

12th Edition

# The Blue Book

OCULAR DISORDERS  
PRESUMED TO BE INHERITED  
IN PUREBRED DOGS

GENETICS COMMITTEE OF  
THE AMERICAN COLLEGE  
OF VETERINARY  
OPHTHALMOLOGISTS

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2019-2020

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## Foreword

Ocular disorders, proven or presumed to be inherited in purebred dogs, have been a topic of intense dialogue by Diplomates of the American College of Veterinary Ophthalmologists (ACVO) for many years. Discussions commenced in the latter half of the 20<sup>th</sup> century during the early days of this College's inception, have continued into the 21<sup>st</sup> century, and will no doubt continue for years to come. Our knowledge of the existence, nature, progression, and inheritance of ocular disorders continues to expand as this field of veterinary science evolves. The Genetics Committee of the ACVO was originally formed in response to requests by registries, breed groups, and veterinarians, with the intent to provide a scientific advisory panel and guidelines regarding ocular disorders in purebred dogs. The Genetics Committee of today remains engaged in an ongoing effort to update information on ocular disorders for this purpose.

The content of this production has originated from several sources as the ACVO recently created a Companion Animal Eye Registry (CAER), which is a joint effort between the Orthopedic Foundation for Animals (OFA) and the ACVO. The addition of eye examination results to the OFA database makes the OFA the most complete source of canine health screening results in the world, allowing responsible breeders to make more informed breeding decisions in an effort to reduce the incidence of inherited disease.

The generation of statistical information is made possible by the efforts of dedicated breeders of purebred dogs who present their dogs to Diplomates of the ACVO for an OFA Companion Animal Eye Registry examination. The research copies of these examinations are then conscientiously submitted to OFA by the examining Veterinary Ophthalmologists. These data generate annual statistics. The statistics for each breed are then reviewed by the Genetics Committee for the most recent year and from the previous 5 years. Recommendations regarding the ocular disorders listed for each breed and the breeding advice are compiled following guidelines detailed elsewhere in this publication. A comprehensive review of the scientific literature since the last published edition was undertaken by all committee members. The scientific articles and breed disorders from the statistical and literature review have been added to the information on each breed in the production of this document. The collective educated clinical experience of the committee members is utilized to reach a consensus of opinion in areas where there remains a paucity of hard scientific proof regarding certain identified breed problems.

The current Genetics Committee has instituted an annual scientific literature search, in addition to the previously established yearly statistical data review. This information is compiled and submitted in an effort to maintain a bank of current information for future editions and versions of this document. The content of all editions past, present, and future will remain dynamic and ever changing as more precise technologies advance the study of the canine genome, as continued scientific research expands our knowledge, and as the database grows.

It is an honor and a privilege to serve the ACVO, our fellow Diplomates, reputable dog breeders, and our most trusted canine companions in this endeavor.

### Genetics Committee 2019

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**12th Edition**  
**2019 - 2020 Version Acknowledgements**

The following groups and individuals deserve credit for the production of this edition of Ocular Disorders Presumed to be Inherited in Purebred Dogs ("The Blue Book"):

The ACVO Board of Regents

Genetics Committee Chairs Dr. Andras Komaromy 2006-2008, Dr. Katie Diehl (2009-2011), Dr. Jacqueline Pearce (2011-2012), Dr. Carrie Breaux (2011-2013), Dr. Kenneth Pierce (2014), Dr. Wendy Townsend (2015), Ellen Belknap (2016), Jessica Meekins (2017), Renee Carter (2018), Adam King (2019), Jane Ashley Huey (2020) and all previous Genetics Committee members

Eddie Dziuk, Chief Operating Officer, and Erika Werne, CAER Program Manager, for the OFA

## Introduction

### What is the purpose of this book?

The Orthopedic Foundation for Animals (OFA), Canine Eye Registration Foundation (CERF), other breed registry groups, breed clubs, and practicing veterinarians have requested that the American College of Veterinary Ophthalmologists (ACVO) provide a scientific advisory panel to furnish guidelines regarding ocular disorders of major concern to purebred dogs. The Genetics Committee of the ACVO was formed in response to these requests and is engaged in an ongoing effort to update information on ocular disorders proven or suspected to be hereditary in purebred dogs. The compendium of ocular disorders and breeding recommendations which follow are interim guidelines. They are reviewed regularly and revised whenever additional information becomes available.

### How can this information be used?

**National and international breed clubs** are encouraged to submit their input regarding breeding decisions for ocular disorders found in their breeds. **Local breed clubs** can participate by encouraging and organizing ocular examination clinics and forwarding their requests and concerns to their national organization. **Practicing veterinarians** are encouraged to contribute by informing all owners of potential breeding animals of the value and availability of ocular examinations, prior to breeding. Information regarding ocular disorders found in litters or individuals can be forwarded to the Genetics Committee via any ACVO diplomate. **Individual breeders** wishing to uphold high ethical standards for the improvement of their breed are urged to contribute by annual examination of their breeding animals and by encouraging the same from other breeders. Further information can be obtained from the Orthopedic Foundation for Animals (OFA): 2300 E Nifong Boulevard, Columbia, MO, 65201-3806, 573-442-0418. Only through increased awareness of the problems and a sustained cooperative effort to disseminate accurate information, will we be able to control and/or eliminate hereditary eye diseases in purebred dogs.

### How do we identify an inherited eye disease?

Although there are noteworthy exceptions, most of the ocular diseases of dogs which are presumed to be hereditary have not been adequately documented. Genetic studies require examination of large numbers of related animals in order to characterize the disorder (age of onset, characteristic appearance, rate of progression) and to define the mode of inheritance (recessive, dominant). In a clinical situation, related animals are frequently not available for examination once a disorder suspected to be inherited is identified in an individual dog. Maintaining a number of dogs for controlled breeding trials through several generations is a long and costly process. Both of these obstacles are compounded by the fact that many ocular conditions do not develop until later in life. Due to the potential for disease to arise from inherited genetic defects at any age, the Genetics Committee recommends annual eye exams.

Until the genetic basis of an ocular disorder is defined in a published report, we rely on what statistical information is available from registry organizations, informed opinions and consensus from ACVO diplomates, and must satisfy ourselves with terms like "presumed inherited" and "suspected to be inherited." Several companies provide information on genetic testing which greatly assists in providing more information and data to aid in defining the canine genetics of ocular diseases.

### **When do we suspect that a disorder is inherited in a given breed?**

- When the frequency is greater than in other breeds
- When the frequency increases in a given breed as a whole
- When the frequency is greater in related dogs within a breed
- When it has a characteristic appearance and location
- When it has a characteristic age of onset and course of progression (predictable stages of development and time for each stage to develop)
- When it looks identical to an entity which has been proven to be inherited in another breed

Special thank you to the “Father of Veterinary Medical Genetics,” Donald F. Patterson, DVM, DSc. Dr. Patterson, who died in 2013, was Emeritus Professor of Medicine and Medical Genetics, University of Pennsylvania School of Veterinary Medicine and Emeritus Professor of Human Genetics, University of Pennsylvania School of Medicine. These guidelines on the heritability of disorders in dogs are based on his lectures and publications.

## **Guidelines Used by the ACVO Genetics Committee in Making Breeding Recommendations**

In this book, we chose the term "**BREEDING ADVICE**" and intentionally avoided the words "certifiable" and "registerable." The ACVO does not serve as a registry organization. Registry organizations operate independently of the ACVO and set their own standards for registration. However, the OFA does follow the guidelines set forth by the ACVO Genetics Committee in this publication. Any registry organization may use the information in this compendium and results of examinations performed by ACVO Diplomates in the registering of animals with regard to breeding suitability as they see fit.

It is important to recognize that the sensitivity of genetic disorder detection is greater when large numbers of dogs are examined. The extensive number of disorders listed in this book for some breeds may reflect the popularity of the breed and the numbers of animals evaluated. Conversely, the lack of disorders listed for other breeds often reflects only the paucity of examinations reported for each breed. For these reasons, the ACVO Genetics Committee strongly recommends annual evaluations of dogs of all breeds as the imperative first step in the control of hereditary ocular disorders. We would like to acknowledge the contribution of the Orthopedic Foundation for Animals (OFA) and Canine Eye Registration Foundation (CERF) for providing statistical summaries of ophthalmic examinations from their files.

**For each breed, specific ocular disorders have been listed which are known or suspected to be inherited based on one or more of the following criteria:**

- 1) There are published reports in the scientific literature regarding a condition in a particular breed with evidence of inheritance.
- 2) The incidence of affected animals (from OFA and CERF reports) is greater than or equal to 1% of the examined population with a minimum of five affected animals per five year period. Regardless of the population of dogs examined, if 50 or more affected individuals are identified in a five year period, the entity will be listed for that breed.
- 3) A specific request from a breed club that a condition be included for their breed may be considered at the ACVO annual meeting of the Genetics Committee if information is received by August 1. Such requests are reviewed critically and must include specific documentation as to the disorder in question and the numbers seen. Further information from the breed club may be requested. The request must receive agreement by a majority of the committee.
- 4) There is overwhelming opinion by a majority of the Genetics Committee members that clinical experience by ACVO Diplomates would indicate a particular condition should be listed for a breed, in spite of the absence of direct evidence of affected animals on OFA or CERF reports.
- 5) Results of genetic laboratory research and genetic testing.

**The "Breeding Advice" given is determined by the significance of the condition to vision and/or very strong evidence of heritability:**

Two categories of advice regarding breeding have been established:

**NO:** Substantial evidence exists to support the heritability of this entity AND/OR the entity represents a potential compromise of vision or other ocular function.

**BREEDER OPTION:** Entity is suspected to be inherited but does not represent potential compromise of vision or other ocular function.

When the breeding advice is "**NO**," even a minor clinical form of the entity would make this animal unsuitable for breeding. When the advice is "**BREEDER OPTION**," caution is advised. In time, it may be appropriate to modify this stand to "**NO**" based on accumulated evidence. If, in time, it becomes apparent that there is insufficient evidence that an entity is inherited, it may be deleted from the list.

**There are currently eleven disorders for which there is an unequivocal recommendation against breeding in all breeds:**

These are conditions which frequently result in blindness and for which there is definite evidence of heritability in one or more breeds. However, these disorders will not be listed on the individual breed page for a given breed, unless they also meet the criteria described above.

- **Keratoconjunctivitis sicca (KCS)** – Breeding is not recommended for any animal demonstrating keratitis consistent with KCS. The prudent approach is to assume KCS to be hereditary except in cases suspected to be non-genetic in origin. See \*note.
- **Glaucoma** – See \*note.
- **Persistent Pupillary Membranes**
  - **Iris to Lens**
  - **Iris to Cornea**
  - **Iris Sheets**
  - **Endothelial Opacity/No Strands**
- **Cataract** – Breeding is not recommended for any animal demonstrating partial or complete opacity of the lens or its capsule *unless the examiner has also checked the box for “suspect not inherited” or unless specified otherwise for the particular breed.* See \*note.
- **Lens luxation or subluxation** – See \*note.
- **Persistent hyperplastic primary vitreous (PHPV)/persistent hyperplastic tunica vasculosa lentis (PHTVL)** – See \*note.
- **Retinal detachment** – See \*note.
- **Retinal atrophy – generalized (PRA)** - Breeding is not advised for any animal demonstrating bilaterally symmetric retinal degeneration (considered to be PRA unless proven otherwise).
- **Retinal dysplasia, geographic or detached forms** – See \*note.
- **Optic nerve coloboma**
- **Optic nerve hypoplasia**

\*Note: The prudent approach of these disorders is to assume they are hereditary except in cases specifically known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, or nutritional deficiencies.

In breeds recognized with Persistent Pupillary Membrane (PPM) as an inherited problem there is an unequivocal recommendation against breeding when there is PPM iris to lens, or PPM iris to cornea, or iris sheets. Breeding advice is “**NO**.”

**The following breeds are recommended to have a preliminary examination prior to initial pharmacological dilation to best facilitate identification of these disorders:**

**Dalmatian** – iris hypoplasia/sphincter dysplasia

**Australian Shepherd** – iris coloboma

**Miniature American Shepherd/Miniature Australian Shepherd** – iris coloboma

**Toy Australian Shepherd** – iris coloboma

### **What can be detected during an Eye Certification Examination?**

A routine eye screening examination includes indirect ophthalmoscopy and slit lamp biomicroscopy following pharmacological dilation of the pupils. Gonioscopy, tonometry, Schirmer tear test, electroretinography, and ultrasonography are not routinely performed; thus, dogs with goniodysgenesis, glaucoma, keratoconjunctivitis sicca, or some early cases of progressive retinal atrophy might not be detected without further testing.

The diagnoses obtained during an ophthalmic eye certification examination refer only to the **phenotype** (clinical appearance) of an animal. Thus, it is possible for a clinically normal animal to be a carrier (abnormal **genotype**) of genetic abnormalities.

An individual ACVO Diplomate may disagree with the breeding advice contained in this compendium. It is appropriate for this examiner to contact the ACVO Genetics Committee to voice disagreement, initiate change, or suggest additions. The members of the Genetics Committee represent the ACVO but acknowledge that the information generated for a breed may not agree with the knowledge and clinical experience of every individual ACVO Diplomate.

### **What is the role of the responsible dog breeder?**

The final beneficiary of the information in this book is the dog breeder. It is up to the conscientious breeder to use this information along with other criteria in selecting which animals to breed. To assist this determination, current certification is recommended. Animals currently free of heritable eye disease will be issued a certificate on receipt of the examination/application by OFA. To avoid confusion between a normal animal (no evidence of heritable eye disorders) and one that may have a minor fault coming under the advice of Breeder Option, the Breeder Option category will be printed on the certificate. This is intended to stimulate conversation as to the specific nature of the Breeder Option condition found in that particular animal, allowing breeders using a dog in a breeding program to make an informed decision.

**There are many ocular conditions which are a direct result of selection for a facial conformation considered desirable by breeders.**

These include:

- Entropion
- Ectropion
- Macropblepharon
- Exposure keratopathy syndrome

Facial conformation with excessively prominent eyes, heavy facial folds, or eyelids which are either inverted or everted predispose animals to corneal irritation, discomfort, and if left untreated, can lead to loss of vision. A responsible breeding program should recognize and select away from these exaggerated facial features.



## THE ROLE OF GENETIC TESTING IN THE DETECTION OF OCULAR DISEASE

Genetic testing plays a very important role in the diagnosis of disease. However, it is important to be aware of the limitations of genetic testing and understand its role in the detection and control of genetically inherited diseases.

Genetically inherited diseases are caused by a deleterious sequence change (mutation) in the DNA that results in an abnormal protein (protein can be absent, have insufficient function, or have an abnormal function) that results in disease.

Genetic tests are developed by comparing the DNA sequence of a normal animal to that of an animal with disease. This allows the identification of a particular DNA sequence that can be causally associated with the disease. This is an extremely powerful tool that, in some cases, allows for identification of disease even before it is evident clinically.

However, a particular test is only capable of detecting the DNA sequence it was designed to detect. That is, the DNA test only tests for a specific change in the DNA that can cause disease. For example, a DNA test specific for the *PDE6B* gene mutation (responsible for the rcd1 form of PRA in the Irish Setter) will not detect any abnormalities in other breeds or mixed breeds that have other mutations in the same gene. Thus the specificity of a DNA test is also its limitation, and in the case of PRA in Irish Setters it is specific for the Irish Setter defect and not for any other defects.

In polygenic disorders, a genetic test cannot evaluate the integrity of all the proteins that make up a particular cellular process. Thus, it is possible for a DNA test that has been associated with a disease to be normal and yet the disease can still be present. The disease could be caused by an abnormality in one of the other genes that are involved with that particular cellular process. The defect in the other protein still results in an abnormal cellular process, which still results in disease. A perfect example of this is observed in oculo-skeletal dysplasia in Labrador Retrievers and Samoyed dogs. In both breeds the diseases are clinically identical, yet caused by mutations in different genes involved in fibril formation of a specific kind of collagen molecule.

Thus, obtaining a DNA test that is normal does not guarantee absence of disease. It only guarantees that the particular change the DNA test was designed to detect is not present, and that disease from that particular change will not occur. This is why genetic testing should be combined with ophthalmic examination for maximum efficacy. An ophthalmic exam evaluates the sum total or “result” of all the cellular processes required to maintain ocular health and result in vision, and is an essential part of the ocular wellness exam to ensure that other important clinically recognizable diseases are not present.

## Breeder Option Codes

### A – Eyelids

- A1 Entropion
- A2 Ectropion
- A3 Distichiasis
- A4 Ectopic Cilia
- A6 Imperforate Lacrimal Punctum

### B – Nictans

- B1 Cartilage Anomaly/Eversion
- B2 Gland Prolapse

### C – Cornea

- C1 Corneal Dystrophy – Epithelial/Stromal
- C2 Corneal Dystrophy – Endothelial
- C4 Pigmentary Keratitis/Keratopathy

### D – Uvea

- D1a Uveal Cyst – Free Floating
- D1b Uveal Cyst – Single
- D1c Uveal Cyst – Multiple
- D2 Iris Coloboma
- D3 Persistent Pupillary Membranes – Iris to Iris
- D4 Iris Hypoplasia

### E – Lens

- E1 Cataract – Suspect Not Inherited
- E2 Posterior Y Tip Suture Opacities

### F – Vitreous

- F1 Persistent Hyaloid Artery
- F2a Vitreous Degeneration – Syneresis
- F2b Vitreous Degeneration – Anterior Chamber

### G – Fundus

- G1 Retinal Dysplasia – Folds
- G5 Micropapilla
- G6 Retinopathy

## Breeds Not Listed for Insufficient Data

Attempts have been made to confirm information on the following list of breeds/rare breeds. This list is not an endorsement of the breed status and may change from time to time as additional information is available.

To date there are no published reports of inherited ocular conditions in these breeds and/or the numbers of individuals for which examinations are recorded are too low to identify the presence of significant ocular disorders. Examinations are encouraged to accumulate information and reduce the likelihood of undetected conditions becoming problematic.

|                               |                              |
|-------------------------------|------------------------------|
| Aatu Tamaskan                 | French Spaniel               |
| Alano                         | German Longhaired Pointer    |
| Alapaha Blue-Blood Bulldog    | German Spaniel               |
| Akbash                        | Grand Basset Griffon Vendeen |
| American Alsatian             | Greenland Dog                |
| American Bandogge Mastiff     | Hanoverian Hound             |
| American Bully                | Harrier                      |
| American English Coonhound    | Hovawart                     |
| American Foxhound             | Jindo                        |
| American Husky                | Kishu Ken                    |
| American Leopard Hound        | Korean Poongsan              |
| Anatolian Shepherd            | Kromforhlander               |
| Armenian Gampr                | Large Munsterlander          |
| Australian Koolie             | Llewellyn Setter             |
| Azawakh                       | Magyar Agar                  |
| Bavarian Mountain Scent Hound | McNab                        |
| Bergamasco                    | Munsterlander                |
| Berger des Pyrenees           | Native American Indian Dog   |
| Biewer Terrier                | Native American Village Dog  |
| Blue Lacy                     | Nederlandse Kooikerhondje    |
| Blue Mountain Shepherd        | New Zealand Huntaway         |
| Bluetick Coonhound            | North American Shepherd      |
| Boz Shepherd                  | Norwegian Lundehund          |
| Braque d'Auvergne             | Old English Bulldogge        |
| Braque du Bourbonnais         | Otterhound                   |
| Braque Francais Pyrenees      | Picardy Spaniel              |
| Bull Terrier                  | Polish Tatra Sheepdog        |
| Ca De Bou                     | Porcelaine Hound             |
| Cao De Castro Laboreiro       | Portuguese Podengo           |
| Carolina Dog                  | Portuguese Pointer           |
| Catalan Sheepdog              | Pudelpointer                 |
| Caucasian Shepherd            | Pumi                         |
| Central Asian Shepherd        | Pyrenean Mastiff             |
| Cesky Terrier                 | Redbone Coonhound            |
| Chart Polski                  | Saluki                       |
| Chinook Hybrid                | Scottish Deerhound           |
| Cirneco Dell'Etna             | Seppala Siberian Sled Dog    |
| Czechoslovakian Vlcak         | Shorty Bull                  |
| Danish Broholmer              | Skye Terrier                 |
| Danish Swedish Farmhound      | Slovakian Wirehaired Pointer |
| Dogo d'Argentino              | Small Munsterlander          |
| Drentsch Partrijshond         | Spanish Greyhound            |
| Drever                        | Spanish Mastiff              |
| ECT Landseer                  | Stabyhoun                    |
| English Coonhound             | Treeing Walker               |
| English Foxhound              | Wachtelhund                  |
| English Jack Russell Terrier  | Welsh Sheepdog               |
| Epagneul Breton               | White Swiss Shepherd         |
| Estrela Mountain Dog          | Windsprite                   |
| Fila Brasileiro               | Working Kelpie               |
| Finnish Spitz                 | Yakutian Laika               |
| French Pointer                |                              |

## Glossary of Terms

*(For more detailed definitions, the reader is referred to medical and genetic scientific texts.)*

**Achromatopsia:** see **Day blindness**

**Canine multifocal retinopathy:** characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). The condition includes numerous distinct (i.e. multi-focal), roughly circular patches of elevated retina with accumulation of material that produces gray-tan-pink colored lesions (multifocal bullous retinal detachments). These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs and might not progress or progress slowly, or may appear to heal with discrete areas of tapetal hyper-reflectivity or hyperpigmentation. Most dogs exhibit no noticeable problem with vision despite their abnormal appearing retinas.

**Cataract:** any opacity of the lens and/or its capsule, regardless of size or location within the lens. Cataracts are assumed to be hereditary unless associated with known trauma, ocular inflammation, specific metabolic diseases, or nutritional deficiencies.

**Ceroid lipofuscinosis:** an inherited disease of man and animals characterized by the accumulation of lipopigment in various tissues of the body including the eye. It results in progressive neurologic disease including blindness. (Also called Batten's disease.)

**Choroidal hypoplasia:** a congenital, inherited, non-progressive defect primarily affecting the choroid resulting in some or all of the following: decreased or lack of pigment in the retinal pigment epithelium or choroid, tapetal thinning, and reduced or abnormal choroidal blood vessels.

**Chronic superficial keratitis (CSK):** see **Pannus**

**Collie eye anomaly:** a congenital syndrome of ocular anomalies characterized by bilateral and often symmetrical defects including any combination of **choroidal hypoplasia**, **coloboma**, and **retinal detachment(s)**.

**Coloboma:** a congenital abnormality in ocular development usually characterized by focal absence of tissue, commonly (though not exclusively) located at the 6 o'clock position associated with failure of closure of the optic fissure.

**Cone degeneration:** the loss of photopic vision caused by selective degeneration of the cone photoreceptors. Also known as day blindness, hemeralopia, or achromatopsia.

**Corneal degeneration:** opacification of one or more of the corneal layers frequently resulting from deposition of lipid or mineral and occurring secondary to chronic inflammation.

**Corneal dystrophy:** non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers (**epithelium**, **stroma**, **endothelium**). The term dystrophy implies an inherited condition. It is usually bilateral although not necessarily symmetrical and the onset in one eye may precede the other.

**Corneal dystrophy - endothelial:** breed-related loss or dysfunction of corneal endothelial cells resulting in bilateral, progressive corneal edema.

**Corneal dystrophy - epithelial, stromal:** breed-related, non-inflammatory, white to silver-colored opacification of the corneal epithelium and/or stroma frequently resulting from deposition of lipid.

**Day blindness:** see **Cone degeneration**

**Dermoid:** a congenital, non-cancerous growth occurring on the cornea, conjunctiva, or eyelid typified by the presence of skin-like structures.

**Distichiasis:** the presence of abnormally oriented eyelashes, frequently protruding from Meibomian gland ductal openings.

**Dry eye:** see **Keratoconjunctivitis sicca**

**Dysplasia:** abnormality of development.

**Dystrophy:** non-inflammatory, developmental, nutritional, or metabolic abnormality; dystrophy implies a possible hereditary basis and is usually bilateral.

**Ectopic cilia:** aberrant hairs emerging through the palpebral conjunctiva which often causes ocular discomfort and corneal disease.

**Ectropion:** a conformational defect resulting in eversion of the eyelid margin, which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several factors defining the skin and other structures, which make up the eyelids, orbital contents, and conformation of the skull.

**Entropion:** a conformational defect resulting in inversion of the eyelid margin which may cause ocular irritation. It is likely that entropion is influenced by several factors defining the skin and other structures, which make up the eyelids, orbital contents, and conformation of the skull.

**Euryblepharon:** an exceptionally long eyelid marginal length, which may lead to Ectropion or Entropion. Euryblepharon is synonymous with the term macropalpebral fissure.

**Exposure/pigmentary keratitis:** a condition characterized by variable degrees of superficial vascularization, fibrosis, and/or pigmentation of the cornea. May be associated with excessive exposure/irritation of the globe due to shallow orbits, lower eyelid medial entropion, lagophthalmos, and macropalpebral fissure.

**Glaucoma:** characterized by an elevation of intraocular pressure (IOP) which causes optic nerve and retinal degeneration and results in blindness. Diagnosis and classification of glaucoma requires tonometry and gonioscopy, which are not part of a routine eye certification examination.

**Glaucoma, pigmentary:** see **Ocular melanosis**

**Goniodysgenesis:** congenital anomaly characterized by the persistence of a variably fenestrated sheet of uveal tissue spanning the iridocorneal angle, extending from the iris base to the peripheral cornea. Diagnosis is by gonioscopy, which is not part of a routine eye certification examination.

**Hemeralopia:** see **Cone degeneration**

**Imperforate lacrimal punctum:** developmental anomaly resulting in an imperforate opening of the lacrimal puncta. An imperforate lower punctum may result in epiphora, an overflow of tears onto the face.

**Iridocorneal angle:** the junction between the iris and the cornea; the drainage angle. Aqueous humor leaves the anterior chamber via the trabecular meshwork within the iridocorneal angle into the venous circulation.

**Iris coloboma:** a congenital abnormality in iris development usually characterized by a full-thickness defect in iris tissue, commonly (though not exclusively) located at the 6 o'clock position associated with failure of closure of the optic fissure. A partial-thickness defect in iris tissue should be recorded as iris hypoplasia on the eye certification form.

**Iris cyst:** see **Uveal cyst**

**Iris hypoplasia:** a congenital abnormality in iris development usually characterized by a reduced quantity of tissue identified as a partial-thickness defect in iris tissue. Full-thickness iris hypoplasia is rare and should be recorded as an iris coloboma on the eye certification form.

**Iris melanoma:** see **Uveal melanoma**

**Iris sphincter dysplasia:** a congenital abnormality in iris development usually characterized by a full-thickness defect in iris tissue at the level of the iris sphincter, causing pupillary dilation. This abnormality has been noted in the Dalmatian breed.

**Keratitis:** inflammation of the cornea.

**Keratitis, punctate:** inflammation of the cornea accompanied by multifocal, coalescing areas of stromal corneal ulceration of variable depth.

**Keratoconjunctivitis sicca (KCS):** an abnormality of the tear film attributed to deficiency of the aqueous portion of the tears. Progressive KCS may result in ocular surface irritation and/or vision impairment via corneal opacification. Also called dry eye. The test for this condition is the Schirmer Tear Test, which is not part of a routine eye certification examination.

**Lens subluxation/luxation:** partial (subluxation) or complete displacement of the lens from the normal anatomic site. Lens luxation may result in elevated intraocular pressure (secondary glaucoma), causing vision impairment, pain, and/or retinal detachment.

**Lenticonus:** an anomaly of the lens in which the anterior or posterior surface protrudes in a conical form; usually congenital.

**Macroblepharon:** an exceptionally large palpebral fissure. Macroblepharon in conjunction with laxity of the lateral canthal structures may lead to lower lid ectropion and upper lid entropion. Either of these conditions may lead to severe ocular irritation.

**Merle:** an incompletely dominant phenotype in which heterozygous (M/m) dogs exhibit a coat color phenotype of various dilute color patches, while homozygous (M/M) dogs exhibit marked hypopigmentation and ocular defects, including microphthalmia, blindness and colobomas, and deafness. Deafness and ocular defects are sometimes seen in heterozygous individuals.

**Micropapilla:** a congenital anomaly which results in a small optic disk diameter without vision loss. Contrast with optic nerve hypoplasia, which may have a similar ophthalmoscopic appearance with vision loss.

**Microphakia:** a congenital anomaly in which there is an abnormally small lens.

**Microphthalmos:** a congenital anomaly in which the globe is abnormally small. Commonly associated with multiple ocular malformations and when severe, may affect vision.

**Nictitans cartilage anomaly/eversion:** a congenital anomaly in the nictitating membrane in which the T-shaped cartilage is malformed and/or folded.

**Nictitans gland prolapse:** protrusion of the tear-producing gland of the nictitating membrane from its normal position posterior to the nictitating membrane, to a position superior to the free margin of this structure.

**Nodular granulomatous episclerokeratitis (NGE):** an inflammatory disorder of the sclera and episclera, with occasional corneal involvement, characterized by granulomatous infiltrates. Previously known as **Proliferative keratoconjunctivitis**. This condition is most commonly seen in the Collie.

**Nyctalopia:** loss of scotopic (night) vision. Causes include genetic defects in photoreceptors and in retinal pigment epithelium, either dystrophy or degeneration of affected cells.

**Ocular melanosis:** progressive bilateral and sometimes asymmetrical increase in pigmentation with melanocytic accumulation the uveal tract and adjacent tissues. Ultimately progresses to glaucoma and loss of vision in most cases (melanocytic glaucoma). Not associated with systemic disease or metastases. Most often recognized in Cairn Terriers.

**Optic nerve coloboma:** a congenital abnormality of the optic nerve commonly associated with failure of closure of the optic fissure, resulting in a defect in the optic nerve in the anterior-posterior plane. May result in partial or total vision loss.

**Optic nerve hypoplasia:** a congenital anomaly, which results in a small optic disk diameter and vision loss. Contrast with micropapilla, which may have a similar ophthalmoscopic appearance but without loss of vision.

**Pannus:** a bilateral inflammatory disease of the cornea which usually starts as a grayish haze to the inferior or inferiortemporal cornea, followed by the formation of a vascularized subepithelial opacity that begins to spread toward the central cornea; pigmentation may follow the vascularization. If severe, vision impairment occurs. Plasma cell infiltration of the nictitans may occur in conjunction with CSK, or on its own. (Also called "CSK".)

**Persistent hyaloid artery (PHA):** congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small patent vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

**Persistent hyperplastic primary vitreous (PHPV):** congenital defect resulting from abnormalities in the regression of the hyaloid artery (the primary vitreous) and the interaction of the blood vessel with the posterior lens capsule/cortex during embryogenesis. This condition is often associated with congenital cataracts and frequently seen with PHTVL.

**Persistent hyperplastic tunica vasculosa lentis (PHTVL):** congenital defect resulting from failure of regression of the embryonic vascular network which surrounds the developing lens. Often associated with PHPV and a patent hyaloid artery.

**Persistent pupillary membranes (PPM):** persistent blood vessel remnants in the anterior chamber which fail to regress normally by 3 months of age. These strands arise from the iris collaret and may bridge from iris to iris, iris to lens, iris to cornea, or form sheets of tissue in the anterior chamber.

**Persistent tunica vasculosa lentis (PTVL):** clinically insignificant posterior epicapsular lenticular opacities resulting from incomplete regression of the embryonic vascular network which surrounds the developing lens.

**Pigmentary glaucoma:** see **Ocular melanosis**

**Pigmentary uveitis:** see **Uveitis, pigmentary**

**Pigmentary keratopathy:** a condition reported in Pugs in which the cornea becomes pigmented, often resulting in vision impairment. Development of pigmentary keratopathy is associated with congenital uveal pathology – iris hypoplasia and the presence of persistent pupillary membranes – but not with other factors such as Schirmer tear test values or medial canthal entropion.

**Plasmoma:** see **Pannus**. Also called Atypical Pannus. Bilateral thickening and depigmentation of the nictitans due to invasion of lymphocytes and plasma cells. It may or may not be associated with corneal involvement (Pannus).

**Progressive rod-cone degeneration (PRCD)** (see also **PRA**): Typically refers to recessively inherited generalized loss of rod photoreceptors followed by cone degeneration. Many different genetic mutations result in a similar phenotypic presentation.

**Progressive retinal atrophy (PRA):** an umbrella term used to describe a group of inherited dysplastic, dystrophic, or degenerative diseases of the retinal visual cells (photoreceptors, retinal pigment epithelium, or both).

**Proliferative keratoconjunctivitis:** see **Nodular granulomatous episclerokeratitis**

**Retinal atrophy:** a non-specific term used to describe a decrease in the number and deterioration of the cells of the retina, regardless of cause.

**Retinal degeneration:** see **Retinal atrophy**

**Retinal detachment:** a separation of the neurosensory retina from the retinal pigment epithelium.

**Retinal dysplasia:** abnormal development of the retina present at birth. This condition is non-progressive and recognized in 3 forms: **folds**, **geographic**, **detached**.

**Retinal dysplasia – folds:** seen ophthalmoscopically as linear, triangular, curved or curvilinear foci of retinal folding. May be single or multiple. In puppies, retinal folds can be seen as a transient phenomenon, resolving as the eye attains maturity.

**Retinal dysplasia – geographic:** an irregularly shaped area of retinal development containing both areas of thinning and areas of elevation. This form may be associated with visual impairment.

**Retinal dysplasia – detached:** severe retinal disorganization associated with separation of the neurosensory retina from the retinal pigmented epithelium. This form results in visual impairment.



**Retinopathy:** any non-inflammatory condition of the retina. These conditions can usually be detected by ophthalmoscopic examination, but an electroretinogram (ERG) may be required in some instances (e.g. canine multifocal retinopathy).

**Rod-cone dysplasia:** an inherited retinal disease characterized by abortive or abnormal development of rods and cones. Affected animals become blind early in life, usually within the first 6 months, with the exception of *rca4* in the Gordon and Irish Setter dogs. See specific breed pages for rod-cone dysplasia type descriptions.

**Rod dysplasia:** abnormal development of the visual cells resulting in vision impairment in dim light by 6 months and total blindness at 3-5 years.

**Uveal cyst:** a pigmented, fluid-filled epithelial-lined structure arising from the posterior iris or ciliary body epithelium. Cysts may remain attached to the pupil margin, iris, or ciliary body, or may detach and be free-floating within the anterior chamber. They may rupture and adhere to the cornea or anterior lens capsule. Uveal cysts may occur in any breed. Uveal cysts are commonly benign, although they may be associated with other pathologic conditions in various breeds.

**Uveal cyst, anterior chamber:** a pigmented, fluid-filled, epithelial-lined structure arising from the posterior iris or ciliary body epithelium which has detached from its site of origin and is free-floating in the anterior chamber.

**Uveal cyst, ciliary body:** a pigmented, fluid-filled, epithelial-lined structure arising from the ciliary body epithelium and attached to the ciliary body.

**Uveal cyst, iris:** a pigmented, fluid-filled, epithelial-lined structure arising from the posterior iris epithelium and attached to the iris.

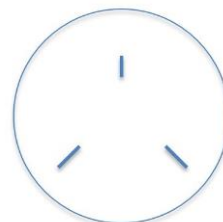
**Uveal melanoma:** a locally invasive melanocytic neoplasm arising within the uveal tract, may be benign (melanocytoma) or malignant (malignant melanoma). Uveal melanomas are reported in higher frequency in German Shepherd Dogs and Labrador Retrievers. Inherited iris melanoma has been reported in Labrador Retrievers.

**Uveitis, pigmentary:** a specific form of uveitis most commonly seen in middle-aged to older Golden Retrievers. Clinically manifests early as pigment deposition in a radial fashion on the anterior lens capsule with iridociliary cysts. Later stages are associated with posterior synechia, fibrinous anterior uveitis, cataract, and ultimately glaucoma. Not associated with systemic disease; may be asymmetric in presentation.

**Uveodermatologic syndrome:** an immune-mediated syndrome of anterior uveitis, chorioretinitis, dermal depigmentation (vitiligo), and hair depigmentation (poliosis). A similar syndrome in humans, called Vogt-Koyanagi-Harada syndrome (VKH), is an autoimmune disease directed against melanocytes. Secondary glaucoma and/or retinal detachment are frequent complications of this disease. Seen most commonly in the Akita, Samoyed, and Siberian Husky breeds.

**Vitreous degeneration:** Liquefaction of the vitreous gel which may predispose to retinal detachment resulting in blindness.

**Y-suture tip opacity:** These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



# AFFENPINSCHER

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Affenpinscher breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT AFFENPINSCHER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |       |
|---|---------------------|-----------|------|-----------|-------|
|   |                     | #         | %    | #         | %     |
| <b>GLOBE</b>  |                     |           |      |           |       |
| 0.110 microphthalmia  |                     | 1         | 0.3% | 0         |       |
| <b>EYELIDS</b>  |                     |           |      |           |       |
| 20.140 ectopic cilia  |                     | 1         | 0.3% | 2         | 1.4%  |
| 25.110 distichiasis   |                     | 21        | 5.8% | 3         | 2.1%  |
| <b>NASOLACRIMAL</b>   |                     |           |      |           |       |
| 40.910 keratoconjunctivitis sicca                                   |                     | 0         |      | 1         | 0.7%  |
| <b>NICTITANS</b>  |                     |           |      |           |       |
| 52.110 prolapsed gland of the third eyelid                          |                     | 1         | 0.3% | 0         |       |
| <b>CORNEA</b>   |                     |           |      |           |       |
| 70.220 pigmentary keratitis   |                     | 0         |      | 2         | 1.4%  |
| 70.700 corneal dystrophy  |                     | 6         | 1.7% | 5         | 3.4%  |
| <b>UVEA</b>   |                     |           |      |           |       |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 26        | 7.2% | 18        | 12.3% |
| 93.730 persistent pupillary membranes, iris to cornea               |                     | 1         | 0.3% | 0         |       |
| 93.740 persistent pupillary membranes, iris sheets                  |                     | 1         | 0.3% | 0         |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 4         | 1.1% | 2         | 1.4%  |
| <b>LENS</b>   |                     |           |      |           |       |
| 100.200 cataract, unspecified                                       |                     | 3         | 0.8% | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 8         | 2.2% | 1         | 0.7%  |
| 100.302 punctate cataract, posterior cortex                         |                     | 1         | 0.3% | 0         |       |
| 100.311 incipient cataract, anterior cortex                         |                     | 1         | 0.3% | 0         |       |
| 100.312 incipient cataract, posterior cortex                        |                     | 3         | 0.8% | 1         | 0.7%  |
| 100.316 incipient cataract, nucleus                                 |                     | 0         |      | 1         | 0.7%  |
| 100.328 y-suture tip opacities                                      |                     | 0         |      | 1         | 0.7%  |
| 100.330 generalized/complete cataract                               |                     | 3         | 0.8% | 0         |       |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 11        | 3.0% | 3         | 2.1%  |
| <b>VITREOUS</b>   |                     |           |      |           |       |
| 110.200 vitreous degeneration-anterior chamber                      |                     | 0         |      | 1         | 0.7%  |
| 110.320 vitreal degeneration  |                     | 4         | 1.1% | 0         |       |
| <b>RETINA</b>   |                     |           |      |           |       |
| 120.170 retinal dysplasia, folds                                    |                     | 2         | 0.6% | 0         |       |
| <b>OPTIC NERVE</b>  |                     |           |      |           |       |
| 130.120 optic nerve hypoplasia                                      |                     | 0         |      | 1         | 0.7%  |
| <b>OTHER</b>  |                     |           |      |           |       |
| 900.000 other, unspecified  |                     | 3         | 0.8% | 0         |       |
| 900.100 other, not inherited  |                     | 8         | 2.2% | 1         | 0.7%  |
| 900.110 other. suspect not inherited/significance unknown           |                     | 1         | 0.3% | 3         | 2.1%  |

|                                     | 1991-2015 | 2016-2020 |
|-------------------------------------|-----------|-----------|
| <b>NORMAL</b><br>0.000 normal globe | 306 84.5% | 109 74.7% |

# AFGHAN HOUND

|    | DISORDER  | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|---|-------------|-----------|-----------------|
| A. | Distichiasis  | Not defined | 1         | Breeder option  |
| B. | Corneal dystrophy<br>- epithelial/stromal           | Not defined | 1, 2      | Breeder option  |
| C. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| D. | Cataract  | Not defined | 1, 3-6    | NO              |
| E. | Y-suture tip opacity                                | Not defined | 1         | Breeder Option  |

## Description and Comments

A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

B. Corneal Dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

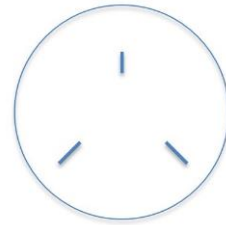
D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

The characteristic cataract in the Afghan Hound begins as equatorial lens vacuoles in dogs from 4 months to 2 years of age. The opacities then extend into the anterior and posterior cortices. Rapid progression can occur with visual impairment in young adults. Test breedings have been done which support a hereditary basis; however, the exact mode of inheritance is unknown.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Vainisi SJ, Goldberg MF. *Animal models of inherited disease. In: Genetic and Metabolic Eye Disease* Little Brown and Company, Boston, 1974.
3. Roberts SR, Helper LC. Cataracts in Afghan hounds. *J Am Vet Med Assoc.* 1972; 160: 427.
4. Roberts SR. Hereditary cataracts. *Vet Clin North Am.* 1973; 3: 433.
5. Barnett KC. The diagnosis and differential diagnosis of cataract in the dog. *J Small Anim Pract.* 1985; 26: 305.
6. Barnett KC. Hereditary cataract in the dog. *J Small Anim Pract.* 1978; 19: 109-120.

# OCULAR DISORDERS REPORT AFGHAN HOUND

| Diagnostic Name   | TOTAL DOGS EXAMINED |       | 1991-2015<br>2,160 |       | 2016-2020<br>400 |   |
|---|---------------------|-------|--------------------|-------|------------------|---|
|   | #                   | %     | #                  | %     | #                | % |
| <b>GLOBE</b>  |                     |       |                    |       |                  |   |
| 0.110 microphthalmia  | 0                   |       | 1                  | 0.2%  |                  |   |
| 10.000 glaucoma   | 2                   | 0.1%  | 0                  |       |                  |   |
| <b>EYELIDS</b>  |                     |       |                    |       |                  |   |
| 21.000 entropion, unspecified   | 2                   | 0.1%  | 0                  |       |                  |   |
| 25.110 distichiasis   | 26                  | 1.2%  | 3                  | 0.8%  |                  |   |
| <b>NASOLACRIMAL</b>   |                     |       |                    |       |                  |   |
| 32.110 imperforate lower nasolacrimal punctum                         | 1                   | 0.0%  | 0                  |       |                  |   |
| 40.910 keratoconjunctivitis sicca                                     | 1                   | 0.0%  | 0                  |       |                  |   |
| <b>NICTITANS</b>  |                     |       |                    |       |                  |   |
| 51.100 third eyelid cartilage anomaly                                 | 1                   | 0.0%  | 0                  |       |                  |   |
| <b>CORNEA</b>   |                     |       |                    |       |                  |   |
| 70.210 corneal pannus   | 3                   | 0.1%  | 0                  |       |                  |   |
| 70.700 corneal dystrophy  | 232                 | 10.7% | 49                 | 12.2% |                  |   |
| 70.730 corneal endothelial degeneration                               | 3                   | 0.1%  | 0                  |       |                  |   |
| <b>UVEA</b>   |                     |       |                    |       |                  |   |
| 93.710 persistent pupillary membranes, iris to iris                   | 57                  | 2.6%  | 14                 | 3.5%  |                  |   |
| 93.720 persistent pupillary membranes, iris to lens                   | 1                   | 0.0%  | 0                  |       |                  |   |
| 93.730 persistent pupillary membranes, iris to cornea                 | 1                   | 0.0%  | 0                  |       |                  |   |
| 93.740 persistent pupillary membranes, iris sheets                    | 2                   | 0.1%  | 0                  |       |                  |   |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   | 0                   |       | 1                  | 0.2%  |                  |   |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands | 1                   | 0.0%  | 0                  |       |                  |   |
| 93.810 uveal melanoma   | 0                   |       | 1                  | 0.2%  |                  |   |
| 93.999 uveal cysts  | 4                   | 0.2%  | 0                  |       |                  |   |
| <b>LENS</b>   |                     |       |                    |       |                  |   |
| 100.200 cataract, unspecified   | 9                   | 0.4%  | 0                  |       |                  |   |
| 100.210 cataract. suspect not inherited/significance unknown          | 126                 | 5.8%  | 32                 | 8.0%  |                  |   |
| 100.301 punctate cataract, anterior cortex                            | 1                   | 0.0%  | 2                  | 0.5%  |                  |   |
| 100.302 punctate cataract, posterior cortex                           | 2                   | 0.1%  | 0                  |       |                  |   |
| 100.303 punctate cataract, equatorial cortex                          | 1                   | 0.0%  | 1                  | 0.2%  |                  |   |
| 100.305 punctate cataract, posterior sutures                          | 7                   | 0.3%  | 4                  | 1.0%  |                  |   |
| 100.306 punctate cataract, nucleus                                    | 1                   | 0.0%  | 0                  |       |                  |   |
| 100.307 punctate cataract, capsular                                   | 3                   | 0.1%  | 0                  |       |                  |   |
| 100.311 incipient cataract, anterior cortex                           | 4                   | 0.2%  | 3                  | 0.8%  |                  |   |
| 100.312 incipient cataract, posterior cortex                          | 2                   | 0.1%  | 2                  | 0.5%  |                  |   |
| 100.313 incipient cataract, equatorial cortex                         | 2                   | 0.1%  | 0                  |       |                  |   |
| 100.314 incipient cataract, anterior sutures                          | 2                   | 0.1%  | 1                  | 0.2%  |                  |   |
| 100.315 incipient cataract, posterior sutures                         | 10                  | 0.5%  | 2                  | 0.5%  |                  |   |
| 100.316 incipient cataract, nucleus                                   | 3                   | 0.1%  | 1                  | 0.2%  |                  |   |
| 100.317 incipient cataract, capsular                                  | 2                   | 0.1%  | 1                  | 0.2%  |                  |   |
| 100.321 incomplete cataract, anterior cortex                          | 1                   | 0.0%  | 2                  | 0.5%  |                  |   |
| 100.322 incomplete cataract, posterior cortex                         | 1                   | 0.0%  | 2                  | 0.5%  |                  |   |
| 100.323 incomplete cataract, equatorial cortex                        | 0                   |       | 2                  | 0.5%  |                  |   |
| 100.324 incomplete cataract, anterior sutures                         | 1                   | 0.0%  | 0                  |       |                  |   |



| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.325 incomplete cataract, posterior sutures            | 1 0.0%           | 0                |
| 100.326 incomplete cataract, nucleus                      | 1 0.0%           | 3 0.8%           |
| 100.328 y-suture tip opacities                            | 3 0.1%           | 13 3.2%          |
| 100.330 generalized/complete cataract                     | 2 0.1%           | 0                |
| 100.345 <i>significant cataracts (summary)</i>            | 59 2.7%          | 39 9.8%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 1 0.0%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 1 0.0%           | 1 0.2%           |
| 110.135 PHPV/PTVL   | 1 0.0%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 1 0.0%           | 3 0.8%           |
| 110.320 vitreal degeneration                              | 8 0.4%           | 3 0.8%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.120 coloboma   | 2 0.1%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 5 0.2%           | 1 0.2%           |
| 120.180 retinal dysplasia, geographic                     | 2 0.1%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 9 0.4%           | 0                |
| 120.960 retinopathy                                       | 2 0.1%           | 1 0.2%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 0                | 1 0.2%           |
| 130.150 optic disc coloboma                               | 3 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 20 0.9%          | 0                |
| 900.100 other, not inherited                              | 34 1.6%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 23 1.1%          | 12 3.0%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,720 79.6%      | 269 67.2%        |

# AIREDALE TERRIER

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--------------------------------|-------------|-----------|-----------------|
| A. | Distichiasis                   | Not defined | 1         | Breeder option  |
| B. | Persistent pupillary membranes |             |           |                 |
|    | - iris to iris                 | Not defined | 1         | Breeder option  |
|    | - iris to cornea               | Not defined | 1         | NO              |
| C. | Cataract                       | Not defined | 1         | NO              |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and the more severe forms of retinal dysplasia is undetermined.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Airedale Terrier breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT AIREDALE TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 0.110 microphthalmia  |                     | 3         | 0.4% | 0         |      |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 20.140 ectopic cilia  |                     | 2         | 0.3% | 0         |      |
| 21.000 entropion, unspecified   |                     | 4         | 0.5% | 0         |      |
| 25.110 distichiasis   |                     | 54        | 7.0% | 12        | 7.8% |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.210 corneal pannus   |                     | 1         | 0.1% | 0         |      |
| 70.700 corneal dystrophy  |                     | 9         | 1.2% | 0         |      |
| 70.730 corneal endothelial degeneration                               |                     | 3         | 0.4% | 0         |      |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 25        | 3.2% | 7         | 4.6% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 7         | 0.9% | 1         | 0.7% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 21        | 2.7% | 1         | 0.7% |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 2         | 0.3% | 0         |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 4         | 0.5% | 5         | 3.3% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 3         | 0.4% | 7         | 4.6% |
| 93.999 uveal cysts  |                     | 1         | 0.1% | 0         |      |
| 97.150 chorioretinal coloboma, congenital                             |                     | 0         |      | 1         | 0.7% |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.200 cataract, unspecified   |                     | 7         | 0.9% | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 50        | 6.5% | 9         | 5.9% |
| 100.301 punctate cataract, anterior cortex                            |                     | 7         | 0.9% | 2         | 1.3% |
| 100.302 punctate cataract, posterior cortex                           |                     | 6         | 0.8% | 0         |      |
| 100.303 punctate cataract, equatorial cortex                          |                     | 2         | 0.3% | 0         |      |
| 100.304 punctate cataract, anterior sutures                           |                     | 1         | 0.1% | 0         |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 4         | 0.5% | 1         | 0.7% |
| 100.306 punctate cataract, nucleus                                    |                     | 1         | 0.1% | 0         |      |
| 100.307 punctate cataract, capsular                                   |                     | 1         | 0.1% | 0         |      |
| 100.311 incipient cataract, anterior cortex                           |                     | 9         | 1.2% | 0         |      |
| 100.312 incipient cataract, posterior cortex                          |                     | 9         | 1.2% | 0         |      |
| 100.313 incipient cataract, equatorial cortex                         |                     | 6         | 0.8% | 1         | 0.7% |
| 100.315 incipient cataract, posterior sutures                         |                     | 4         | 0.5% | 0         |      |
| 100.316 incipient cataract, nucleus                                   |                     | 2         | 0.3% | 0         |      |
| 100.317 incipient cataract, capsular                                  |                     | 3         | 0.4% | 0         |      |
| 100.321 incomplete cataract, anterior cortex                          |                     | 0         |      | 1         | 0.7% |
| 100.322 incomplete cataract, posterior cortex                         |                     | 0         |      | 1         | 0.7% |
| 100.328 y-suture tip opacities  |                     | 0         |      | 1         | 0.7% |
| 100.330 generalized/complete cataract                                 |                     | 4         | 0.5% | 0         |      |
| 100.345 significant cataracts (summary)                               |                     | 66        | 8.5% | 7         | 4.6% |
| <b>VITREOUS</b>   |                     |           |      |           |      |
| 110.120 persistent hyaloid artery/remnant                             |                     | 3         | 0.4% | 2         | 1.3% |
| 110.135 PHPV/PTVL   |                     | 1         | 0.1% | 0         |      |
| 110.320 vitreal degeneration  |                     | 7         | 0.9% | 0         |      |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>FUNDUS</b>   |           |           |
| 97.120 coloboma   | 1 0.1%    | 0         |
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 20 2.6%   | 2 1.3%    |
| 120.180 retinal dysplasia, geographic                     | 9 1.2%    | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 12 1.6%   | 0         |
| 120.910 retinal detachment without dialysis               | 1 0.1%    | 0         |
| <b>OPTIC NERVE</b>  |           |           |
| 130.110 micropapilla                                      | 0         | 1 0.7%    |
| 130.120 optic nerve hypoplasia                            | 0         | 1 0.7%    |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 8 1.0%    | 0         |
| 900.100 other, not inherited                              | 35 4.5%   | 0         |
| 900.110 other. suspect not inherited/significance unknown | 11 1.4%   | 17 11.1%  |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 569 73.6% | 97 63.4%  |

# **OCULAR DISORDERS REPORT AKBASH DOG**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the AKBASH DOG breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT

## AKBASH DOG

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |   |
|--|---------------------|-----------|-------|-----------|---|
|  |                     | #         | %     | #         | % |
| <b>GLOBE</b>   |                     |           |       |           |   |
| 0.110 microphthalmia   |                     | 1         | 2.6%  | 0         |   |
| <b>EYELIDS</b>   |                     |           |       |           |   |
| 21.000 entropion, unspecified                                |                     | 3         | 7.7%  | 0         |   |
| 22.000 ectropion, unspecified                                |                     | 1         | 2.6%  | 0         |   |
| <b>UVEA</b>  |                     |           |       |           |   |
| 93.999 uveal cysts   |                     | 2         | 5.1%  | 0         |   |
| <b>LENS</b>  |                     |           |       |           |   |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 2         | 5.1%  | 0         |   |
| 100.303 punctate cataract, equatorial cortex                 |                     | 1         | 2.6%  | 0         |   |
| 100.316 incipient cataract, nucleus                          |                     | 1         | 2.6%  | 0         |   |
| 100.330 generalized/complete cataract                        |                     | 1         | 2.6%  | 0         |   |
| 100.345 <i>significant cataracts (summary)</i>               |                     | 3         | 7.7%  | 0         |   |
| <b>VITREOUS</b>  |                     |           |       |           |   |
| 110.120 persistent hyaloid artery/remnant                    |                     | 1         | 2.6%  | 0         |   |
| <b>NORMAL</b>  |                     |           |       |           |   |
| 0.000 normal globe   |                     | 32        | 82.1% | 0         |   |

# AKITA

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Microphthalmia with multiple ocular defects      | Not defined        | 1, 2             | NO                     |
| B. | Entropion  | Not defined        | 1, 3             | Breeder option         |
| C. | Distichiasis                                     | Not defined        | 1                | Breeder option         |
| D. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |
| E. | Cataract   | Not defined        | 1                | NO                     |
| F. | Y-suture tip opacity                             | Not defined        | 1                | Breeder option         |
| G. | Retinal dysplasia<br>- folds                     | Not defined        | 1                | Breeder option         |
| H. | Uveodermatologic syndrome                        | Not defined        | 1, 4-10          | NO                     |

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## Description and Comments

### A. Microphthalmia with multiple ocular defects

Multiple ocular defects consisting of small eye (microphthalmia), opacity of the lens (cataract), conical shape of the posterior lens (posterior lenticonus), and folding of the retina into rosettes (retinal dysplasia) have been reported in related Akita pups. Cataracts affected primarily the nuclear and cortical lens. Retinal dysplasia affected the superior retina overlying the tapetal fundus. Affected dogs may have severe visual dysfunction. An autosomal recessive mode of inheritance is suspected but not proven.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. OFA/CERF data indicates that entropion in the Akita usually occurs by 2 years of age.



C. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make strong recommendations with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

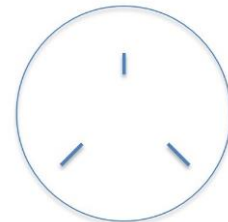
In the Akita, many of these strands bridge between the iris and lens, thus resulting in focal cataract and possible vision impairment.

E. Cataract

Lens opacity which may affect one or both eyes and may involve the lens partially or completely. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membranes, persistent hyaloid, or nutritional deficiencies.

F. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which

should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

G. Retinal Dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and the more severe forms of retinal dysplasia is undetermined.

H. Uveodermatologic syndrome

Uveodermatologic syndrome in the Akita bears many similarities to a condition in people called Vogt-Koyanagi-Harada (or VKH) syndrome. Thus, the condition in dogs is often referred to as VKH or VKH-like syndrome. It is an immune-mediated disease in which pigmented cells (melanocytes) in the eye and in the skin are destroyed by white blood cells (lymphocytes). The first clinical signs are usually inflammation of the intraocular structures (or uveitis) in both eyes. The uveitis is very difficult to control medically and ultimately results in blindness in most affected dogs. Whitening of the hair (poliosis) and skin (vitiligo) may also be noted in advanced cases. The genetics of this condition are unclear, but some genetic predisposition is indicated by the higher prevalence of this disorder in Akitas compared with other dog breeds. Affected dogs are generally young, ranging in age between 1 ½ to 4 years.

## References

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# OCULAR DISORDERS REPORT AKITA

| Diagnostic Name   | TOTAL DOGS EXAMINED |  | 1991-2015<br>10,752 |      | 2016-2020<br>888 |      |
|---|---------------------|--|---------------------|------|------------------|------|
|   |                     |  | #                   | %    | #                | %    |
| <b>GLOBE</b>  |                     |  |                     |      |                  |      |
| 0.110 microphthalmia  |                     |  | 33                  | 0.3% | 5                | 0.6% |
| 10.000 glaucoma   |                     |  | 2                   | 0.0% | 0                |      |
| <b>EYELIDS</b>  |                     |  |                     |      |                  |      |
| 21.000 entropion, unspecified   |                     |  | 103                 | 1.0% | 10               | 1.1% |
| 22.000 ectropion, unspecified   |                     |  | 15                  | 0.1% | 0                |      |
| 25.110 distichiasis   |                     |  | 67                  | 0.6% | 11               | 1.2% |
| <b>NASOLACRIMAL</b>   |                     |  |                     |      |                  |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     |  | 6                   | 0.1% | 4                | 0.5% |
| <b>NICTITANS</b>  |                     |  |                     |      |                  |      |
| 51.100 third eyelid cartilage anomaly                                 |                     |  | 7                   | 0.1% | 0                |      |
| <b>CORNEA</b>   |                     |  |                     |      |                  |      |
| 70.700 corneal dystrophy  |                     |  | 54                  | 0.5% | 7                | 0.8% |
| <b>UVEA</b>   |                     |  |                     |      |                  |      |
| 93.150 iris coloboma  |                     |  | 1                   | 0.0% | 0                |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     |  | 268                 | 2.5% | 24               | 2.7% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     |  | 37                  | 0.3% | 1                | 0.1% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     |  | 23                  | 0.2% | 5                | 0.6% |
| 93.740 persistent pupillary membranes, iris sheets                    |                     |  | 3                   | 0.0% | 1                | 0.1% |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     |  | 7                   | 0.1% | 12               | 1.4% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     |  | 2                   | 0.0% | 2                | 0.2% |
| 93.999 uveal cysts  |                     |  | 1                   | 0.0% | 0                |      |
| <b>LENS</b>   |                     |  |                     |      |                  |      |
| 100.200 cataract, unspecified   |                     |  | 28                  | 0.3% | 0                |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     |  | 243                 | 2.3% | 22               | 2.5% |
| 100.301 punctate cataract, anterior cortex                            |                     |  | 6                   | 0.1% | 1                | 0.1% |
| 100.302 punctate cataract, posterior cortex                           |                     |  | 7                   | 0.1% | 1                | 0.1% |
| 100.303 punctate cataract, equatorial cortex                          |                     |  | 4                   | 0.0% | 0                |      |
| 100.304 punctate cataract, anterior sutures                           |                     |  | 3                   | 0.0% | 0                |      |
| 100.305 punctate cataract, posterior sutures                          |                     |  | 33                  | 0.3% | 7                | 0.8% |
| 100.306 punctate cataract, nucleus                                    |                     |  | 2                   | 0.0% | 0                |      |
| 100.307 punctate cataract, capsular                                   |                     |  | 5                   | 0.0% | 4                | 0.5% |
| 100.311 incipient cataract, anterior cortex                           |                     |  | 10                  | 0.1% | 2                | 0.2% |
| 100.312 incipient cataract, posterior cortex                          |                     |  | 38                  | 0.4% | 0                |      |
| 100.313 incipient cataract, equatorial cortex                         |                     |  | 8                   | 0.1% | 0                |      |
| 100.314 incipient cataract, anterior sutures                          |                     |  | 2                   | 0.0% | 0                |      |
| 100.315 incipient cataract, posterior sutures                         |                     |  | 16                  | 0.1% | 2                | 0.2% |
| 100.316 incipient cataract, nucleus                                   |                     |  | 6                   | 0.1% | 1                | 0.1% |
| 100.317 incipient cataract, capsular                                  |                     |  | 9                   | 0.1% | 1                | 0.1% |
| 100.321 incomplete cataract, anterior cortex                          |                     |  | 0                   |      | 1                | 0.1% |
| 100.322 incomplete cataract, posterior cortex                         |                     |  | 1                   | 0.0% | 0                |      |
| 100.324 incomplete cataract, anterior sutures                         |                     |  | 0                   |      | 1                | 0.1% |
| 100.326 incomplete cataract, nucleus                                  |                     |  | 0                   |      | 1                | 0.1% |
| 100.328 y-suture tip opacities  |                     |  | 3                   | 0.0% | 8                | 0.9% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.330 generalized/complete cataract                     | 26               | 0.2%  | 2                | 0.2%  |
| 100.345 significant cataracts (summary)                   | 207              | 1.9%  | 32               | 3.6%  |
| 100.375 subluxation/luxation, unspecified                 | 1                | 0.0%  | 0                |       |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 16               | 0.1%  | 4                | 0.5%  |
| 110.135 PHPV/PTVL   | 5                | 0.0%  | 1                | 0.1%  |
| 110.320 vitreal degeneration                              | 10               | 0.1%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 200              | 1.9%  | 17               | 1.9%  |
| 120.180 retinal dysplasia, geographic                     | 21               | 0.2%  | 5                | 0.6%  |
| 120.190 retinal dysplasia, detached                       | 4                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 89               | 0.8%  | 1                | 0.1%  |
| 120.910 retinal detachment without dialysis               | 6                | 0.1%  | 0                |       |
| 120.960 retinopathy                                       | 0                |       | 1                | 0.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.120 optic nerve hypoplasia                            | 8                | 0.1%  | 1                | 0.1%  |
| 130.150 optic disc coloboma                               | 2                | 0.0%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 52               | 0.5%  | 0                |       |
| 900.100 other, not inherited                              | 179              | 1.7%  | 2                | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown | 93               | 0.9%  | 29               | 3.3%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 9,600            | 89.3% | 716              | 80.6% |

# ALASKAN KLEE KAI

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Distichiasis                                     | Not defined | 1         | Breeder option  |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined | 2         | Breeder option  |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Alaskan Klee Kai breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT ALASKAN KLEE KAI

| Diagnostic Name     | TOTAL DOGS EXAMINED                                  | 1991-2015<br>607 |       | 2016-2020<br>228 |       |
|---------------------|--|------------------|-------|------------------|-------|
|                     |  | #                | %     | #                | %     |
| <b>EYELIDS</b>      |  |                  |       |                  |       |
| 25.110              | distichiasis   | 48               | 7.9%  | 9                | 3.9%  |
| <b>NASOLACRIMAL</b> |  |                  |       |                  |       |
| 32.110              | imperforate lower nasolacrimal punctum               | 1                | 0.2%  | 3                | 1.3%  |
| <b>CORNEA</b>       |  |                  |       |                  |       |
| 70.220              | pigmentary keratitis                                 | 1                | 0.2%  | 0                |       |
| 70.700              | corneal dystrophy                                    | 11               | 1.8%  | 1                | 0.4%  |
| 70.730              | corneal endothelial degeneration                     | 2                | 0.3%  | 0                |       |
| <b>UVEA</b>         |  |                  |       |                  |       |
| 93.710              | persistent pupillary membranes, iris to iris         | 6                | 1.0%  | 5                | 2.2%  |
| 93.730              | persistent pupillary membranes, iris to cornea       | 1                | 0.2%  | 0                |       |
| 93.740              | persistent pupillary membranes, iris sheets          | 5                | 0.8%  | 0                |       |
| <b>LENS</b>         |  |                  |       |                  |       |
| 100.210             | cataract. suspect not inherited/significance unknown | 11               | 1.8%  | 10               | 4.4%  |
| 100.301             | punctate cataract, anterior cortex                   | 0                |       | 1                | 0.4%  |
| 100.304             | punctate cataract, anterior sutures                  | 0                |       | 1                | 0.4%  |
| 100.307             | punctate cataract, capsular                          | 1                | 0.2%  | 0                |       |
| 100.311             | incipient cataract, anterior cortex                  | 7                | 1.2%  | 1                | 0.4%  |
| 100.312             | incipient cataract, posterior cortex                 | 1                | 0.2%  | 0                |       |
| 100.345             | significant cataracts (summary)                      | 9                | 1.5%  | 3                | 1.3%  |
| <b>VITREOUS</b>     |  |                  |       |                  |       |
| 110.320             | vitreal degeneration                                 | 9                | 1.5%  | 0                |       |
| <b>RETINA</b>       |  |                  |       |                  |       |
| 120.170             | retinal dysplasia, folds                             | 5                | 0.8%  | 0                |       |
| <b>OTHER</b>        |  |                  |       |                  |       |
| 900.000             | other, unspecified                                   | 6                | 1.0%  | 0                |       |
| 900.100             | other, not inherited                                 | 4                | 0.7%  | 1                | 0.4%  |
| 900.110             | other. suspect not inherited/significance unknown    | 8                | 1.3%  | 6                | 2.6%  |
| <b>NORMAL</b>       |  |                  |       |                  |       |
| 0.000               | normal globe   | 527              | 86.8% | 194              | 85.1% |

# ALASKAN MALAMUTE

| DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE           |
|---|------------------------|-----------|--------------------|--------------------------------------|
| A. Distichiasis                                     | Not defined            | 1         | Breeder option     |                                      |
| B. Persistent pupillary membranes<br>- iris to iris | Not defined            | 1         | Breeder option     |                                      |
| C. Cataract   | Not defined            | 1         | NO                 |                                      |
| D. Cone degeneration<br>- day blindness             | Autosomal<br>recessive | 1, 2-8    | NO                 | Mutation in the<br><i>CNGB3</i> gene |

## Descriptions and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.



D. Cone degeneration - day blindness or hemeralopia

Autosomal recessively inherited early degeneration of the cone photoreceptors. Affected puppies develop day-blindness, color blindness, and photophobia between 8 and 12 weeks of age. Affected dogs remain ophthalmoscopically normal their entire life. Electroretinography is required to definitively diagnose the disorder. Genetically, the condition results from a deletion in the *CNGB3* gene. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Rubin LF, Bourns TKR, Lord LH. Hemeralopia in Dogs - Heredity of Hemeralopia in Alaskan Malamutes. *Am J Vet Res.* 1967;28:355-7.
3. Rubin LF. Clinical Features of Hemeralopia in Adult Alaskan Malamute. *J Am Vet Med Assoc.* 1971;158:1696-8.
4. Rubin LF. Hemeralopia in Alaskan Malamute Pups. *J Am Vet Med Assoc.* 1971;158:1699-701.
5. Aguirre GD, Rubin LF. Pathology of hemeralopia in the Alaskan malamute dog. *Invest Ophthalmol.* 1974;13:231-235.
6. Aguirre GD, Rubin LF. The electroretinogram in dogs with inherited cone degeneration. *Invest Ophthalmol.* 1975;14:840-847.
7. Seddon JM, Hampson ECGM, Smith RIE, et al. Genetic heterogeneity of day blindness in Alaskan Malamute. *Anim Genet.* 2006;37:407-410.
8. Sidjanin DJ, Lowe JK, McElwee JL, et al. Canine CNGB3 mutations establish cone degeneration as orthologous to the human achromatopsia locus ACHM3. *Hum Mol Genet.* 2002;11:1823-1833.

# OCULAR DISORDERS REPORT ALASKAN MALAMUTE

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>8,712 |      | 2016-2020<br>1,067 |      |
|---------------------|--|--------------------|------|--------------------|------|
|                     |  | #                  | %    | #                  | %    |
| <b>GLOBE</b>        |  |                    |      |                    |      |
| 0.110               | microphthalmia   | 2                  | 0.0% | 0                  |      |
| 10.000              | glaucoma   | 2                  | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                    |      |                    |      |
| 20.140              | ectopic cilia  | 1                  | 0.0% | 0                  |      |
| 21.000              | entropion, unspecified   | 5                  | 0.1% | 0                  |      |
| 22.000              | ectropion, unspecified   | 1                  | 0.0% | 0                  |      |
| 25.110              | distichiasis   | 198                | 2.3% | 14                 | 1.3% |
| <b>NASOLACRIMAL</b> |  |                    |      |                    |      |
| 40.910              | keratoconjunctivitis sicca                                     | 2                  | 0.0% | 0                  |      |
| <b>NICTITANS</b>    |  |                    |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 1                  | 0.0% | 0                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 1                  | 0.0% | 0                  |      |
| <b>CORNEA</b>       |  |                    |      |                    |      |
| 70.220              | pigmentary keratitis   | 0                  |      | 1                  | 0.1% |
| 70.700              | corneal dystrophy  | 72                 | 0.8% | 13                 | 1.2% |
| <b>UVEA</b>         |  |                    |      |                    |      |
| 93.140              | corneal endothelial pigment without PPM                        | 1                  | 0.0% | 0                  |      |
| 93.150              | iris coloboma  | 3                  | 0.0% | 0                  |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 559                | 6.4% | 83                 | 7.8% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 35                 | 0.4% | 7                  | 0.7% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 12                 | 0.1% | 0                  |      |
| 93.740              | persistent pupillary membranes, iris sheets                    | 4                  | 0.0% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 8                  | 0.1% | 7                  | 0.7% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 4                  | 0.0% | 0                  |      |
| 93.810              | uveal melanoma   | 2                  | 0.0% | 0                  |      |
| 93.999              | uveal cysts  | 6                  | 0.1% | 2                  | 0.2% |
| <b>LENS</b>         |  |                    |      |                    |      |
| 100.200             | cataract, unspecified  | 125                | 1.4% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 347                | 4.0% | 54                 | 5.1% |
| 100.301             | punctate cataract, anterior cortex                             | 21                 | 0.2% | 5                  | 0.5% |
| 100.302             | punctate cataract, posterior cortex                            | 137                | 1.6% | 12                 | 1.1% |
| 100.303             | punctate cataract, equatorial cortex                           | 14                 | 0.2% | 3                  | 0.3% |
| 100.304             | punctate cataract, anterior sutures                            | 16                 | 0.2% | 1                  | 0.1% |
| 100.305             | punctate cataract, posterior sutures                           | 64                 | 0.7% | 3                  | 0.3% |
| 100.306             | punctate cataract, nucleus                                     | 8                  | 0.1% | 5                  | 0.5% |
| 100.307             | punctate cataract, capsular                                    | 28                 | 0.3% | 4                  | 0.4% |
| 100.311             | incipient cataract, anterior cortex                            | 27                 | 0.3% | 6                  | 0.6% |
| 100.312             | incipient cataract, posterior cortex                           | 349                | 4.0% | 30                 | 2.8% |
| 100.313             | incipient cataract, equatorial cortex                          | 40                 | 0.5% | 4                  | 0.4% |
| 100.314             | incipient cataract, anterior sutures                           | 8                  | 0.1% | 1                  | 0.1% |
| 100.315             | incipient cataract, posterior sutures                          | 78                 | 0.9% | 6                  | 0.6% |
| 100.316             | incipient cataract, nucleus                                    | 19                 | 0.2% | 3                  | 0.3% |
| 100.317             | incipient cataract, capsular                                   | 39                 | 0.4% | 1                  | 0.1% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.321 incomplete cataract, anterior cortex              | 2 0.0%           | 3 0.3%           |
| 100.322 incomplete cataract, posterior cortex             | 13 0.1%          | 15 1.4%          |
| 100.323 incomplete cataract, equatorial cortex            | 1 0.0%           | 0                |
| 100.324 incomplete cataract, anterior sutures             | 1 0.0%           | 0                |
| 100.325 incomplete cataract, posterior sutures            | 2 0.0%           | 1 0.1%           |
| 100.326 incomplete cataract, nucleus                      | 2 0.0%           | 1 0.1%           |
| 100.327 incomplete cataract, capsular                     | 0                | 5 0.5%           |
| 100.328 y-suture tip opacities                            | 3 0.0%           | 1 0.1%           |
| 100.330 generalized/complete cataract                     | 80 0.9%          | 1 0.1%           |
| 100.345 <i>significant cataracts (summary)</i>            | 1,077 12.4%      | 111 10.4%        |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 7 0.1%           | 1 0.1%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 9 0.1%           | 3 0.3%           |
| 110.135 PHPV/PTVL   | 6 0.1%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 0                | 1 0.1%           |
| 110.320 vitreal degeneration                              | 13 0.1%          | 1 0.1%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 3 0.0%           | 0                |
| 97.120 coloboma   | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 60 0.7%          | 0                |
| 120.180 retinal dysplasia, geographic                     | 19 0.2%          | 2 0.2%           |
| 120.190 retinal dysplasia, detached                       | 2 0.0%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 17 0.2%          | 1 0.1%           |
| 120.400 retinal hemorrhage                                | 2 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 10 0.1%          | 0                |
| 120.920 retinal detachment with dialysis                  | 0                | 1 0.1%           |
| 120.960 retinopathy                                       | 1 0.0%           | 1 0.1%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 2 0.0%           | 1 0.1%           |
| 130.120 optic nerve hypoplasia                            | 9 0.1%           | 0                |
| 130.150 optic disc coloboma                               | 2 0.0%           | 2 0.2%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 75 0.9%          | 0                |
| 900.100 other, not inherited                              | 257 2.9%         | 3 0.3%           |
| 900.110 other. suspect not inherited/significance unknown | 108 1.2%         | 46 4.3%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 6,864 78.8%      | 767 71.9%        |

# ALASKAN NOBLE COMPANION DOG

| DISORDER  | INHERITANCE | REFERENCE | BREEDING ADVICE |
|---|-------------|-----------|-----------------|
| A. Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Alaskan Noble Companion Dog breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT ALASKAN NOBLE COMPANION DOG

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015<br>53 |       | 2016-2020<br>33 |       |
|--|---------------------|-----------------|-------|-----------------|-------|
|  |                     | #               | %     | #               | %     |
| <b>UVEA</b>  |                     |                 |       |                 |       |
| 93.710 persistent pupillary membranes, iris to iris          |                     | 4               | 7.5%  | 1               | 3.0%  |
| 93.999 uveal cysts   |                     | 0               |       | 1               | 3.0%  |
| <b>LENS</b>  |                     |                 |       |                 |       |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 0               |       | 1               | 3.0%  |
| <b>RETINA</b>  |                     |                 |       |                 |       |
| 120.170 retinal dysplasia, folds                             |                     | 1               | 1.9%  | 0               |       |
| <b>OTHER</b>   |                     |                 |       |                 |       |
| 900.110 other. suspect not inherited/significance unknown    |                     | 0               |       | 1               | 3.0%  |
| <b>NORMAL</b>  |                     |                 |       |                 |       |
| 0.000 normal globe   |                     | 50              | 94.3% | 30              | 90.9% |

# AMERICAN BULLDOG

| DISORDER                                | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|---|---------------------|-----------|-----------------|-----------------------------------|
| A. Glaucoma                             | Not defined         | 2         | NO              |                                   |
| B. Distichiasis                         | Not defined         | 1         | Breeder option  |                                   |
| C. Multifocal retinopathy - <i>cmr1</i> | Autosomal recessive | 3         | Breeder Option  | Mutation in the <i>BEST1</i> gene |

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## Description and Comments

### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure which, when sustained even for a brief period of time, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine breed eye screening exam.

American Bulldogs with glaucoma were reported to have uveal cysts (evident on ophthalmic exam, ultrasound biomicroscopy and/or histopathology), goniodysgenesis, and anterior segment inflammation. Consistent clinical findings among reported individuals included an absent menace response, diminished to absent light perception, mydriasis, and elevated intraocular pressures.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### C. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (*cmr1*) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is

minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas. A DNA test is available.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Pumphrey SA, Pizzirani S, Pirie CG, et al. Glaucoma associated with uveal cysts and goniodysgenesis in American Bulldogs: a case series. *Vet Ophthalmol*. 2012;1-9.
3. Guziewicz KE, Zangerl B, Lindauer SJ, et al. Bestrophin gene mutations cause canine multifocal retinopathy: a novel animal model for best disease. *Invest Ophthalmol Vis Sci*. 2007 May;48:1959-1967.

# OCULAR DISORDERS REPORT AMERICAN BULLDOG

| Diagnostic Name     | TOTAL DOGS EXAMINED                                  | 1991-2015<br>120 |       | 2016-2020<br>28 |       |
|---------------------|--|------------------|-------|-----------------|-------|
|                     |  | #                | %     | #               | %     |
| <b>EYELIDS</b>      |  |                  |       |                 |       |
| 20.160              | macropalpebral fissure                               | 3                | 2.5%  | 0               |       |
| 21.000              | entropion, unspecified                               | 9                | 7.5%  | 0               |       |
| 22.000              | ectropion, unspecified                               | 2                | 1.7%  | 0               |       |
| 25.110              | distichiasis   | 30               | 25.0% | 1               | 3.6%  |
| <b>NASOLACRIMAL</b> |  |                  |       |                 |       |
| 40.910              | keratoconjunctivitis sicca                           | 4                | 3.3%  | 0               |       |
| <b>CORNEA</b>       |  |                  |       |                 |       |
| 70.220              | pigmentary keratitis                                 | 1                | 0.8%  | 0               |       |
| <b>UVEA</b>         |  |                  |       |                 |       |
| 93.710              | persistent pupillary membranes, iris to iris         | 1                | 0.8%  | 4               | 14.3% |
| 93.720              | persistent pupillary membranes, iris to lens         | 0                |       | 1               | 3.6%  |
| 93.730              | persistent pupillary membranes, iris to cornea       | 1                | 0.8%  | 0               |       |
| 93.999              | uveal cysts  | 1                | 0.8%  | 0               |       |
| <b>LENS</b>         |  |                  |       |                 |       |
| 100.210             | cataract. suspect not inherited/significance unknown | 1                | 0.8%  | 2               | 7.1%  |
| <b>RETINA</b>       |  |                  |       |                 |       |
| 120.170             | retinal dysplasia, folds                             | 3                | 2.5%  | 0               |       |
| <b>OTHER</b>        |  |                  |       |                 |       |
| 900.000             | other, unspecified                                   | 16               | 13.3% | 0               |       |
| 900.110             | other. suspect not inherited/significance unknown    | 1                | 0.8%  | 0               |       |
| <b>NORMAL</b>       |  |                  |       |                 |       |
| 0.000               | normal globe   | 84               | 70.0% | 21              | 75.0% |



# AMERICAN ESKIMO DOG

(all varieties)

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|------------------------------------|------------------------|-----------|--------------------|---|
| A. | Cataract                           | Not defined            | 1         | NO                 |   |
| B. | Lens luxation                      | Autosomal<br>recessive | 2         | NO                 | Mutation in the<br><i>ADAMTS17</i> gene |
| C. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 3         | NO                 | Mutation in the<br><i>prcd</i> gene     |

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## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### B. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

### C. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the American Eskimo is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. However in the American Eskimo Dog the phenotype can be very variable in the age of onset. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease

begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

Other forms of retinal degeneration that are not *prcd* are recognized in the breed. The currently available genetic test will not detect these other forms of PRA.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gould D, Pettitt L, McLaughlin B, et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011;14:378-384.
3. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics.* 2006;88:551-563. PMID: 16938425

# OCULAR DISORDERS REPORT AMERICAN ESKIMO DOG

| Diagnostic Name     | TOTAL DOGS EXAMINED                                  | 1991-2015<br>2,423 |      | 2016-2020<br>222 |      |
|---------------------|--|--------------------|------|------------------|------|
|                     |  | #                  | %    | #                | %    |
| <b>EYELIDS</b>      |  |                    |      |                  |      |
| 21.000              | entropion, unspecified                               | 4                  | 0.2% | 0                |      |
| 25.110              | distichiasis   | 18                 | 0.7% | 1                | 0.5% |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |      |
| 32.110              | imperforate lower nasolacrimal punctum               | 1                  | 0.0% | 0                |      |
| <b>CORNEA</b>       |  |                    |      |                  |      |
| 70.700              | corneal dystrophy                                    | 9                  | 0.4% | 3                | 1.4% |
| 70.730              | corneal endothelial degeneration                     | 4                  | 0.2% | 0                |      |
| <b>UVEA</b>         |  |                    |      |                  |      |
| 93.710              | persistent pupillary membranes, iris to iris         | 21                 | 0.9% | 0                |      |
| 93.720              | persistent pupillary membranes, iris to lens         | 1                  | 0.0% | 0                |      |
| 93.730              | persistent pupillary membranes, iris to cornea       | 4                  | 0.2% | 1                | 0.5% |
| 93.740              | persistent pupillary membranes, iris sheets          | 4                  | 0.2% | 0                |      |
| 93.999              | uveal cysts  | 4                  | 0.2% | 0                |      |
| <b>LENS</b>         |  |                    |      |                  |      |
| 100.200             | cataract, unspecified                                | 3                  | 0.1% | 0                |      |
| 100.210             | cataract. suspect not inherited/significance unknown | 136                | 5.6% | 16               | 7.2% |
| 100.301             | punctate cataract, anterior cortex                   | 24                 | 1.0% | 2                | 0.9% |
| 100.302             | punctate cataract, posterior cortex                  | 9                  | 0.4% | 1                | 0.5% |
| 100.303             | punctate cataract, equatorial cortex                 | 6                  | 0.2% | 0                |      |
| 100.304             | punctate cataract, anterior sutures                  | 3                  | 0.1% | 0                |      |
| 100.305             | punctate cataract, posterior sutures                 | 4                  | 0.2% | 0                |      |
| 100.306             | punctate cataract, nucleus                           | 3                  | 0.1% | 0                |      |
| 100.307             | punctate cataract, capsular                          | 3                  | 0.1% | 1                | 0.5% |
| 100.311             | incipient cataract, anterior cortex                  | 21                 | 0.9% | 5                | 2.3% |
| 100.312             | incipient cataract, posterior cortex                 | 23                 | 0.9% | 1                | 0.5% |
| 100.313             | incipient cataract, equatorial cortex                | 13                 | 0.5% | 0                |      |
| 100.314             | incipient cataract, anterior sutures                 | 5                  | 0.2% | 0                |      |
| 100.315             | incipient cataract, posterior sutures                | 3                  | 0.1% | 0                |      |
| 100.316             | incipient cataract, nucleus                          | 6                  | 0.2% | 1                | 0.5% |
| 100.317             | incipient cataract, capsular                         | 6                  | 0.2% | 0                |      |
| 100.323             | incomplete cataract, equatorial cortex               | 1                  | 0.0% | 0                |      |
| 100.326             | incomplete cataract, nucleus                         | 0                  |      | 1                | 0.5% |
| 100.327             | incomplete cataract, capsular                        | 1                  | 0.0% | 1                | 0.5% |
| 100.330             | generalized/complete cataract                        | 10                 | 0.4% | 1                | 0.5% |
| 100.340             | resorbing/hypermature cataract                       | 1                  | 0.0% | 0                |      |
| 100.345             | <i>significant cataracts (summary)</i>               | 145                | 6.0% | 14               | 6.3% |
| 100.375             | <i>subluxation/luxation, unspecified</i>             | 3                  | 0.1% | 0                |      |
| <b>VITREOUS</b>     |  |                    |      |                  |      |
| 110.120             | persistent hyaloid artery/remnant                    | 6                  | 0.2% | 2                | 0.9% |
| 110.135             | PHPV/PTVL  | 3                  | 0.1% | 0                |      |
| 110.200             | vitreous degeneration-anterior chamber               | 1                  | 0.0% | 0                |      |
| 110.320             | vitreal degeneration                                 | 18                 | 0.7% | 0                |      |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 8 0.3%      | 1 0.5%    |
| 120.180 retinal dysplasia, geographic                     | 2 0.1%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 182 7.5%    | 3 1.4%    |
| 120.910 retinal detachment without dialysis               | 1 0.0%      | 0         |
| 120.960 retinopathy                                       | 1 0.0%      | 0         |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 2 0.1%      | 0         |
| 130.120 optic nerve hypoplasia                            | 1 0.0%      | 0         |
| 130.150 optic disc coloboma                               | 3 0.1%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 8 0.3%      | 0         |
| 900.100 other, not inherited                              | 86 3.5%     | 0         |
| 900.110 other. suspect not inherited/significance unknown | 25 1.0%     | 9 4.1%    |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,938 80.0% | 180 81.1% |

# AMERICAN HAIRLESS TERRIER

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|------------------------------------|------------------------|-----------|--------------------|---|
| A. | Lens luxation                      | Autosomal<br>recessive | 2, 3      | NO                 | Mutation in the<br><i>ADAMTS17</i> gene |
| B. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>prcd</i> gene     |

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## Description and Comments

### A. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

### B. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the American Hairless Terrier is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. However in the American Eskimo Dog the phenotype can be very variable in the age of onset. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

Other forms of retinal degeneration that are not PRCD are recognized in the breed. The currently available genetic test will not detect these other forms of PRA.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Farias FH, Johnson GS, Taylor JF, et al. An ADAMTS17 splice donor site mutation in dogs with primary lens luxation. *Invest Ophthalmol Vis Sci*. 2010 Sep;51:4716-4721.
3. Gould D, Pettitt L, McLaughlin B, et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol*. 2011 Nov;14:378-384.

# OCULAR DISORDERS REPORT AMERICAN HAIRLESS TERRIER

| Diagnostic Name | TOTAL DOGS EXAMINED                                  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 0         |       | 1         | 1.5%  |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris         | 1         | 3.8%  | 1         | 1.5%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown | 1         | 3.8%  | 0         |       |
| 100.305         | punctate cataract, posterior sutures                 | 0         |       | 1         | 1.5%  |
| 100.345         | <i>significant cataracts (summary)</i>               | 0         |       | 1         | 1.5%  |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                             | 0         |       | 1         | 1.5%  |
| 120.910         | retinal detachment without dialysis                  | 1         | 3.8%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.000         | other, unspecified                                   | 1         | 3.8%  | 0         |       |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 23        | 88.5% | 62        | 93.9% |

# AMERICAN PIT BULL TERRIER

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|--|---------------------|-----------|-----------------|-----------------------------------|
| A. | Retinal atrophy - cone-rod dystrophy 2 ( <i>crd2</i> )       | Autosomal recessive | 2-4       | NO              | Mutation in the <i>IQCB1</i> gene |
| B. | Retinal atrophy - cone-rod dystrophy 1 ( <i>CRD1/rcd1b</i> ) | Autosomal recessive | 1         | NO              | Mutation in the <i>PDE6B</i> gene |

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## Description and Comments

### A. Retinal atrophy - cone-rod dystrophy 2 (*crd2*)

A cone rod dystrophy characterized by initial loss of cones (day vision) followed by degeneration of the rods (night vision). Evidence of vision loss is evident at an early age with severe retinal degeneration and complete blindness by a year of age. The disease is a severe early onset retinal blindness more appropriately considered a form of Leber congenital amaurosis (LCA). The condition is inherited as an autosomal recessive trait and caused by a mutation in *IQCB1*. A DNA test is available.

### B. Retinal Atrophy - Rod-cone dysplasia 1b [previously considered cone-rod dystrophy 1 (*crd1*)]

The disease was previously considered a cone-rod dystrophy (*crd1*) based on incorrect phenotype ascertainment using ERG. The term *crd1* should no longer be used to refer to the disease in this breed. The disease is more appropriately classified as rod-cone dysplasia 1b (*rcd1b*). In affected dogs there is evidence of vision loss at an early age with severe retinal degeneration and complete blindness by early adulthood, and ophthalmoscopic evidence of advanced retinal degeneration by 1 year of age. The disease is caused by a mutation in the *PDE6B* gene, with clinical abnormalities similar to what is found in *rcd1*-affected Irish Setters, and *rcd1a* affected Sloughis and Spanish Water Dogs. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Miyadera K, Acland GM, Aguirre GD. Genetic and phenotypic variations of inherited retinal diseases in dogs: the power of within- and across-breed studies. *Mamm Genome*. 2012;23:40-61.
3. Goldstein O, Mezey JG, Schweitzer P, et al. *IQCB1* and *PDE6B* mutations cause similar early onset retinal degenerations in two closely related terrier dog breeds. *Invest Ophthalmol*. 2013;54:7005-7019.



4. Kijas JW, Zanger B, Miller B, et al. Cloning of the canine ABCA4 gene and evaluation in canine cone-rod dystrophies and progressive retinal atrophies. *Mol Vis*. 2004;10:223-232.

# OCULAR DISORDERS REPORT AMERICAN PIT BULL TERRIER

| Diagnostic Name | TOTAL DOGS EXAMINED                                  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 5         | 2.6%  | 3         | 6.7%  |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.700          | corneal dystrophy                                    | 1         | 0.5%  | 0         |       |
| 70.730          | corneal endothelial degeneration                     | 1         | 0.5%  | 0         |       |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris         | 4         | 2.1%  | 2         | 4.4%  |
| 93.720          | persistent pupillary membranes, iris to lens         | 2         | 1.1%  | 0         |       |
| 93.730          | persistent pupillary membranes, iris to cornea       | 2         | 1.1%  | 0         |       |
| 93.740          | persistent pupillary membranes, iris sheets          | 1         | 0.5%  | 0         |       |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown | 7         | 3.7%  | 0         |       |
| 100.301         | punctate cataract, anterior cortex                   | 1         | 0.5%  | 0         |       |
| 100.302         | punctate cataract, posterior cortex                  | 2         | 1.1%  | 0         |       |
| 100.305         | punctate cataract, posterior sutures                 | 1         | 0.5%  | 0         |       |
| 100.326         | incomplete cataract, nucleus                         | 1         | 0.5%  | 0         |       |
| 100.328         | y-suture tip opacities                               | 0         |       | 1         | 2.2%  |
| 100.345         | <i>significant cataracts (summary)</i>               | 5         | 2.6%  | 1         | 2.2%  |
| 100.375         | <i>subluxation/luxation, unspecified</i>             | 1         | 0.5%  | 0         |       |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                             | 2         | 1.1%  | 0         |       |
| 120.180         | retinal dysplasia, geographic                        | 1         | 0.5%  | 0         |       |
| 120.310         | generalized progressive retinal atrophy (PRA)        | 2         | 1.1%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.000         | other, unspecified                                   | 1         | 0.5%  | 0         |       |
| 900.100         | other, not inherited                                 | 10        | 5.3%  | 1         | 2.2%  |
| 900.110         | other. suspect not inherited/significance unknown    | 0         |       | 2         | 4.4%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 162       | 85.7% | 36        | 80.0% |

## AMERICAN STAFFORDSHIRE TERRIER\*

\*Please note that since 1972 the AKC considers the Staffordshire Bull Terrier a different breed from the American Staffordshire Terrier. Since the latter breed evolved from the former, it is possible that the same genetic diseases exist in both.

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE           |
|----|---|------------------------|-----------|--------------------|--------------------------------------|
| A. | Distichiasis  | Not defined            | 1         | Breeder<br>option  |                                      |
| B. | Persistent pupillary<br>membranes<br>- iris to iris                 | Not defined            | 1         | Breeder<br>option  |                                      |
| C. | Cataract  | Not defined            | 1, 2, 3   | NO                 |                                      |
| D. | Retinal atrophy - cone-<br>rod dystrophy 2 ( <i>crd2</i> )          | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>IQCB1</i> gene |
| E. | Retinal atrophy - cone-<br>rod dystrophy 1<br>( <i>CRD1/rcd1b</i> ) | Autosomal<br>recessive | 4-6       | NO                 | Mutation in the<br><i>PDE6B</i> gene |

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### Description and Comments

#### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make strong recommendations with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

#### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

#### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes

of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In this breed, cataracts usually develop by one year of age. There is initial opacification of the suture lines progressing to nuclear and cortical cataract formation; complete cataracts and blindness develop by three years of age. A simple autosomal recessive mode of inheritance has been proposed; however, the genetics have not been defined and additional studies will be required.

D. Retinal Atrophy - Cone-rod dystrophy 2 (*crd2*)

A cone rod dystrophy characterized by initial loss of cones (day vision) followed by degeneration of the rods (night vision). Evidence of vision loss is evident at an early age with severe retinal degeneration and complete blindness by a year of age. The disease is a severe early onset retinal blindness more appropriately considered a form of Leber congenital amaurosis (LCA). The condition is inherited as an autosomal recessive trait and caused by a mutation in *IQCB1*. A DNA test is available.

E. Retinal Atrophy - Rod-cone dysplasia 1b [previously considered cone-rod dystrophy 1 (*crd1*)]

The disease was previously considered a cone-rod dystrophy (*crd1*) based on incorrect phenotype ascertainment using ERG (Aguirre, personal communication, 2016). The term *crd1* should no longer be used to refer to the disease in this breed. The disease is more appropriately classified as rod-cone dysplasia 1b (*rcd1b*). In affected dogs there is evidence of vision loss at an early age with severe retinal degeneration and complete blindness by early adulthood, and ophthalmoscopic evidence of advanced retinal degeneration by 1 year of age. The disease is caused by a mutation in the *PDE6B* gene, with clinical abnormalities similar to what is found in *rcd1*-affected Irish Setters, and *rcd1a* affected Sloughis and Spanish Water Dogs. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Barnett KC. Hereditary cataract in the dog. *J Small Anim Pract.* 1978;19:109-120.
3. Barnett KC. The diagnosis and differential diagnosis of cataract in the dog. *J Small Anim Pract.* 1985;26:305-316.
4. Kijas JW, Zanger B, Miller B, et al. Cloning of the canine ABCA4 gene and evaluation in canine cone-rod dystrophies and progressive retinal atrophies. *Mol Vis.* 2004;10:223-232.
5. Miyadera K, Acland GM, Aguirre GD. Genetic and phenotypic variations of inherited retinal diseases in dogs: the power of within- and across-breed studies. *Mamm Genome.* 2012;23:40-61.
6. Goldstein O, Mezey JG, Schweitzer P, et al. IQCB1 and PDE6B mutations cause similar early onset retinal degenerations in two closely related terrier dog breeds. *Invest Ophthalmol.* 2013;54:7005-7019.

# OCULAR DISORDERS REPORT AMERICAN STAFFORDSHIRE TERRIER

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015<br>726 |      | 2016-2020<br>98 |      |
|-----------------|--|------------------|------|-----------------|------|
|                 |  | #                | %    | #               | %    |
| <b>EYELIDS</b>  |  |                  |      |                 |      |
| 21.000          | entropion, unspecified   | 2                | 0.3% | 0               |      |
| 25.110          | distichiasis   | 34               | 4.7% | 2               | 2.0% |
| <b>CORNEA</b>   |  |                  |      |                 |      |
| 70.210          | corneal pannus   | 1                | 0.1% | 0               |      |
| 70.220          | pigmentary keratitis   | 1                | 0.1% | 1               | 1.0% |
| 70.730          | corneal endothelial degeneration                               | 1                | 0.1% | 0               |      |
| <b>UVEA</b>     |  |                  |      |                 |      |
| 93.110          | iris hypoplasia  | 0                |      | 1               | 1.0% |
| 93.710          | persistent pupillary membranes, iris to iris                   | 30               | 4.1% | 3               | 3.1% |
| 93.720          | persistent pupillary membranes, iris to lens                   | 2                | 0.3% | 0               |      |
| 93.730          | persistent pupillary membranes, iris to cornea                 | 1                | 0.1% | 0               |      |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands   | 1                | 0.1% | 0               |      |
| 93.760          | persistent pupillary membranes, endothelial opacity/no strands | 1                | 0.1% | 0               |      |
| 93.999          | uveal cysts  | 2                | 0.3% | 1               | 1.0% |
| <b>LENS</b>     |  |                  |      |                 |      |
| 100.200         | cataract, unspecified  | 1                | 0.1% | 0               |      |
| 100.210         | cataract. suspect not inherited/significance unknown           | 28               | 3.9% | 5               | 5.1% |
| 100.301         | punctate cataract, anterior cortex                             | 1                | 0.1% | 0               |      |
| 100.302         | punctate cataract, posterior cortex                            | 2                | 0.3% | 0               |      |
| 100.303         | punctate cataract, equatorial cortex                           | 2                | 0.3% | 0               |      |
| 100.304         | punctate cataract, anterior sutures                            | 1                | 0.1% | 0               |      |
| 100.305         | punctate cataract, posterior sutures                           | 1                | 0.1% | 0               |      |
| 100.311         | incipient cataract, anterior cortex                            | 4                | 0.6% | 0               |      |
| 100.312         | incipient cataract, posterior cortex                           | 3                | 0.4% | 0               |      |
| 100.313         | incipient cataract, equatorial cortex                          | 4                | 0.6% | 0               |      |
| 100.323         | incomplete cataract, equatorial cortex                         | 0                |      | 1               | 1.0% |
| 100.328         | y-suture tip opacities   | 0                |      | 1               | 1.0% |
| 100.330         | generalized/complete cataract                                  | 1                | 0.1% | 0               |      |
| 100.345         | <i>significant cataracts (summary)</i>                         | 20               | 2.8% | 2               | 2.0% |
| 100.375         | <i>subluxation/luxation, unspecified</i>                       | 2                | 0.3% | 0               |      |
| <b>VITREOUS</b> |  |                  |      |                 |      |
| 110.120         | persistent hyaloid artery/remnant                              | 2                | 0.3% | 0               |      |
| 110.320         | vitreal degeneration   | 3                | 0.4% | 0               |      |
| <b>RETINA</b>   |  |                  |      |                 |      |
| 120.170         | retinal dysplasia, folds                                       | 8                | 1.1% | 0               |      |
| 120.180         | retinal dysplasia, geographic                                  | 2                | 0.3% | 0               |      |
| 120.310         | generalized progressive retinal atrophy (PRA)                  | 3                | 0.4% | 0               |      |
| <b>OTHER</b>    |  |                  |      |                 |      |
| 900.000         | other, unspecified   | 8                | 1.1% | 0               |      |
| 900.100         | other, not inherited   | 30               | 4.1% | 0               |      |
| 900.110         | other. suspect not inherited/significance unknown              | 8                | 1.1% | 5               | 5.1% |

|                                     | 1991-2015 | 2016-2020 |
|-------------------------------------|-----------|-----------|
| <b>NORMAL</b><br>0.000 normal globe | 620 85.4% | 80 81.6%  |

# AMERICAN WATER SPANIEL

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--|-------------|-----------|-------------------------|
| A. | Distichiasis   | Not defined | 1         | Breeder option          |
| B. | Persistent pupillary membranes<br>- lens pigment foci/no strands | Not defined | 1         | Passes with no notation |
| C. | Cataract   | Not defined | 1         | NO                      |
| D. | Y-suture tip opacity   | Not defined | 1         | Breeder Option          |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

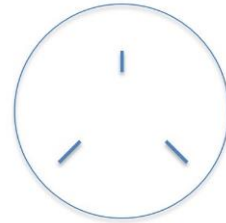
Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

#### D. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the American Water Spaniel breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.



# OCULAR DISORDERS REPORT AMERICAN WATER SPANIEL

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 0.110 microphthalmia  |                     | 2         | 0.2%  | 0         |       |
| 10.000 glaucoma   |                     | 3         | 0.3%  | 0         |       |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 20.160 macropalpebral fissure                                       |                     | 2         | 0.2%  | 0         |       |
| 21.000 entropion, unspecified                                       |                     | 7         | 0.7%  | 1         | 0.8%  |
| 22.000 ectropion, unspecified                                       |                     | 2         | 0.2%  | 0         |       |
| 25.110 distichiasis   |                     | 336       | 32.0% | 54        | 43.2% |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.220 pigmentary keratitis   |                     | 1         | 0.1%  | 0         |       |
| 70.700 corneal dystrophy  |                     | 5         | 0.5%  | 2         | 1.6%  |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.150 iris coloboma  |                     | 2         | 0.2%  | 0         |       |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 11        | 1.0%  | 1         | 0.8%  |
| 93.730 persistent pupillary membranes, iris to cornea               |                     | 1         | 0.1%  | 0         |       |
| 93.740 persistent pupillary membranes, iris sheets                  |                     | 2         | 0.2%  | 0         |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 6         | 0.6%  | 4         | 3.2%  |
| 93.999 uveal cysts  |                     | 1         | 0.1%  | 0         |       |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.200 cataract, unspecified                                       |                     | 5         | 0.5%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 38        | 3.6%  | 8         | 6.4%  |
| 100.301 punctate cataract, anterior cortex                          |                     | 5         | 0.5%  | 0         |       |
| 100.302 punctate cataract, posterior cortex                         |                     | 7         | 0.7%  | 0         |       |
| 100.303 punctate cataract, equatorial cortex                        |                     | 2         | 0.2%  | 0         |       |
| 100.305 punctate cataract, posterior sutures                        |                     | 5         | 0.5%  | 3         | 2.4%  |
| 100.306 punctate cataract, nucleus                                  |                     | 1         | 0.1%  | 0         |       |
| 100.307 punctate cataract, capsular                                 |                     | 2         | 0.2%  | 0         |       |
| 100.311 incipient cataract, anterior cortex                         |                     | 7         | 0.7%  | 0         |       |
| 100.312 incipient cataract, posterior cortex                        |                     | 11        | 1.0%  | 4         | 3.2%  |
| 100.315 incipient cataract, posterior sutures                       |                     | 5         | 0.5%  | 1         | 0.8%  |
| 100.317 incipient cataract, capsular                                |                     | 1         | 0.1%  | 1         | 0.8%  |
| 100.322 incomplete cataract, posterior cortex                       |                     | 0         |       | 1         | 0.8%  |
| 100.327 incomplete cataract, capsular                               |                     | 0         |       | 1         | 0.8%  |
| 100.328 y-suture tip opacities                                      |                     | 1         | 0.1%  | 2         | 1.6%  |
| 100.330 generalized/complete cataract                               |                     | 1         | 0.1%  | 0         |       |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 53        | 5.0%  | 13        | 10.4% |
| 100.375 <i>subluxation/luxation, unspecified</i>                    |                     | 0         |       | 1         | 0.8%  |
| <b>VITREOUS</b>   |                     |           |       |           |       |
| 110.120 persistent hyaloid artery/remnant                           |                     | 2         | 0.2%  | 0         |       |
| 110.135 PHPV/PTVL   |                     | 0         |       | 1         | 0.8%  |
| 110.320 vitreal degeneration  |                     | 0         |       | 2         | 1.6%  |
| <b>RETINA</b>   |                     |           |       |           |       |
| 120.170 retinal dysplasia, folds                                    |                     | 8         | 0.8%  | 0         |       |
| 120.180 retinal dysplasia, geographic                               |                     | 1         | 0.1%  | 0         |       |
| 120.310 generalized progressive retinal atrophy (PRA)               |                     | 5         | 0.5%  | 0         |       |

| <b>RETINA CONTINUED</b>                                   | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 120.960 retinopathy                                       | 1 0.1%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 0                | 2 1.6%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 5 0.5%           | 0                |
| 900.100 other, not inherited                              | 18 1.7%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 2 0.2%           | 7 5.6%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 659 62.8%        | 50 40.0%         |

# **OCULAR DISORDERS REPORT ARGENTINE DOGO**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the ARGENTINE DOGO breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT ARGENTINE DOGO

| Diagnostic Name | TOTAL DOGS EXAMINED                                  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 1         | 0.8%  | 0         |       |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.700          | corneal dystrophy                                    | 1         | 0.8%  | 2         | 9.5%  |
| 70.730          | corneal endothelial degeneration                     | 1         | 0.8%  | 0         |       |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris         | 14        | 10.9% | 0         |       |
| 93.720          | persistent pupillary membranes, iris to lens         | 1         | 0.8%  | 0         |       |
| <b>LENS</b>     |  |           |       |           |       |
| 100.200         | cataract, unspecified                                | 1         | 0.8%  | 0         |       |
| 100.210         | cataract. suspect not inherited/significance unknown | 1         | 0.8%  | 0         |       |
| 100.302         | punctate cataract, posterior cortex                  | 1         | 0.8%  | 0         |       |
| 100.312         | incipient cataract, posterior cortex                 | 2         | 1.6%  | 1         | 4.8%  |
| 100.316         | incipient cataract, nucleus                          | 2         | 1.6%  | 0         |       |
| 100.330         | generalized/complete cataract                        | 1         | 0.8%  | 0         |       |
| 100.345         | <i>significant cataracts (summary)</i>               | 7         | 5.4%  | 1         | 4.8%  |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.120         | persistent hyaloid artery/remnant                    | 1         | 0.8%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.100         | other, not inherited                                 | 1         | 0.8%  | 0         |       |
| 900.110         | other. suspect not inherited/significance unknown    | 1         | 0.8%  | 0         |       |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 109       | 84.5% | 18        | 85.7% |

# AUSTRALIAN CATTLE DOG

(Queensland Heeler or Blue Heeler)

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|--|---------------------|-----------|-----------------|--------------------------------------|
| A. | Glaucoma   | Not defined         | 2         | NO              |                                      |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option  |                                      |
| C. | Cataract   | Not defined         | 1         | NO              |                                      |
| D. | Y-suture tip opacity                             | Not defined         | 1         | Breeder option  |                                      |
| E. | Lens luxation                                    | Autosomal recessive | 3, 4      | NO              | Mutation in the <i>ADAMTS17</i> gene |
| F. | Retinal atrophy ( <i>prcd</i> )                  | Autosomal recessive | 5, 6      | NO              | Mutation in the <i>prcd</i> gene     |
| G. | Retinal atrophy - rod-cone dysplasia type 4      | Autosomal recessive | 7         | NO              | Mutation in the <i>C2orf71</i> gene  |

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## Description and Comments

### A. Glaucoma

An elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy).

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

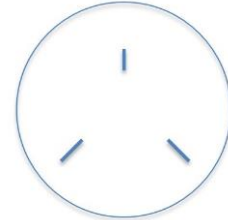
### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane,

persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

E. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

F. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that one form of PRA in the Australian Cattle Dog is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. However in the Australian Cattle Dog the

phenotype can be very variable in the age of onset. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

Other forms of retinal degeneration that are not PRCD are recognized in the breed. The currently available genetic test will not detect these other forms of PRA.

G. Retinal atrophy - rod-cone dysplasia, type 4 (*rcd4*)

A form of PRA identified also in the Australian Cattle Dog breed. Clinical night blindness is observed on average as late as 10 years of age and progresses to total blindness. This form of PRA has been referred to as late-onset PRA (LOPRA). The disorder is caused by a mutation present in the *C2orf71* gene. A DNA test is available. The test is accurate only for this mutation and is of no value in identifying other forms of PRA.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gelatt KN, MacKay EO. Prevalence of the breed-related glaucomas in pure-bred dogs in North America. *Vet Ophthalmol*. 2004;7:97-111.
3. Gould D, Pettitt L, McLaughlin B, et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol*. 2011;14:378-384.
4. Farias FH, Johnson GS, Taylor JF, et al. An ADAMTS17 splice donor site mutation in dogs with primary lens luxation. *Invest Ophthalmol Vis Sci*. 2010;51:4716-4721.
5. Dekomien G, Epplen JT. Exclusion of the PDE6A gene for generalised progressive retinal atrophy in 11 breeds of dog. *Anim Genet*. 2000;31:135-139.
6. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006;88:551-563. PMID: 16938425
7. Downs LM, Bell JS, Freeman J, et al. Late-onset progressive retinal atrophy in the Gordon and Irish Setter breeds is associated with a frameshift mutation in *C2orf71*. *Anim Genet*. 2012;44:169-177.

# OCULAR DISORDERS REPORT AUSTRALIAN CATTLE DOG

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 0.110 microphthalmia  |                     | 0         |      | 1         | 0.2% |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 22.000 ectropion, unspecified   |                     | 1         | 0.0% | 0         |      |
| 25.110 distichiasis   |                     | 15        | 0.3% | 2         | 0.4% |
| <b>NASOLACRIMAL</b>   |                     |           |      |           |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 1         | 0.0% | 1         | 0.2% |
| <b>NICTITANS</b>  |                     |           |      |           |      |
| 50.210 pannus of third eyelid   |                     | 2         | 0.0% | 0         |      |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.210 corneal pannus   |                     | 2         | 0.0% | 0         |      |
| 70.700 corneal dystrophy  |                     | 23        | 0.5% | 5         | 0.9% |
| 70.730 corneal endothelial degeneration                               |                     | 4         | 0.1% | 0         |      |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 42        | 0.9% | 6         | 1.1% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 2         | 0.0% | 0         |      |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 3         | 0.1% | 0         |      |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 6         | 0.1% | 0         |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 0         |      | 3         | 0.5% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 2         | 0.0% | 0         |      |
| 93.999 uveal cysts  |                     | 11        | 0.2% | 4         | 0.7% |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.200 cataract, unspecified   |                     | 35        | 0.8% | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 283       | 6.1% | 30        | 5.5% |
| 100.301 punctate cataract, anterior cortex                            |                     | 38        | 0.8% | 6         | 1.1% |
| 100.302 punctate cataract, posterior cortex                           |                     | 35        | 0.8% | 5         | 0.9% |
| 100.303 punctate cataract, equatorial cortex                          |                     | 19        | 0.4% | 1         | 0.2% |
| 100.304 punctate cataract, anterior sutures                           |                     | 3         | 0.1% | 0         |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 14        | 0.3% | 4         | 0.7% |
| 100.306 punctate cataract, nucleus                                    |                     | 4         | 0.1% | 0         |      |
| 100.307 punctate cataract, capsular                                   |                     | 4         | 0.1% | 0         |      |
| 100.311 incipient cataract, anterior cortex                           |                     | 45        | 1.0% | 10        | 1.8% |
| 100.312 incipient cataract, posterior cortex                          |                     | 70        | 1.5% | 2         | 0.4% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 51        | 1.1% | 5         | 0.9% |
| 100.314 incipient cataract, anterior sutures                          |                     | 2         | 0.0% | 4         | 0.7% |
| 100.315 incipient cataract, posterior sutures                         |                     | 18        | 0.4% | 2         | 0.4% |
| 100.316 incipient cataract, nucleus                                   |                     | 4         | 0.1% | 3         | 0.5% |
| 100.317 incipient cataract, capsular                                  |                     | 4         | 0.1% | 3         | 0.5% |
| 100.321 incomplete cataract, anterior cortex                          |                     | 0         |      | 3         | 0.5% |
| 100.322 incomplete cataract, posterior cortex                         |                     | 0         |      | 3         | 0.5% |
| 100.323 incomplete cataract, equatorial cortex                        |                     | 0         |      | 2         | 0.4% |
| 100.326 incomplete cataract, nucleus                                  |                     | 1         | 0.0% | 3         | 0.5% |
| 100.327 incomplete cataract, capsular                                 |                     | 0         |      | 2         | 0.4% |
| 100.328 y-suture tip opacities  |                     | 1         | 0.0% | 5         | 0.9% |



| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.330 generalized/complete cataract                     | 22 0.5%          | 2 0.4%           |
| 100.340 resorbing/hypermature cataract                    | 0                | 1 0.2%           |
| 100.345 <i>significant cataracts (summary)</i>            | 370 8.0%         | 66 12.0%         |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 3 0.1%           | 1 0.2%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 8 0.2%           | 0                |
| 110.135 PHPV/PTVL   | 1 0.0%           | 0                |
| 110.320 vitreal degeneration                              | 13 0.3%          | 2 0.4%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 3 0.1%           | 0                |
| 97.120 coloboma   | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 37 0.8%          | 1 0.2%           |
| 120.180 retinal dysplasia, geographic                     | 12 0.3%          | 3 0.5%           |
| 120.190 retinal dysplasia, detached                       | 1 0.0%           | 1 0.2%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 251 5.4%         | 6 1.1%           |
| 120.400 retinal hemorrhage                                | 1 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 3 0.1%           | 0                |
| 120.920 retinal detachment with dialysis                  | 0                | 2 0.4%           |
| 120.960 retinopathy                                       | 1 0.0%           | 7 1.3%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.120 optic nerve hypoplasia                            | 2 0.0%           | 0                |
| 130.150 optic disc coloboma                               | 1 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 20 0.4%          | 0                |
| 900.100 other, not inherited                              | 126 2.7%         | 1 0.2%           |
| 900.110 other. suspect not inherited/significance unknown | 32 0.7%          | 45 8.2%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 3,811 82.4%      | 415 75.6%        |

# AUSTRALIAN KELPIE

| DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE    |
|--|---------------------|-----------|-----------------|----------------------------|
| A. Cataract  | Not defined         | 1         | NO              |                            |
| B. Choroidal hypoplasia (Collie Eye Anomaly)<br>- optic nerve coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- staphyloma/ coloboma | Autosomal recessive | 2         | NO              | Mutation in the NHEJ1 gene |

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## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### B. Choroidal hypoplasia (Collie Eye Anomaly)

- staphyloma/coloboma
- retinal detachment
- retinal hemorrhage
- optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Kucharczyk, N., et al. (2019). "Collie Eye Anomaly in Australian Kelpie dogs in Poland." BMC Vet Res 15(1): 392. PMID: 31684941.

# OCULAR DISORDERS REPORT AUSTRALIAN KELPIE

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.700          | corneal dystrophy  | 1         | 0.4%  | 0         |       |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 1         | 0.4%  | 0         |       |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 0         |       | 1         | 5.0%  |
| 93.810          | uveal melanoma   | 3         | 1.3%  | 0         |       |
| <b>LENS</b>     |  |           |       |           |       |
| 100.200         | cataract, unspecified  | 5         | 2.2%  | 0         |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 28        | 12.4% | 5         | 25.0% |
| 100.301         | punctate cataract, anterior cortex                           | 7         | 3.1%  | 0         |       |
| 100.302         | punctate cataract, posterior cortex                          | 8         | 3.6%  | 0         |       |
| 100.306         | punctate cataract, nucleus                                   | 1         | 0.4%  | 0         |       |
| 100.311         | incipient cataract, anterior cortex                          | 9         | 4.0%  | 0         |       |
| 100.312         | incipient cataract, posterior cortex                         | 7         | 3.1%  | 0         |       |
| 100.313         | incipient cataract, equatorial cortex                        | 2         | 0.9%  | 0         |       |
| 100.315         | incipient cataract, posterior sutures                        | 1         | 0.4%  | 0         |       |
| 100.330         | generalized/complete cataract                                | 1         | 0.4%  | 0         |       |
| 100.345         | <i>significant cataracts (summary)</i>                       | 41        | 18.2% | 0         |       |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.320         | vitreal degeneration   | 3         | 1.3%  | 0         |       |
| <b>FUNDUS</b>   |  |           |       |           |       |
| 97.110          | choroidal hypoplasia   | 1         | 0.4%  | 0         |       |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                                     | 5         | 2.2%  | 0         |       |
| 120.310         | generalized progressive retinal atrophy (PRA)                | 11        | 4.9%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.000         | other, unspecified   | 7         | 3.1%  | 0         |       |
| 900.100         | other, not inherited   | 8         | 3.6%  | 0         |       |
| 900.110         | other. suspect not inherited/significance unknown            | 1         | 0.4%  | 1         | 5.0%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 171       | 76.0% | 13        | 65.0% |

# AUSTRALIAN LABRADOODLE

(Labradoodle, Australian Cobber Dog)

|    | DISORDER   | INHERITANCE                        | REFERENCE | BREEDING ADVICE                                   | GENETIC TESTS AVAILABLE               |
|----|--|------------------------------------|-----------|---|---------------------------------------|
| A. | Entropion  | Not defined                        | 1-3       | Breeder option                                    |                                       |
| B. | Ectropion  | Not defined                        | 1         | Breeder option                                    |                                       |
| C. | Distichiasis   | Not defined                        | 1         | Breeder option                                    |                                       |
| D. | Corneal dystrophy<br>- epithelial/stromal  | Not defined                        | 1, 4      | Breeder option                                    |                                       |
| E. | Uveal cysts  | Not defined                        | 5         | Breeder option                                    |                                       |
| F. | Persistent pupillary<br>membranes<br>- iris to iris<br>- lens pigment foci no<br>strands | Not defined<br>Not defined         | 1<br>1    | Breeder option<br>Passes with no<br>notation      |                                       |
| G. | Cataract   | Not defined                        | 1         | NO  |                                       |
| H. | Y-suture tip opacity   | Not defined                        | 1         | Breeder option                                    |                                       |
| I. | Persistent hyaloid<br>artery   | Not defined                        | 1         | Breeder option                                    |                                       |
| J. | Vitreous degeneration  | Not defined                        | 1         | Breeder option                                    |                                       |
| K. | Retinal atrophy ( <i>prcd</i> )  | Autosomal<br>recessive             | 1, 5-9    | NO  | Mutation of the<br><i>prcd</i> gene   |
| L. | Achromatopsia Type<br>1 (ACHM – Type 1)  | Autosomal<br>recessive             | 10        | NO  | Deletion in the<br><i>CNGA3</i> gene  |
| M. | Retinal dysplasia<br>- folds   | Presumed<br>autosomal<br>recessive | 1, 11-20  | NO<br>(Breeder option<br>with Normal DNA<br>test) | Mutation of the<br><i>COL9A3</i> gene |
| N. | Retinal dysplasia<br>- geographic/<br>detached (without<br>skeletal defects)             | Presumed<br>autosomal<br>recessive | 1, 11-21  | NO  |                                       |

|    | <b>DISORDER</b>   | <b>INHERITANCE</b>   | <b>REFERENCE</b> | <b>BREEDING<br/>ADVICE</b> | <b>GENETIC TESTS<br/>AVAILABLE</b>    |
|----|---|--|------------------|----------------------------|---------------------------------------|
| O. | Retinal dysplasia<br>- folds/geographic/<br>detached (with<br>skeletal defects) | Autosomal<br>recessive<br>with incomplete<br>dominance for<br>the eyes | 1, 11-21         | NO                         | Mutation of the<br><i>COL9A3</i> gene |
| P. | Limbal melanoma   | Not defined  | 22               | NO                         |                                       |

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## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. Selection should be directed against entropion and toward a head conformation that reduces or eliminates the likelihood of the defect.

### B. Ectropion

A conformational defect resulting in eversion of the eyelid(s), which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### C. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### D. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral. In Labrador Retrievers in Europe, one form of corneal dystrophy has been shown to be caused by accumulations of glycosaminoglycans in the corneal stroma. This form of corneal dystrophy is caused by a mutation in the *CHST6* gene.

E. Uveal cysts

Fluid filled sacs arising from the posterior surface of the iris, to which they may remain attached or break free and float into the anterior chamber. Usually occur in mature dogs.

This disorder may be observed in any breed but retriever breeds tend to be predisposed. There is usually no effect on vision unless the cysts are heavily clustered and impinge on the pupillary area. Less frequently, the cysts may rupture and adhere to the cornea or anterior lens capsule. Multiple cysts may occlude the iridocorneal angle and cause glaucoma.

F. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

G. Cataract

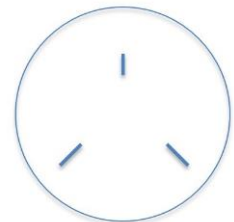
A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

The most frequently reported cataracts in the Labradoodle (Australian) are bilateral or unilateral, focal, posterior polar (posterior cortical)/subcapsular cataracts, which usually present between 1-3 years of age. These are generally stationary or very slowly progressive and generally do not interfere with vision. It has been suggested that these cataracts are inherited as dominant with incomplete penetrance, but definitive breeding studies are still required to verify this hypothesis.

A second type of cataract is a progressive cortical cataract which may involve the entire lens. It is not clear whether this is a distinct entity, or an aberrant form of the posterior polar cataract.

H. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-



diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.

These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

I. Persistent hyaloid artery (PHA)

A congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

J. Vitreous degeneration

Liquefaction of the vitreous gel, which may predispose to retinal detachment.

K. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Labradoodle is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

L. Achromatopsia Type 2 (ACHM – Type 2)

A congenital form of day blindness. Visual deficits become apparent between 8-10 weeks of age. Normal vision is present in low light conditions. Clinical examination is normal. Cone responses are absent on an electroretinogram. The causative genetic mutation of the *CNGA3* gene (3nt deletion in exon 7). A DNA test is available.

M. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its



significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness.

In the Labrador Retriever, the presence of retinal folds may be seen in the heterozygous state described in “R” below, thus the recommendation against breeding.

The breeding advice for Labrador Retrievers and Samoyeds diagnosed with "retinal dysplasia - folds" will be changed from "No" to "Breeder option" if the owner of the dog provides the registering office with results of the DNA test for the affected dog, showing that it is not a carrier of the *COL9A3* mutation.

N. Retinal dysplasia - geographic, detached without skeletal defects

Abnormal development of the retina present at birth; however, in the Golden Retriever, Labrador Retriever, and German Shepherd Dog it has been demonstrated that the geographic form of retinal dysplasia may not be apparent before dogs are 10 weeks of age.

**Retinal dysplasia - geographic:** Any irregularly shaped area of abnormal retinal development containing both areas of thinning and areas of elevation representing folds and retinal disorganization.

**Retinal dysplasia - detached:** Severe retinal disorganization associated with separation (detachment) of the retina.

These two forms are associated with vision impairment or blindness. Retinal dysplasia is known to be inherited in many breeds. The genetic relationship between the three forms of retinal dysplasia is not known for all breeds

In Europe, this condition has been documented as an autosomal recessive condition and results in early retinal detachment and blindness. Lens and corneal opacities can also be present, but skeletal abnormalities (see below) are not present. The condition of generalized retinal dysplasia with retinal detachment but without skeletal abnormalities has been reported primarily in Europe, and is rarely if ever seen in the United States.

In the United States, the milder forms of retinal dysplasia (folds/geographic) are seen in Labradors. These may represent the heterozygous form of the condition in which the homozygote also displays skeletal malformations (see “R” below) or it may represent a genetically distinct entity with an undetermined mode of inheritance. It is not possible clinically to make this distinction. Thus, Labradors with any form of retinal dysplasia should not be used for breeding.

O. Retinal dysplasia - folds or detachment with skeletal defects

This condition is also known as oculo-skeletal dysplasia (OSD) or dwarfism with retinal dysplasia type 1 (DRD1) in the Labrador Retriever. A similar condition, DRD2, occurs in the Samoyed. The condition is autosomal recessive and homozygous affected dogs have shortened forelimbs (“downhill” conformation) with valgus deformity. They have severe ocular defects including cataract, retinal folds/multifocal retinal dysplasia, vitreal degeneration and retinal detachment. The ocular abnormalities result in blindness in most dogs. Heterozygous dogs can have either a normal ocular exam or have multiple retinal folds, vitreal membranes, or vitreal

degeneration suggesting a semi-dominant mechanism with respect to the eyes. It is important to note that generally the retinal folds present in heterozygous dogs tend to cluster around the major superior blood vessels of the central tapetal region. The condition is caused by a 1 base pair insertion of *COL9A3*. A DNA test is available.

P. Limbal melanoma

Most limbal melanomas are really epibulbar melanocytomas, but there is a possibility of an extension of an intraocular melanoma extending outward and presenting as a limbal melanoma. An epibulbar melanocytoma originates from the superficial pigment lining the limbus and the lesion may eventually extend into the eye. Metastasis has not been documented and the mass is characterized by large epithelioid cells. The lesion presents as a subconjunctival smooth mass most commonly in the dorsolateral limbal region and extends later into the cornea and posterior on the sclera. Breed predisposition has been noted in the German Shepherd, Labrador and Golden Retriever.

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# OCULAR DISORDERS REPORT AUSTRALIAN LABRADOODLE

| Diagnostic Name     | TOTAL DOGS EXAMINED  |     | 1991-2015<br>4,250 |     | 2016-2020<br>7,487 |   |
|---------------------|--|-----|--------------------|-----|--------------------|---|
|                     | #  | %   | #                  | %   | #                  | % |
| <b>EYELIDS</b>      |  |     |                    |     |                    |   |
| 21.000              | entropion, unspecified   | 1   | 0.0%               | 0   |                    |   |
| 25.110              | distichiasis   | 41  | 1.0%               | 165 | 2.2%               |   |
| <b>NASOLACRIMAL</b> |  |     |                    |     |                    |   |
| 32.110              | imperforate lower nasolacrimal punctum                         | 1   | 0.0%               | 15  | 0.2%               |   |
| 40.910              | keratoconjunctivitis sicca                                     | 0   |                    | 2   | 0.0%               |   |
| <b>NICTITANS</b>    |  |     |                    |     |                    |   |
| 51.100              | third eyelid cartilage anomaly                                 | 1   | 0.0%               | 3   | 0.0%               |   |
| <b>CORNEA</b>       |  |     |                    |     |                    |   |
| 70.210              | corneal pannus   | 1   | 0.0%               | 1   | 0.0%               |   |
| 70.700              | corneal dystrophy  | 40  | 0.9%               | 130 | 1.7%               |   |
| <b>UVEA</b>         |  |     |                    |     |                    |   |
| 93.110              | iris hypoplasia  | 0   |                    | 1   | 0.0%               |   |
| 93.150              | iris coloboma  | 0   |                    | 1   | 0.0%               |   |
| 93.710              | persistent pupillary membranes, iris to iris                   | 163 | 3.8%               | 521 | 7.0%               |   |
| 93.720              | persistent pupillary membranes, iris to lens                   | 4   | 0.1%               | 5   | 0.1%               |   |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 1   | 0.0%               | 1   | 0.0%               |   |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 71  | 1.7%               | 263 | 3.5%               |   |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 0   |                    | 3   | 0.0%               |   |
| 93.810              | uveal melanoma   | 1   | 0.0%               | 0   |                    |   |
| 97.150              | chorioretinal coloboma, congenital                             | 1   | 0.0%               | 0   |                    |   |
| <b>LENS</b>         |  |     |                    |     |                    |   |
| 100.210             | cataract. suspect not inherited/significance unknown           | 102 | 2.4%               | 209 | 2.8%               |   |
| 100.301             | punctate cataract, anterior cortex                             | 10  | 0.2%               | 23  | 0.3%               |   |
| 100.302             | punctate cataract, posterior cortex                            | 1   | 0.0%               | 11  | 0.1%               |   |
| 100.303             | punctate cataract, equatorial cortex                           | 1   | 0.0%               | 4   | 0.1%               |   |
| 100.304             | punctate cataract, anterior sutures                            | 1   | 0.0%               | 1   | 0.0%               |   |
| 100.305             | punctate cataract, posterior sutures                           | 13  | 0.3%               | 30  | 0.4%               |   |
| 100.306             | punctate cataract, nucleus                                     | 4   | 0.1%               | 6   | 0.1%               |   |
| 100.307             | punctate cataract, capsular                                    | 1   | 0.0%               | 24  | 0.3%               |   |
| 100.311             | incipient cataract, anterior cortex                            | 6   | 0.1%               | 13  | 0.2%               |   |
| 100.312             | incipient cataract, posterior cortex                           | 1   | 0.0%               | 14  | 0.2%               |   |
| 100.313             | incipient cataract, equatorial cortex                          | 2   | 0.0%               | 5   | 0.1%               |   |
| 100.314             | incipient cataract, anterior sutures                           | 0   |                    | 2   | 0.0%               |   |
| 100.315             | incipient cataract, posterior sutures                          | 0   |                    | 4   | 0.1%               |   |
| 100.316             | incipient cataract, nucleus                                    | 1   | 0.0%               | 6   | 0.1%               |   |
| 100.317             | incipient cataract, capsular                                   | 1   | 0.0%               | 6   | 0.1%               |   |
| 100.321             | incomplete cataract, anterior cortex                           | 1   | 0.0%               | 5   | 0.1%               |   |
| 100.322             | incomplete cataract, posterior cortex                          | 0   |                    | 6   | 0.1%               |   |
| 100.323             | incomplete cataract, equatorial cortex                         | 2   | 0.0%               | 4   | 0.1%               |   |
| 100.325             | incomplete cataract, posterior sutures                         | 2   | 0.0%               | 1   | 0.0%               |   |
| 100.326             | incomplete cataract, nucleus                                   | 1   | 0.0%               | 5   | 0.1%               |   |
| 100.327             | incomplete cataract, capsular                                  | 0   |                    | 3   | 0.0%               |   |
| 100.328             | y-suture tip opacities   | 17  | 0.4%               | 61  | 0.8%               |   |
| 100.330             | generalized/complete cataract                                  | 0   |                    | 2   | 0.0%               |   |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.345 significant cataracts (summary)                   | 65 1.5%          | 236 3.2%         |
| 100.375 subluxation/luxation, unspecified                 | 0                | 1 0.0%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 6 0.1%           | 20 0.3%          |
| 110.135 PHPV/PTVL   | 2 0.0%           | 1 0.0%           |
| 110.200 vitreous degeneration-anterior chamber            | 0                | 4 0.1%           |
| 110.320 vitreal degeneration                              | 3 0.1%           | 9 0.1%           |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 31 0.7%          | 49 0.7%          |
| 120.180 retinal dysplasia, geographic                     | 0                | 2 0.0%           |
| 120.960 retinopathy                                       | 4 0.1%           | 1 0.0%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 10 0.2%          | 8 0.1%           |
| 130.120 optic nerve hypoplasia                            | 0                | 2 0.0%           |
| 130.150 optic disc coloboma                               | 0                | 3 0.0%           |
| <b>OTHER</b>  |                  |                  |
| 900.100 other, not inherited                              | 1 0.0%           | 9 0.1%           |
| 900.110 other. suspect not inherited/significance unknown | 85 2.0%          | 319 4.3%         |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,826 43.0%      | 5,835 77.9%      |

## AUSTRALIAN SHEPHERD

**It is recommended that this breed be examined prior to pharmacological dilation to best facilitate identification of iris coloboma.**

|    | <b>DISORDER</b>                               | <b>INHERITANCE</b>                                      | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> | <b>GENETIC TESTS AVAILABLE</b>    |
|----|---|---|------------------|------------------------|-----------------------------------|
| A. | Microphthalmia with multiple ocular defects   | Presumed autosomal recessive with incomplete penetrance | 1-6              | NO                     |                                   |
| B. | Distichiasis                                  | Not defined   | 1                | Breeder option         |                                   |
| C. | Corneal dystrophy - epithelial/stromal        | Not defined   | 1                | Breeder option         |                                   |
| D. | Iris coloboma                                 | Not defined   | 1                | NO                     |                                   |
| E. | Iris hypoplasia                               | Not defined   | 1                | Breeder option         |                                   |
| F. | Persistent pupillary membranes - iris to iris | Not defined   | 1                | Breeder option         |                                   |
| G. | Cataract                                      | Autosomal co-dominant                                   | 1, 7,8           | NO                     | Mutation in the <i>HSF4</i> gene  |
| H. | Y-suture tip opacity                          | Not defined   | 1                | Breeder option         |                                   |
| I. | Vitreous degeneration                         | Not defined   | 1                | Breeder option         |                                   |
| J. | Persistent hyaloid artery                     | Not defined   | 1                | Breeder option         |                                   |
| K. | Retinal atrophy ( <i>prcd</i> )               | Autosomal recessive                                     | 1, 9, 10         | NO                     | Mutation in the <i>prcd</i> gene  |
| L. | Cone degeneration - day blindness             | Autosomal recessive                                     | 16               | NO                     | Mutation in the <i>CNGB3</i> gene |
| M. | Multifocal retinopathy - <i>cmr1</i>          | Autosomal recessive                                     | 17               | Breeder option         | Mutation in the <i>BEST1</i> gene |
| N. | Retinal dysplasia - folds                     | Not defined   | 1                | Breeder option         |                                   |

|    | <b>DISORDER</b>  | <b>INHERITANCE</b>  | <b>REFERENCE</b> | <b>BREEDING<br/>ADVICE</b> | <b>GENETIC TESTS<br/>AVAILABLE</b> |
|----|--|---------------------|------------------|----------------------------|------------------------------------|
| O. | Choroidal hypoplasia (Collie Eye Anomaly)<br>- optic nerve coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- staphyloma/coloboma | Autosomal recessive | 1, 12-15         | NO                         | Mutation in the <i>NHEJ1</i> gene  |
| P. | Coloboma/staphyloma without microphthalmia   | Not defined         | 1                | NO                         |                                    |
| Q. | Micropapilla   | Not defined         | 1                | Breeder option             |                                    |

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## Description and Comments

### A. Microphthalmia with multiple ocular defects

Microphthalmia is a congenital defect characterized by a small eye with associated defects of the cornea, iris (coloboma), anterior chamber, lens (cataract) and/or retina (dysplasia). In the Australian Shepherd, microphthalmia has long been suspected to be associated with merle coat coloration but a definitive genetic relationship has not been established. The eyes of affected homozygous merle (usually white) dogs have extreme forms of this entity and are usually blind at birth. Affected heterozygous merle-coated dogs demonstrate less severe manifestations.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### C. Corneal Dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

D. Iris coloboma

A congenital abnormality in iris development usually characterized by a full-thickness defect in iris tissue, commonly (though not exclusively) located at the 6 o'clock position associated with failure of closure of the optic fissure. A partial-thickness defect in iris tissue should be recorded as iris hypoplasia on the OFA form.

E. Iris hypoplasia

A congenital abnormality in iris development usually characterized by a reduced quantity of tissue identified as a partial-thickness defect in iris tissue. Full-thickness iris hypoplasia is rare and should be recorded as an iris coloboma on the OFA form.

F. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

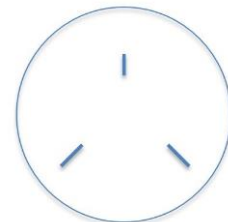
G. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Australian Shepherd, a mutation in *HSF4* (heat shock transcription factor 4), the *HSF4-2* mutation, has been shown to increase the likelihood of cataract formation. The mutation is inherited in a co-dominant manner. Dogs with one copy of the mutation develop bilateral posterior cataracts and homozygotes develop a nuclear cataract that typically progresses to a mature cataract. A DNA test is available for this mutation. Other genetic factors can contribute to cataract formation in this breed and will not be detected by this test.

H. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress





(unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.

These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

I. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

J. Persistent hyaloid artery (PHA)

Congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small patent vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

K. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Australian Shepherd is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

L. Cone degeneration - day blindness or hemeralopia

Autosomal recessively inherited early degeneration of the cone photoreceptors. Affected puppies develop day-blindness, color blindness, and photophobia between 8 and 12 weeks of age. Affected dogs remain ophthalmoscopically normal their entire life. Electroretinography is required to definitively diagnose the disorder. Genetically, the condition results from a mutation in the *CNGB3* gene. A DNA test is available.

M. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (*cmr1*) is characterized by numerous distinct (i.e. multi-focal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff.

N. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

O. Choroidal hypoplasia (Collie Eye Anomaly)

- staphyloma/coloboma
- retinal detachment
- retinal hemorrhage
- optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

P. Coloboma/staphyloma (unassociated with microphthalmia)

A coloboma is a congenital defect which may affect the iris, choroid or optic disc. Iris colobomas are seen as notches in the pupillary margin. Scleral ectasia is defined as a

congenital thinning and secondary distention of the sclera; when lined by uveal tissue it is called a staphyloma. These may be anteriorly located, apparent as a bulge beneath the upper eyelid or posteriorly located, requiring visualization with an ophthalmoscope. These conditions may or may not be genetically related to the same anomalies seen associated with microphthalmia (entity "A" above).

Q. Micropapilla

Micropapilla refers to a small optic disc which is not associated with vision impairment.

Optic nerve hypoplasia refers to a congenital defect of the optic nerve which causes blindness and abnormal pupil response in the affected eye. It may be difficult to differentiate between micropapilla and optic nerve hypoplasia on a routine (dilated) screening ophthalmoscopic exam.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gelatt KN, McGill LD. Clinical characteristics of microphthalmia with colobomas of the Australian Shepherd Dog. *J Am Vet Med Assoc.* 1973;162:393-396.
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# OCULAR DISORDERS REPORT AUSTRALIAN SHEPHERD

| Diagnostic Name     | TOTAL DOGS EXAMINED  |  | 1991-2015<br>96,323 |      | 2016-2020<br>22,420 |      |
|---------------------|--|--|---------------------|------|---------------------|------|
|                     |  |  | #                   | %    | #                   | %    |
| <b>GLOBE</b>        |  |  |                     |      |                     |      |
| 0.110               | microphthalmia   |  | 92                  | 0.1% | 15                  | 0.1% |
| 10.000              | glaucoma   |  | 8                   | 0.0% | 0                   |      |
| <b>EYELIDS</b>      |  |  |                     |      |                     |      |
| 20.110              | eyelid dermoid   |  | 1                   | 0.0% | 0                   |      |
| 20.140              | ectopic cilia  |  | 5                   | 0.0% | 0                   |      |
| 20.160              | macropalpebral fissure   |  | 4                   | 0.0% | 0                   |      |
| 21.000              | entropion, unspecified   |  | 15                  | 0.0% | 1                   | 0.0% |
| 22.000              | ectropion, unspecified   |  | 6                   | 0.0% | 0                   |      |
| 25.110              | distichiasis   |  | 1,578               | 1.6% | 310                 | 1.4% |
| <b>NASOLACRIMAL</b> |  |  |                     |      |                     |      |
| 32.110              | imperforate lower nasolacrimal punctum                         |  | 5                   | 0.0% | 7                   | 0.0% |
| 40.910              | keratoconjunctivitis sicca                                     |  | 1                   | 0.0% | 0                   |      |
| <b>NICTITANS</b>    |  |  |                     |      |                     |      |
| 50.210              | pannus of third eyelid   |  | 0                   |      | 1                   | 0.0% |
| 51.100              | third eyelid cartilage anomaly                                 |  | 4                   | 0.0% | 0                   |      |
| 52.110              | prolapsed gland of the third eyelid                            |  | 2                   | 0.0% | 1                   | 0.0% |
| <b>CORNEA</b>       |  |  |                     |      |                     |      |
| 70.210              | corneal pannus   |  | 8                   | 0.0% | 1                   | 0.0% |
| 70.220              | pigmentary keratitis   |  | 1                   | 0.0% | 1                   | 0.0% |
| 70.700              | corneal dystrophy  |  | 432                 | 0.4% | 163                 | 0.7% |
| 70.730              | corneal endothelial degeneration                               |  | 14                  | 0.0% | 2                   | 0.0% |
| <b>UVEA</b>         |  |  |                     |      |                     |      |
| 93.110              | iris hypoplasia  |  | 208                 | 0.2% | 144                 | 0.6% |
| 93.140              | corneal endothelial pigment without PPM                        |  | 1                   | 0.0% | 0                   |      |
| 93.150              | iris coloboma  |  | 1,430               | 1.5% | 215                 | 1.0% |
| 93.180              | iris sphincter dysplasia                                       |  | 8                   | 0.0% | 21                  | 0.1% |
| 93.710              | persistent pupillary membranes, iris to iris                   |  | 4,597               | 4.8% | 1,561               | 7.0% |
| 93.720              | persistent pupillary membranes, iris to lens                   |  | 87                  | 0.1% | 16                  | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea                 |  | 43                  | 0.0% | 7                   | 0.0% |
| 93.740              | persistent pupillary membranes, iris sheets                    |  | 92                  | 0.1% | 0                   |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   |  | 25                  | 0.0% | 23                  | 0.1% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands |  | 22                  | 0.0% | 6                   | 0.0% |
| 93.810              | uveal melanoma   |  | 8                   | 0.0% | 1                   | 0.0% |
| 93.999              | uveal cysts  |  | 39                  | 0.0% | 7                   | 0.0% |
| 97.150              | chorioretinal coloboma, congenital                             |  | 16                  | 0.0% | 11                  | 0.0% |
| <b>LENS</b>         |  |  |                     |      |                     |      |
| 100.200             | cataract, unspecified  |  | 169                 | 0.2% | 0                   |      |
| 100.210             | cataract. suspect not inherited/significance unknown           |  | 2,311               | 2.4% | 442                 | 2.0% |
| 100.301             | punctate cataract, anterior cortex                             |  | 221                 | 0.2% | 37                  | 0.2% |
| 100.302             | punctate cataract, posterior cortex                            |  | 318                 | 0.3% | 41                  | 0.2% |
| 100.303             | punctate cataract, equatorial cortex                           |  | 83                  | 0.1% | 12                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            |  | 30                  | 0.0% | 4                   | 0.0% |
| 100.305             | punctate cataract, posterior sutures                           |  | 200                 | 0.2% | 68                  | 0.3% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |      | <b>2016-2020</b> |      |
|---|------------------|------|------------------|------|
| 100.306 punctate cataract, nucleus                        | 149              | 0.2% | 36               | 0.2% |
| 100.307 punctate cataract, capsular                       | 81               | 0.1% | 35               | 0.2% |
| 100.311 incipient cataract, anterior cortex               | 302              | 0.3% | 46               | 0.2% |
| 100.312 incipient cataract, posterior cortex              | 742              | 0.8% | 72               | 0.3% |
| 100.313 incipient cataract, equatorial cortex             | 189              | 0.2% | 21               | 0.1% |
| 100.314 incipient cataract, anterior sutures              | 24               | 0.0% | 2                | 0.0% |
| 100.315 incipient cataract, posterior sutures             | 151              | 0.2% | 17               | 0.1% |
| 100.316 incipient cataract, nucleus                       | 197              | 0.2% | 23               | 0.1% |
| 100.317 incipient cataract, capsular                      | 109              | 0.1% | 23               | 0.1% |
| 100.321 incomplete cataract, anterior cortex              | 5                | 0.0% | 15               | 0.1% |
| 100.322 incomplete cataract, posterior cortex             | 16               | 0.0% | 27               | 0.1% |
| 100.323 incomplete cataract, equatorial cortex            | 1                | 0.0% | 7                | 0.0% |
| 100.324 incomplete cataract, anterior sutures             | 0                |      | 1                | 0.0% |
| 100.325 incomplete cataract, posterior sutures            | 1                | 0.0% | 4                | 0.0% |
| 100.326 incomplete cataract, nucleus                      | 6                | 0.0% | 5                | 0.0% |
| 100.327 incomplete cataract, capsular                     | 1                | 0.0% | 3                | 0.0% |
| 100.328 y-suture tip opacities                            | 31               | 0.0% | 67               | 0.3% |
| 100.330 generalized/complete cataract                     | 230              | 0.2% | 10               | 0.0% |
| 100.345 <i>significant cataracts (summary)</i>            | 3,256            | 3.4% | 576              | 2.6% |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 18               | 0.0% | 1                | 0.0% |
| <b>VITREOUS</b>   |                  |      |                  |      |
| 110.120 persistent hyaloid artery/remnant                 | 507              | 0.5% | 121              | 0.5% |
| 110.135 PHPV/PTVL   | 108              | 0.1% | 14               | 0.1% |
| 110.200 vitreous degeneration-anterior chamber            | 4                | 0.0% | 11               | 0.0% |
| 110.320 vitreal degeneration                              | 255              | 0.3% | 47               | 0.2% |
| <b>FUNDUS</b>   |                  |      |                  |      |
| 97.110 choroidal hypoplasia                               | 152              | 0.2% | 34               | 0.2% |
| 97.120 coloboma   | 96               | 0.1% | 0                |      |
| <b>RETINA</b>   |                  |      |                  |      |
| 120.170 retinal dysplasia, folds                          | 924              | 1.0% | 200              | 0.9% |
| 120.180 retinal dysplasia, geographic                     | 45               | 0.0% | 4                | 0.0% |
| 120.190 retinal dysplasia, detached                       | 9                | 0.0% | 3                | 0.0% |
| 120.310 generalized progressive retinal atrophy (PRA)     | 133              | 0.1% | 4                | 0.0% |
| 120.400 retinal hemorrhage                                | 13               | 0.0% | 0                |      |
| 120.910 retinal detachment without dialysis               | 61               | 0.1% | 0                |      |
| 120.920 retinal detachment with dialysis                  | 10               | 0.0% | 8                | 0.0% |
| 120.960 retinopathy                                       | 8                | 0.0% | 10               | 0.0% |
| <b>OPTIC NERVE</b>  |                  |      |                  |      |
| 130.110 micropapilla                                      | 200              | 0.2% | 95               | 0.4% |
| 130.120 optic nerve hypoplasia                            | 114              | 0.1% | 19               | 0.1% |
| 130.150 optic disc coloboma                               | 154              | 0.2% | 23               | 0.1% |
| <b>OTHER</b>  |                  |      |                  |      |
| 900.000 other, unspecified                                | 545              | 0.6% | 0                |      |
| 900.100 other, not inherited                              | 1,262            | 1.3% | 29               | 0.1% |
| 900.110 other. suspect not inherited/significance unknown | 558              | 0.6% | 445              | 2.0% |

|                                     | 1991-2015    | 2016-2020    |
|-------------------------------------|--------------|--------------|
| <b>NORMAL</b><br>0.000 normal globe | 84,363 87.6% | 18,511 82.6% |

# AUSTRALIAN STUMPY TAIL CATTLE DOG

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|------------------------------------|------------------------|-----------|--------------------|-------------------------------------|
| A. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>prcd</i> gene |

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## Description and Comments

### A. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Australian Stumpy Tail Cattle Dog is *prcd*, which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Australian Stumpy Tail Cattle Dog. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006 Nov;88:551-563. PMID: 16938425



# OCULAR DISORDERS REPORT AUSTRALIAN STUMPY TAIL CATTLE DOG

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015 |              | 2016-2020 |        |
|--|---------------------|-----------|--------------|-----------|--------|
|  |                     | #         | %            | #         | %      |
| <b>LENS</b>  |                     |           |              |           |        |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 2         | 4.5%         | 0         |        |
| 100.301 punctate cataract, anterior cortex                   |                     | 1         | 2.3%         | 0         |        |
| 100.305 punctate cataract, posterior sutures                 |                     | 1         | 2.3%         | 0         |        |
| 100.311 incipient cataract, anterior cortex                  |                     | 1         | 2.3%         | 0         |        |
| 100.312 incipient cataract, posterior cortex                 |                     | 2         | 4.5%         | 0         |        |
| 100.313 incipient cataract, equatorial cortex                |                     | 2         | 4.5%         | 0         |        |
| 100.316 incipient cataract, nucleus                          |                     | 1         | 2.3%         | 0         |        |
| <i>100.345 significant cataracts (summary)</i>               |                     | <i>8</i>  | <i>18.2%</i> | <i>0</i>  |        |
| <b>RETINA</b>  |                     |           |              |           |        |
| 120.170 retinal dysplasia, folds                             |                     | 1         | 2.3%         | 0         |        |
| 120.180 retinal dysplasia, geographic                        |                     | 1         | 2.3%         | 0         |        |
| 120.310 generalized progressive retinal atrophy (PRA)        |                     | 3         | 6.8%         | 0         |        |
| <b>OTHER</b>   |                     |           |              |           |        |
| 900.100 other, not inherited                                 |                     | 1         | 2.3%         | 0         |        |
| 900.110 other. suspect not inherited/significance unknown    |                     | 1         | 2.3%         | 0         |        |
| <b>NORMAL</b>  |                     |           |              |           |        |
| 0.000 normal globe   |                     | 38        | 86.4%        | 1         | 100.0% |

# AUSTRALIAN TERRIER

|    | DISORDER   | INHERITANCE                | REFERENCE | BREEDING ADVICE                           |
|----|--|----------------------------|-----------|---|
| A. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined | 1<br>1    | Breeder option<br>Passes with no notation |
| B. | Cataract   | Not defined                | 1         | NO  |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Australian Terrier breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT AUSTRALIAN TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 10.000 glaucoma   |                     | 1         | 0.1% | 0         |      |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 21.000 entropion, unspecified   |                     | 2         | 0.2% | 0         |      |
| 25.110 distichiasis   |                     | 3         | 0.4% | 0         |      |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.220 pigmentary keratitis   |                     | 0         |      | 1         | 0.5% |
| 70.700 corneal dystrophy  |                     | 4         | 0.5% | 1         | 0.5% |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 38        | 4.5% | 18        | 8.8% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 1         | 0.1% | 0         |      |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 3         | 0.4% | 0         |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 5         | 0.6% | 10        | 4.9% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1         | 0.1% | 1         | 0.5% |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.200 cataract, unspecified   |                     | 2         | 0.2% | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 28        | 3.3% | 10        | 4.9% |
| 100.301 punctate cataract, anterior cortex                            |                     | 3         | 0.4% | 0         |      |
| 100.302 punctate cataract, posterior cortex                           |                     | 2         | 0.2% | 0         |      |
| 100.303 punctate cataract, equatorial cortex                          |                     | 2         | 0.2% | 1         | 0.5% |
| 100.305 punctate cataract, posterior sutures                          |                     | 2         | 0.2% | 0         |      |
| 100.306 punctate cataract, nucleus                                    |                     | 2         | 0.2% | 0         |      |
| 100.311 incipient cataract, anterior cortex                           |                     | 5         | 0.6% | 2         | 1.0% |
| 100.312 incipient cataract, posterior cortex                          |                     | 4         | 0.5% | 0         |      |
| 100.313 incipient cataract, equatorial cortex                         |                     | 5         | 0.6% | 0         |      |
| 100.314 incipient cataract, anterior sutures                          |                     | 1         | 0.1% | 0         |      |
| 100.316 incipient cataract, nucleus                                   |                     | 1         | 0.1% | 0         |      |
| 100.317 incipient cataract, capsular                                  |                     | 0         |      | 1         | 0.5% |
| 100.323 incomplete cataract, equatorial cortex                        |                     | 1         | 0.1% | 0         |      |
| 100.326 incomplete cataract, nucleus                                  |                     | 1         | 0.1% | 0         |      |
| 100.330 generalized/complete cataract                                 |                     | 8         | 1.0% | 0         |      |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 39        | 4.6% | 4         | 2.0% |
| 100.375 <i>subluxation/luxation, unspecified</i>                      |                     | 1         | 0.1% | 0         |      |
| <b>VITREOUS</b>   |                     |           |      |           |      |
| 110.320 vitreal degeneration  |                     | 3         | 0.4% | 0         |      |
| <b>RETINA</b>   |                     |           |      |           |      |
| 120.170 retinal dysplasia, folds                                      |                     | 3         | 0.4% | 0         |      |
| 120.310 generalized progressive retinal atrophy (PRA)                 |                     | 3         | 0.4% | 0         |      |
| 120.400 retinal hemorrhage  |                     | 1         | 0.1% | 0         |      |
| <b>OPTIC NERVE</b>  |                     |           |      |           |      |
| 130.110 micropapilla  |                     | 1         | 0.1% | 0         |      |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 4 0.5%    | 0         |
| 900.100 other, not inherited                              | 8 1.0%    | 2 1.0%    |
| 900.110 other. suspect not inherited/significance unknown | 3 0.4%    | 8 3.9%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 737 87.5% | 162 79.0% |

# BARBET

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE         | GENETIC TESTS AVAILABLE          |
|----|--|---------------------|-----------|-------------------------|----------------------------------|
| A. | Distichiasis   | Not defined         | 1         | Breeder option          |                                  |
| B. | Persistent Pupillary Membranes<br>- lens pigment foci no strands | Not defined         | 1         | Passes with no notation |                                  |
| C. | Cataract   | Not defined         | 1         | NO                      |                                  |
| D. | Y suture tip opacity   | Not defined         | 1         | Breeder option          |                                  |
| E. | Retinal atrophy ( <i>prcd</i> )                                  | Autosomal recessive | 2         | NO                      | Mutation in the <i>prcd</i> gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

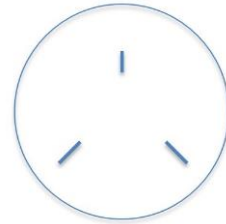
### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume

cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

E. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Barbet is *prcd*, which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006 Nov;88:551-563. PMID: 16938425

# OCULAR DISORDERS REPORT BARBET

| Diagnostic Name    | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|--------------------|--|-----------|-------|-----------|-------|
|                    |  | #         | %     | #         | %     |
| <b>EYELIDS</b>     |  |           |       |           |       |
| 25.110             | distichiasis   | 10        | 6.5%  | 6         | 3.3%  |
| <b>CORNEA</b>      |  |           |       |           |       |
| 70.700             | corneal dystrophy  | 0         |       | 1         | 0.5%  |
| <b>UVEA</b>        |  |           |       |           |       |
| 93.710             | persistent pupillary membranes, iris to iris                 | 4         | 2.6%  | 4         | 2.2%  |
| 93.750             | persistent pupillary membranes, lens pigment foci/no strands | 4         | 2.6%  | 5         | 2.7%  |
| 93.999             | uveal cysts  | 0         |       | 2         | 1.1%  |
| <b>LENS</b>        |  |           |       |           |       |
| 100.210            | cataract. suspect not inherited/significance unknown         | 20        | 12.9% | 18        | 9.8%  |
| 100.301            | punctate cataract, anterior cortex                           | 1         | 0.6%  | 0         |       |
| 100.303            | punctate cataract, equatorial cortex                         | 1         | 0.6%  | 1         | 0.5%  |
| 100.305            | punctate cataract, posterior sutures                         | 0         |       | 1         | 0.5%  |
| 100.307            | punctate cataract, capsular                                  | 0         |       | 1         | 0.5%  |
| 100.311            | incipient cataract, anterior cortex                          | 0         |       | 2         | 1.1%  |
| 100.312            | incipient cataract, posterior cortex                         | 0         |       | 2         | 1.1%  |
| 100.313            | incipient cataract, equatorial cortex                        | 0         |       | 1         | 0.5%  |
| 100.315            | incipient cataract, posterior sutures                        | 0         |       | 1         | 0.5%  |
| 100.317            | incipient cataract, capsular                                 | 0         |       | 1         | 0.5%  |
| 100.328            | y-suture tip opacities                                       | 2         | 1.3%  | 5         | 2.7%  |
| 100.330            | generalized/complete cataract                                | 0         |       | 1         | 0.5%  |
| 100.345            | significant cataracts (summary)                              | 4         | 2.6%  | 16        | 8.7%  |
| <b>VITREOUS</b>    |  |           |       |           |       |
| 110.320            | vitreal degeneration   | 0         |       | 1         | 0.5%  |
| <b>FUNDUS</b>      |  |           |       |           |       |
| 97.110             | choroidal hypoplasia   | 1         | 0.6%  | 0         |       |
| <b>RETINA</b>      |  |           |       |           |       |
| 120.170            | retinal dysplasia, folds                                     | 1         | 0.6%  | 0         |       |
| 120.310            | generalized progressive retinal atrophy (PRA)                | 0         |       | 2         | 1.1%  |
| 120.920            | retinal detachment with dialysis                             | 0         |       | 1         | 0.5%  |
| 120.960            | retinopathy  | 0         |       | 3         | 1.6%  |
| <b>OPTIC NERVE</b> |  |           |       |           |       |
| 130.110            | micropapilla   | 0         |       | 2         | 1.1%  |
| <b>OTHER</b>       |  |           |       |           |       |
| 900.000            | other, unspecified   | 2         | 1.3%  | 0         |       |
| 900.100            | other, not inherited   | 0         |       | 2         | 1.1%  |
| 900.110            | other. suspect not inherited/significance unknown            | 5         | 3.2%  | 7         | 3.8%  |
| <b>NORMAL</b>      |  |           |       |           |       |
| 0.000              | normal globe   | 126       | 81.3% | 135       | 73.8% |



# BASENJI

|    | DISORDER                                  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|---|------------------------|-----------|--------------------|-------------------------------------|
| A. | Corneal dystrophy<br>- epithelial/stromal | Not defined            | 1         | Breeder option     |                                     |
| B. | Corneal dystrophy<br>- endothelial        | Not defined            | 1         | NO                 |                                     |
| C. | Persistent pupillary<br>membranes         |                        |           |                    |                                     |
|    | - iris to iris                            | Not defined            | 1, 2-9    | Breeder option     |                                     |
|    | - iris to cornea                          | Not defined            | 1         | NO                 |                                     |
|    | - iris to lens                            | Not defined            | 1         | NO                 |                                     |
|    | - endothelial<br>opacity/no strands       | Not defined            | 1         | NO                 |                                     |
| D. | Cataract                                  | Not defined            | 1         | NO                 |                                     |
| E. | Y suture tip opacity                      | Not defined            | 1         | Breeder option     |                                     |
| F. | Retinal atrophy                           | Not defined            | 1, 6, 7   | NO                 |                                     |
|    | - generalized                             |                        |           |                    |                                     |
|    | - Bas_PRA1                                | Autosomal<br>recessive | 1, 6, 7   | NO                 | Mutation in the S-<br>antigen (SAG) |
| G. | Optic nerve coloboma                      | Not defined            | 1, 2      | NO                 |                                     |

## Description and Comments

### A. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

### B. Corneal dystrophy - endothelial

Corneal endothelial dystrophy is an abnormal loss of the inner lining of the cornea that causes progressive fluid retention (edema). With time the edema results in keratitis and decreased vision. This usually does not occur until the animal is older. In the Basenji, this condition is less common than corneal endothelial disease caused by attachment of persistent pupillary membranes.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

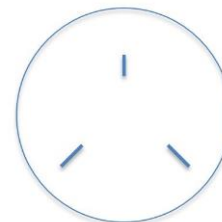
In the Basenji, this is a particularly significant problem with many cases reported where the strands bridge between the iris and the cornea resulting in localized corneal opacities which may cause vision impairment. This has also been associated with optic nerve coloboma (see "F" below).

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Y-suture tip opacity

These are prominent (or "highlighted" or "more dense") distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a "peace sign" as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the "Lens" section of the CAER form. The newest version of the form (3/16/21) has boxes that say, "posterior Y-suture tip opacities" which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: "punctate posterior sutures" AND ALSO MARK "suspect not inherited/significance unknown" (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: "E2" or "posterior suture tip opacities." This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

F. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. Except for X-linked PRA in the Siberian Husky, in all breeds studied to date, PRA is inherited as an autosomal recessive trait.

Bas\_PRA1

A specific mutation has been located in the S-antigen (*SAG*) gene that causes a late onset form of retinal degeneration in the Basenji. The condition is inherited in an autosomal recessive fashion. Initial thinning of the retina evidenced by irregular hypo and hyper-reflectivity of the tapetal fundus is typically noted at 5 years of age with retinal vascular attenuation noted by 6-7 years of age. Clinically the disease closely resembles *prcd*-PRA. The retinal degeneration progresses gradually and ultimately results in complete vision loss. This mutation is responsible for the majority, but not all cases of PRA within the Basenji breed.

G. Optic nerve coloboma

A congenital cavity in the optic nerve which, if large, may cause blindness or vision impairment.

In the Basenji, this condition has been associated with persistent pupillary membranes (see "C" above).

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Barnett KC and Knight CG. Persistent pupillary membrane and associated defects in the Basenji. *Vet Rec.* 1969 Aug 30;85:242-248.
3. Roberts SR and Bistner SI. Persistent pupillary membrane in Basenji dogs. *J Am Vet Med Assoc.* 1968 Sep 1;153:533-542.
4. Mason TA. Persistent pupillary membrane in the Basenji. *Aust Vet J.* 1976 Aug;52:343-344.
5. Bistner SI, Rubin LF and Roberts SR. A review of persistent pupillary membranes in the Basenji dog. *J Am Anim Hosp Assoc.* 1971;7:143.
6. Priester W. Canine progressive retinal atrophy: Occurrence by age, breed, and sex. *American Journal of Veterinary Research.* 1974;35:571-574.
7. Goldstein O, Jordan JA, Aguirre GD, et al. A non-stop S-antigen gene mutation is associated with late onset hereditary retinal degeneration in dogs. *Mol Vis.* 2013;19:1871-1884.

# OCULAR DISORDERS REPORT BASENJI

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>10,454 |       | 2016-2020<br>1,573 |       |
|---|---------------------|---------------------|-------|--------------------|-------|
|   |                     | #                   | %     | #                  | %     |
| <b>GLOBE</b>  |                     |                     |       |                    |       |
| 0.110 microphthalmia  |                     | 8                   | 0.1%  | 0                  |       |
| <b>EYELIDS</b>  |                     |                     |       |                    |       |
| 20.160 macropalpebral fissure   |                     | 1                   | 0.0%  | 0                  |       |
| 21.000 entropion, unspecified   |                     | 6                   | 0.1%  | 0                  |       |
| 22.000 ectropion, unspecified   |                     | 1                   | 0.0%  | 0                  |       |
| 25.110 distichiasis   |                     | 61                  | 0.6%  | 13                 | 0.8%  |
| <b>CORNEA</b>   |                     |                     |       |                    |       |
| 70.210 corneal pannus   |                     | 2                   | 0.0%  | 0                  |       |
| 70.220 pigmentary keratitis   |                     | 2                   | 0.0%  | 0                  |       |
| 70.700 corneal dystrophy  |                     | 314                 | 3.0%  | 41                 | 2.6%  |
| 70.730 corneal endothelial degeneration                               |                     | 242                 | 2.3%  | 12                 | 0.8%  |
| <b>UVEA</b>   |                     |                     |       |                    |       |
| 90.250 pigmentary uveitis   |                     | 1                   | 0.0%  | 0                  |       |
| 93.110 iris hypoplasia  |                     | 0                   |       | 1                  | 0.1%  |
| 93.140 corneal endothelial pigment without PPM                        |                     | 18                  | 0.2%  | 0                  |       |
| 93.150 iris coloboma  |                     | 9                   | 0.1%  | 0                  |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 5,263               | 50.3% | 1,041              | 66.2% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 449                 | 4.3%  | 33                 | 2.1%  |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 1,100               | 10.5% | 117                | 7.4%  |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 40                  | 0.4%  | 7                  | 0.4%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 14                  | 0.1%  | 23                 | 1.5%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 175                 | 1.7%  | 133                | 8.5%  |
| 93.810 uveal melanoma   |                     | 0                   |       | 1                  | 0.1%  |
| 93.999 uveal cysts  |                     | 2                   | 0.0%  | 6                  | 0.4%  |
| 97.150 chorioretinal coloboma, congenital                             |                     | 1                   | 0.0%  | 0                  |       |
| <b>LENS</b>   |                     |                     |       |                    |       |
| 100.200 cataract, unspecified   |                     | 47                  | 0.4%  | 0                  |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 461                 | 4.4%  | 50                 | 3.2%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 42                  | 0.4%  | 4                  | 0.3%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 16                  | 0.2%  | 2                  | 0.1%  |
| 100.303 punctate cataract, equatorial cortex                          |                     | 9                   | 0.1%  | 0                  |       |
| 100.304 punctate cataract, anterior sutures                           |                     | 5                   | 0.0%  | 0                  |       |
| 100.305 punctate cataract, posterior sutures                          |                     | 63                  | 0.6%  | 19                 | 1.2%  |
| 100.306 punctate cataract, nucleus                                    |                     | 15                  | 0.1%  | 4                  | 0.3%  |
| 100.307 punctate cataract, capsular                                   |                     | 58                  | 0.6%  | 22                 | 1.4%  |
| 100.310 incipient cataract, unspecified                               |                     | 0                   |       | 1                  | 0.1%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 29                  | 0.3%  | 3                  | 0.2%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 26                  | 0.2%  | 8                  | 0.5%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 17                  | 0.2%  | 4                  | 0.3%  |
| 100.314 incipient cataract, anterior sutures                          |                     | 3                   | 0.0%  | 0                  |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 30                  | 0.3%  | 7                  | 0.4%  |
| 100.316 incipient cataract, nucleus                                   |                     | 21                  | 0.2%  | 1                  | 0.1%  |
| 100.317 incipient cataract, capsular                                  |                     | 23                  | 0.2%  | 3                  | 0.2%  |
| 100.328 y-suture tip opacities  |                     | 6                   | 0.1%  | 20                 | 1.3%  |
| 100.330 generalized/complete cataract                                 |                     | 22                  | 0.2%  | 0                  |       |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.345 significant cataracts (summary)                   | 432 4.1%         | 98 6.2%          |
| 100.375 subluxation/luxation, unspecified                 | 9 0.1%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 9 0.1%           | 5 0.3%           |
| 110.135 PHPV/PTVL   | 8 0.1%           | 1 0.1%           |
| 110.200 vitreous degeneration-anterior chamber            | 1 0.0%           | 2 0.1%           |
| 110.320 vitreal degeneration                              | 29 0.3%          | 0                |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 1 0.0%           | 0                |
| 97.120 coloboma   | 13 0.1%          | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 20 0.2%          | 3 0.2%           |
| 120.180 retinal dysplasia, geographic                     | 19 0.2%          | 1 0.1%           |
| 120.190 retinal dysplasia, detached                       | 4 0.0%           | 1 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 379 3.6%         | 2 0.1%           |
| 120.400 retinal hemorrhage                                | 5 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 7 0.1%           | 0                |
| 120.960 retinopathy                                       | 9 0.1%           | 4 0.3%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 1 0.0%           | 0                |
| 130.120 optic nerve hypoplasia                            | 3 0.0%           | 0                |
| 130.150 optic disc coloboma                               | 99 0.9%          | 9 0.6%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 78 0.7%          | 0                |
| 900.100 other, not inherited                              | 223 2.1%         | 6 0.4%           |
| 900.110 other. suspect not inherited/significance unknown | 259 2.5%         | 45 2.9%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 4,130 39.5%      | 361 22.9%        |

# BASSET FAUVE DE BRETAGNE

|    | DISORDER  | INHERITANCE         | REFERENCE | BREEDING ADVICE         | GENETIC TESTS AVAILABLE              |
|----|---|---------------------|-----------|-------------------------|--------------------------------------|
| A. | Glaucoma - POAG   | Autosomal recessive | 2-8       | NO                      | Mutation in the <i>ADAMTS17</i> gene |
| B. | Persistent pupillary membranes - lens pigment foci/no strands | Not defined         | 1         | Passes with no notation |                                      |

## Description and Comments

### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure which, when sustained even for a brief period of time, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening exam.

In the Basset Fauve de Bretagne, both closed angle (PCAG) and open angle (POAG) forms of glaucoma are present. Some Basset Fauve de Bretagnes have an abnormality of the iridocorneal angle termed goniodysgenesis. This abnormality is not visible during routine ophthalmologic examination using an indirect ophthalmoscope or a slit-lamp microscope. There appears to be an association between goniodysgenesis and glaucoma, but the mechanism by which the angle defect results in glaucoma has not been determined. It is suspected that mild to severe anterior uveitis impairs outflow of aqueous through the small perforations that are present in the sheet of tissue in the iridocorneal angle; this results in a secondary and often irreversible rise in intraocular pressure that causes blindness.

The inheritance of PCAG and goniodysgenesis in the Basset Fauve de Bretagne are not known. Until the inheritance is determined, control should be directed to removing dogs from breeding that have glaucoma and have goniodysgenesis, as well as those dogs that produce progeny affected with glaucoma. Three genetic loci, *COL1A2*, *RAB22A*, and *NEB*, have been implicated as possible contributors to the development of PCAG in the Basset Fauve de Bretagne. One is an autosomal recessive missense mutation of a nebulin (*NEB*) residue on chromosome 19. Because 33% of unaffected animals were homozygous for the risk allele, it was hypothesized that modifying factors may be present. A genetic test is not yet available for PCAG.

POAG in the Basset Fauve de Bretagne is caused by a 19 base pair deletion in exon 2 of

*ADAMTS17*. This deletion alters the reading frame and is suspected to cause a truncated protein. The trait shows an autosomal recessive mode of inheritance. A DNA test is available.

B. Persistent pupillary membranes (PPM)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Ahram DF, Cook AC, Kecova H, et al. Identification of genetic loci associated with primary angle-closure glaucoma in the basset hound. *Mol Vis*. 2014;20:497-510.
3. Bedford PG. The aetiology of primary glaucoma in the dog. *J Small Anim Pract*. 1975;16:217-239.
4. Bedford PGC. A gonioscopic study of the iridocorneal angle in the English and America breeds of Cocker Spaniel and the Basset Hound. *J Small Anim Pract*. 1977;18:631-642.
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8. Ahram DF, Grozdanic SD, Kecova H, et al. Variants in Nebulin (NEB) Are Linked to the Development of Familial Primary Angle Closure Glaucoma in Basset Hounds. *PloS one*. 2015;10:e0126660.

# OCULAR DISORDERS REPORT BASSET FAUVE DE BRETAGNE

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 10.000 glaucoma   |                     | 2         | 4.7%  | 0         |       |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 25.110 distichiasis   |                     | 0         |       | 1         | 1.3%  |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 2         | 4.7%  | 0         |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 13        | 30.2% | 9         | 11.7% |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 5         | 11.6% | 4         | 5.2%  |
| <b>VITREOUS</b>   |                     |           |       |           |       |
| 110.120 persistent hyaloid artery/remnant                           |                     | 0         |       | 1         | 1.3%  |
| <b>OTHER</b>  |                     |           |       |           |       |
| 900.110 other. suspect not inherited/significance unknown           |                     | 3         | 7.0%  | 3         | 3.9%  |
| <b>NORMAL</b>   |                     |           |       |           |       |
| 0.000 normal globe  |                     | 23        | 53.5% | 60        | 77.9% |



# BASSET HOUND

|    | DISORDER                                | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|---|------------------------|-----------|--------------------|---|
| A. | Glaucoma - POAG                         | Autosomal<br>recessive | 1-9       | NO                 | Mutation in the<br><i>ADAMTS17</i> gene |
| B. | Entropion                               | Not defined            | 1         | Breeder option     |   |
| C. | Ectropion                               | Not defined            | 1, 10     | Breeder option     |   |
| D. | Distichiasis                            | Not defined            | 11        | Breeder option     |   |
| E. | Nictitans cartilage<br>anomaly/eversion | Not defined            | 2         | Breeder option     |   |
| F. | Persistent pupillary<br>membranes       |                        |           |                    |   |
|    | - iris to iris                          | Not defined            | 1         | Breeder option     |   |
|    | - iris to cornea                        | Not defined            | 1         | NO                 |   |
| G. | Cataract                                | Not defined            | 1         | NO                 |   |

## Description and Comments

### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure which, when sustained even for a brief period of time, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening exam.

In the Basset Hound, both closed angle (PCAG) and open angle (POAG) forms of glaucoma are present. Some Basset Hounds have an abnormality of the iridocorneal angle termed goniodysgenesis. This abnormality is not visible during routine ophthalmologic examination using an indirect ophthalmoscope or a slit-lamp microscope. There appears to be an association between goniodysgenesis and glaucoma, but the mechanism by which the angle defect results in glaucoma has not been determined. It is suspected that mild to severe anterior uveitis impairs outflow of aqueous through the small perforations that are present in the sheet of tissue in the iridocorneal angle; this results in a secondary and often irreversible rise in intraocular pressure that causes blindness.

The inheritance of PCAG and goniodysgenesis in the Basset Hound are not known. Until the inheritance is determined, control should be directed to removing dogs from breeding that have glaucoma and have goniodysgenesis, as well as those dogs that produce progeny

affected with glaucoma. Three genetic loci, *COL1A2*, *RAB22A*, and *NEB*, have been implicated as possible contributors to the development of PCAG in the Basset Hound. One is an autosomal recessive missense mutation of a nebulin (*NEB*) residue on chromosome 19. Because 33% of unaffected animals were homozygous for the risk allele, it was hypothesized that modifying factors may be present. A genetic test is not yet available for PCAG.

POAG in the Basset Hound is caused by a 19 base pair deletion in exon 2 of *ADAMTS17*. This deletion alters the reading frame and is suspected to cause a truncated protein. The trait shows an autosomal recessive mode of inheritance. A DNA test is available.

B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

C. Ectropion

A conformational defect resulting in eversion of the eyelids, which may cause ocular irritation. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

In the Basset Hound, ectropion is associated with an exceptionally large palpebral fissure (macroblepharon) and laxity of the canthal structures. Central lower lid ectropion is often associated with entropion of the adjacent lid segment. This causes severe ocular irritation.

It is acknowledged that factors other than genetics may play a role or be the cause of entropion and/or ectropion. However, when non-genetic factors can be ruled out, selection should be directed to a more normal head conformation that minimizes or eliminates the likelihood of the defects.

D. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

E. Nictitans cartilage anomaly/eversion

A scroll-like curling of the cartilage of the third eyelid, usually everting the margin. This condition may occur in one or both eyes and may cause mild ocular irritation.

F. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

G. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

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# OCULAR DISORDERS REPORT BASSET HOUND

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>1,798 |      | 2016-2020<br>167 |       |
|---|---------------------|--------------------|------|------------------|-------|
|   |                     | #                  | %    | #                | %     |
| <b>GLOBE</b>  |                     |                    |      |                  |       |
| 0.110 microphthalmia  |                     | 1                  | 0.1% | 0                |       |
| <b>EYELIDS</b>  |                     |                    |      |                  |       |
| 20.140 ectopic cilia  |                     | 1                  | 0.1% | 0                |       |
| 20.160 macropalpebral fissure   |                     | 17                 | 0.9% | 0                |       |
| 21.000 entropion, unspecified   |                     | 20                 | 1.1% | 7                | 4.2%  |
| 22.000 ectropion, unspecified   |                     | 126                | 7.0% | 20               | 12.0% |
| 25.110 distichiasis   |                     | 25                 | 1.4% | 1                | 0.6%  |
| <b>NASOLACRIMAL</b>   |                     |                    |      |                  |       |
| 40.910 keratoconjunctivitis sicca                                     |                     | 6                  | 0.3% | 0                |       |
| <b>NICTITANS</b>  |                     |                    |      |                  |       |
| 51.100 third eyelid cartilage anomaly                                 |                     | 20                 | 1.1% | 2                | 1.2%  |
| 52.110 prolapsed gland of the third eyelid                            |                     | 9                  | 0.5% | 3                | 1.8%  |
| <b>CORNEA</b>   |                     |                    |      |                  |       |
| 70.210 corneal pannus   |                     | 3                  | 0.2% | 0                |       |
| 70.220 pigmentary keratitis   |                     | 3                  | 0.2% | 0                |       |
| 70.700 corneal dystrophy  |                     | 4                  | 0.2% | 0                |       |
| 70.730 corneal endothelial degeneration                               |                     | 5                  | 0.3% | 0                |       |
| <b>UVEA</b>   |                     |                    |      |                  |       |
| 93.140 corneal endothelial pigment without PPM                        |                     | 1                  | 0.1% | 0                |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 49                 | 2.7% | 6                | 3.6%  |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 11                 | 0.6% | 1                | 0.6%  |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 28                 | 1.6% | 1                | 0.6%  |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 1                  | 0.1% | 0                |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 1                  | 0.1% | 0                |       |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 4                  | 0.2% | 0                |       |
| 93.999 uveal cysts  |                     | 4                  | 0.2% | 1                | 0.6%  |
| <b>LENS</b>   |                     |                    |      |                  |       |
| 100.200 cataract, unspecified   |                     | 6                  | 0.3% | 0                |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 52                 | 2.9% | 10               | 6.0%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 17                 | 0.9% | 0                |       |
| 100.302 punctate cataract, posterior cortex                           |                     | 9                  | 0.5% | 1                | 0.6%  |
| 100.303 punctate cataract, equatorial cortex                          |                     | 5                  | 0.3% | 0                |       |
| 100.304 punctate cataract, anterior sutures                           |                     | 3                  | 0.2% | 0                |       |
| 100.305 punctate cataract, posterior sutures                          |                     | 7                  | 0.4% | 1                | 0.6%  |
| 100.306 punctate cataract, nucleus                                    |                     | 3                  | 0.2% | 2                | 1.2%  |
| 100.307 punctate cataract, capsular                                   |                     | 6                  | 0.3% | 1                | 0.6%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 7                  | 0.4% | 1                | 0.6%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 13                 | 0.7% | 1                | 0.6%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 2                  | 0.1% | 0                |       |
| 100.314 incipient cataract, anterior sutures                          |                     | 1                  | 0.1% | 0                |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 3                  | 0.2% | 0                |       |
| 100.316 incipient cataract, nucleus                                   |                     | 4                  | 0.2% | 0                |       |
| 100.317 incipient cataract, capsular                                  |                     | 3                  | 0.2% | 2                | 1.2%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.326 incomplete cataract, nucleus                      | 2 0.1%           | 0                |
| 100.327 incomplete cataract, capsular                     | 1 0.1%           | 0                |
| 100.328 y-suture tip opacities                            | 0                | 1 0.6%           |
| 100.330 generalized/complete cataract                     | 5 0.3%           | 0                |
| 100.345 <i>significant cataracts (summary)</i>            | 97 5.4%          | 10 6.0%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 2 0.1%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 7 0.4%           | 0                |
| 110.135 PHPV/PTVL   | 1 0.1%           | 0                |
| 110.320 vitreal degeneration                              | 5 0.3%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 11 0.6%          | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 2 0.1%           | 0                |
| 120.400 retinal hemorrhage                                | 1 0.1%           | 0                |
| 120.910 retinal detachment without dialysis               | 2 0.1%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.120 optic nerve hypoplasia                            | 1 0.1%           | 1 0.6%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 19 1.1%          | 0                |
| 900.100 other, not inherited                              | 43 2.4%          | 7 4.2%           |
| 900.110 other. suspect not inherited/significance unknown | 98 5.5%          | 8 4.8%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,384 77.0%      | 106 63.5%        |

# BEAGLE

|    | DISORDER                                    | INHERITANCE                  | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE           |
|----|---|------------------------------|-----------|--------------------|--------------------------------------|
| A. | Microphthalmia with multiple ocular defects | See below                    | 1, 2      | NO                 |                                      |
| B. | Glaucoma – POAG                             | Presumed autosomal recessive | 5         | NO                 | Mutation in the <i>ADAMTS10</i> gene |
| C. | Distichiasis                                | Not defined                  | 1         | Breeder option     |                                      |
| D. | Corneal dystrophy - epithelial/stromal      | Not defined                  | 16-21     | Breeder option     |                                      |
| E. | Cataract                                    | Not defined                  | 22, 23    | NO                 |                                      |
| F. | Tapetal degeneration                        | Presumed autosomal recessive | 24-27     | Breeder option     |                                      |
| G. | Retinal dysplasia - folds                   | Not defined                  | 1         | Breeder option     |                                      |
| H. | Congenital stationary night blindness       | Autosomal recessive          | 28        | NO                 | Mutation in the <i>LRIT3</i> gene    |

## Description and Comments

### A. Microphthalmia with multiple congenital ocular defects

A developmental anomaly in which the eyeball is abnormally small. This is often associated with other ocular malformations, including defects of the cornea, anterior chamber, lens, and/or retina.

In the Beagle, the condition may be present unilaterally or bilaterally and is characterized by a small globe and associated ocular defects which are variable. Several forms of the condition, all apparently different, are recognized:

1) In one study, complete lens opacities were noted by 5-6 months of age; the severity of the cataract correlated closely with the extent of microphthalmia. Severely microphthalmic eyes also had multiple retinal folds. The disorder appeared to be inherited; the exact mode was not fully defined, although an X-linked disorder could not be ruled out.

2) A different form of microphthalmia is recognized in association with microphakia and

persistent pupillary membrane (PPM). Based on a limited pedigree of one cross, a dominant inheritance was proposed; heterozygotes have PPM and microphakia/cataract and homozygous affected show microphthalmia and multiple congenital ocular anomalies.

3) A third form of microphthalmia is recognized in the breed. This condition is usually unilateral and the fellow eye is normal. The mode of inheritance has not been defined, but autosomal recessive inheritance is suspected.

#### B. Glaucoma

Glaucoma is an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening exam.

Primary open angle glaucoma is present in the breed, and extensive breeding studies have demonstrated its inheritance as autosomal recessive. By one year of age, the intraocular pressure (IOP) is elevated, but the filtration angle is open (early glaucoma). Animals with moderate glaucoma show sustained elevations of IOP, focal disinsertions of the lens zonules and focal closures of the iridocorneal angle. Later the globe enlarges, the lens luxates and the eyes become blind and show the effects of chronic glaucoma. The causative mutation in *ADAMTS10* causes an arginine for glycine substitution at position 661. A DNA test is available.

#### C. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

#### D. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

In the Beagle, corneal dystrophy has been described as an oval opacity located at the junction at the middle and inferior thirds of the cornea. The opacities are caused by accumulation of cholesterol and other lipids within the cornea. Progression was noted with possible vision impairment.

#### E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely

(diffuse) or in a localized region.

Several different types of cataract (anterior capsular, posterior cortical, other) have been reported in the Beagle, but the mode of inheritance of the defects is unknown. When one considers that this breed, particularly the laboratory-bred Beagle, has been the subject of extensive ophthalmological examination, the relatively low incidence of cataracts is surprising.

F. Tapetal degeneration

The tapetum lucidum is a modified choroidal structure present in the eyes of many animals that have good night vision. In Beagles there is a recessively inherited defect of the tapetal layer. Absence of this layer is determined by ophthalmoscopy which shows that the fundus has a uniform reddish coloration. The degeneration of the tapetum occurs as a result of abnormal postnatal development of this structure. The degeneration of the tapetum does not affect vision and does not result in functional or structural damage to the retina. As such, the condition probably represents an insignificant inherited variation of no functional significance.

G. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

H. Congenital stationary night blindness (CSNB)

A non-progressive retinal disease characterized by night blindness; day vision is normal. This condition is very rare and has only been found to date in a research colony in Japan. The condition is inherited in an autosomal recessive manner. Affected dogs had normal retinas on clinical examination, but no detectable rod photoreceptor responses with an electroretinogram (ERG). A DNA test is available.

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# OCULAR DISORDERS REPORT BEAGLE

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 0.110 microphthalmia  |                     | 4         | 0.2%  | 0         |       |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 21.000 entropion, unspecified   |                     | 3         | 0.2%  | 3         | 0.7%  |
| 22.000 ectropion, unspecified   |                     | 1         | 0.1%  | 0         |       |
| 25.110 distichiasis   |                     | 293       | 18.1% | 80        | 18.1% |
| <b>NASOLACRIMAL</b>   |                     |           |       |           |       |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 4         | 0.2%  | 8         | 1.8%  |
| 40.910 keratoconjunctivitis sicca                                     |                     | 3         | 0.2%  | 0         |       |
| <b>NICTITANS</b>  |                     |           |       |           |       |
| 51.100 third eyelid cartilage anomaly                                 |                     | 1         | 0.1%  | 0         |       |
| 52.110 prolapsed gland of the third eyelid                            |                     | 10        | 0.6%  | 1         | 0.2%  |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.220 pigmentary keratitis   |                     | 1         | 0.1%  | 0         |       |
| 70.700 corneal dystrophy  |                     | 6         | 0.4%  | 1         | 0.2%  |
| 70.730 corneal endothelial degeneration                               |                     | 2         | 0.1%  | 0         |       |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.150 iris coloboma  |                     | 0         |       | 1         | 0.2%  |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 20        | 1.2%  | 4         | 0.9%  |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 3         | 0.2%  | 0         |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 1         | 0.1%  | 0         |       |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1         | 0.1%  | 1         | 0.2%  |
| 93.999 uveal cysts  |                     | 2         | 0.1%  | 3         | 0.7%  |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.200 cataract, unspecified   |                     | 9         | 0.6%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 46        | 2.8%  | 13        | 2.9%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 8         | 0.5%  | 1         | 0.2%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 6         | 0.4%  | 0         |       |
| 100.303 punctate cataract, equatorial cortex                          |                     | 2         | 0.1%  | 0         |       |
| 100.305 punctate cataract, posterior sutures                          |                     | 3         | 0.2%  | 0         |       |
| 100.306 punctate cataract, nucleus                                    |                     | 0         |       | 1         | 0.2%  |
| 100.307 punctate cataract, capsular                                   |                     | 3         | 0.2%  | 0         |       |
| 100.311 incipient cataract, anterior cortex                           |                     | 3         | 0.2%  | 0         |       |
| 100.312 incipient cataract, posterior cortex                          |                     | 14        | 0.9%  | 0         |       |
| 100.313 incipient cataract, equatorial cortex                         |                     | 6         | 0.4%  | 2         | 0.5%  |
| 100.315 incipient cataract, posterior sutures                         |                     | 1         | 0.1%  | 0         |       |
| 100.316 incipient cataract, nucleus                                   |                     | 4         | 0.2%  | 1         | 0.2%  |
| 100.317 incipient cataract, capsular                                  |                     | 2         | 0.1%  | 0         |       |
| 100.322 incomplete cataract, posterior cortex                         |                     | 1         | 0.1%  | 0         |       |
| 100.323 incomplete cataract, equatorial cortex                        |                     | 1         | 0.1%  | 0         |       |
| 100.328 y-suture tip opacities  |                     | 1         | 0.1%  | 0         |       |
| 100.330 generalized/complete cataract                                 |                     | 19        | 1.2%  | 0         |       |
| 100.345 significant cataracts (summary)                               |                     | 83        | 5.1%  | 5         | 1.1%  |
| 100.375 subluxation/luxation, unspecified                             |                     | 1         | 0.1%  | 0         |       |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>VITREOUS</b>   |             |           |
| 110.120 persistent hyaloid artery/remnant                 | 1 0.1%      | 0         |
| 110.135 PHPV/PTVL   | 1 0.1%      | 1 0.2%    |
| 110.320 vitreal degeneration                              | 6 0.4%      | 2 0.5%    |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 33 2.0%     | 3 0.7%    |
| 120.180 retinal dysplasia, geographic                     | 6 0.4%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 8 0.5%      | 0         |
| 120.910 retinal detachment without dialysis               | 2 0.1%      | 0         |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 1 0.1%      | 0         |
| 130.120 optic nerve hypoplasia                            | 4 0.2%      | 0         |
| 130.150 optic disc coloboma                               | 1 0.1%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 18 1.1%     | 0         |
| 900.100 other, not inherited                              | 44 2.7%     | 1 0.2%    |
| 900.110 other. suspect not inherited/significance unknown | 24 1.5%     | 17 3.8%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,196 73.8% | 314 70.9% |

## BEARDED COLLIE

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|--|---------------------|-----------|-----------------|-----------------------------------|
| A. | Distichiasis   | Not defined         | 1         | Breeder option  |                                   |
| B. | Corneal dystrophy<br>- epithelial/stromal  | Not defined         | 1         | Breeder option  |                                   |
| C. | Persistent pupillary membranes<br>- iris to iris   | Not defined         | 1         | Breeder option  |                                   |
| D. | Cataract   | Not defined         | 1         | NO              |                                   |
| E. | Y suture tip opacity   | Not defined         | 1         |                 |                                   |
| F. | Retinal dysplasia<br>- folds   | Not defined         | 1         | Breeder option  |                                   |
| G. | Choroidal hypoplasia (Collie Eye Anomaly)<br>- staphyloma/coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- optic nerve coloboma | Autosomal recessive | 1-3       | NO              | Mutation in the <i>NHEJ1</i> gene |

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### Description and Comments

#### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

#### B. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

C. Persistent pupillary membranes (PPMs)

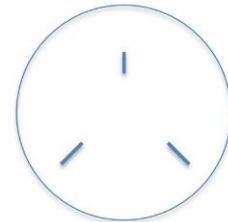
Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

F. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached), which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

- G. Choroidal hypoplasia (Collie Eye Anomaly)
- Staphyloma/coloboma
  - Retinal detachment
  - Retinal hemorrhage
  - Optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Parker HG, Kukekova AV, Akey DT, et al. Breed relationships facilitate fine-mapping studies: a 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. *Genome Res.* 2007 Nov;17:1562-1571.
3. Lowe JK, Kukekova AV, Kirkness EF, et al. Linkage mapping of the primary disease locus for collie eye anomaly. *Genomics.* 2003;82:86-95.

# OCULAR DISORDERS REPORT BEARDED COLLIE

| Diagnostic Name   | TOTAL DOGS EXAMINED |      | 1991-2015 |       | 2016-2020 |   |
|---|---------------------|------|-----------|-------|-----------|---|
|   |                     |      | 3,775     |       | 454       |   |
|   | #                   | %    | #         | %     | #         | % |
| <b>GLOBE</b>  |                     |      |           |       |           |   |
| 0.110 microphthalmia  | 2                   | 0.1% | 0         |       |           |   |
| <b>EYELIDS</b>  |                     |      |           |       |           |   |
| 25.110 distichiasis   | 25                  | 0.7% | 7         | 1.5%  |           |   |
| <b>CORNEA</b>   |                     |      |           |       |           |   |
| 70.700 corneal dystrophy  | 49                  | 1.3% | 3         | 0.7%  |           |   |
| 70.730 corneal endothelial degeneration                             | 1                   | 0.0% | 0         |       |           |   |
| <b>UVEA</b>   |                     |      |           |       |           |   |
| 93.150 iris coloboma  | 1                   | 0.0% | 0         |       |           |   |
| 93.710 persistent pupillary membranes, iris to iris                 | 154                 | 4.1% | 17        | 3.7%  |           |   |
| 93.720 persistent pupillary membranes, iris to lens                 | 9                   | 0.2% | 0         |       |           |   |
| 93.730 persistent pupillary membranes, iris to cornea               | 2                   | 0.1% | 0         |       |           |   |
| 93.740 persistent pupillary membranes, iris sheets                  | 1                   | 0.0% | 0         |       |           |   |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands | 0                   |      | 3         | 0.7%  |           |   |
| 93.999 uveal cysts  | 9                   | 0.2% | 2         | 0.4%  |           |   |
| <b>LENS</b>   |                     |      |           |       |           |   |
| 100.200 cataract, unspecified                                       | 12                  | 0.3% | 0         |       |           |   |
| 100.210 cataract. suspect not inherited/significance unknown        | 374                 | 9.9% | 56        | 12.3% |           |   |
| 100.301 punctate cataract, anterior cortex                          | 38                  | 1.0% | 6         | 1.3%  |           |   |
| 100.302 punctate cataract, posterior cortex                         | 15                  | 0.4% | 1         | 0.2%  |           |   |
| 100.303 punctate cataract, equatorial cortex                        | 28                  | 0.7% | 4         | 0.9%  |           |   |
| 100.304 punctate cataract, anterior sutures                         | 5                   | 0.1% | 0         |       |           |   |
| 100.305 punctate cataract, posterior sutures                        | 21                  | 0.6% | 6         | 1.3%  |           |   |
| 100.306 punctate cataract, nucleus                                  | 6                   | 0.2% | 1         | 0.2%  |           |   |
| 100.307 punctate cataract, capsular                                 | 7                   | 0.2% | 3         | 0.7%  |           |   |
| 100.311 incipient cataract, anterior cortex                         | 37                  | 1.0% | 2         | 0.4%  |           |   |
| 100.312 incipient cataract, posterior cortex                        | 33                  | 0.9% | 1         | 0.2%  |           |   |
| 100.313 incipient cataract, equatorial cortex                       | 23                  | 0.6% | 3         | 0.7%  |           |   |
| 100.314 incipient cataract, anterior sutures                        | 3                   | 0.1% | 0         |       |           |   |
| 100.315 incipient cataract, posterior sutures                       | 10                  | 0.3% | 1         | 0.2%  |           |   |
| 100.316 incipient cataract, nucleus                                 | 12                  | 0.3% | 4         | 0.9%  |           |   |
| 100.317 incipient cataract, capsular                                | 9                   | 0.2% | 3         | 0.7%  |           |   |
| 100.321 incomplete cataract, anterior cortex                        | 2                   | 0.1% | 1         | 0.2%  |           |   |
| 100.322 incomplete cataract, posterior cortex                       | 0                   |      | 1         | 0.2%  |           |   |
| 100.327 incomplete cataract, capsular                               | 0                   |      | 1         | 0.2%  |           |   |
| 100.328 y-suture tip opacities                                      | 3                   | 0.1% | 8         | 1.8%  |           |   |
| 100.330 generalized/complete cataract                               | 5                   | 0.1% | 0         |       |           |   |
| 100.345 <i>significant cataracts (summary)</i>                      | 269                 | 7.1% | 46        | 10.1% |           |   |
| 100.375 <i>subluxation/luxation, unspecified</i>                    | 6                   | 0.2% | 1         | 0.2%  |           |   |
| <b>VITREOUS</b>   |                     |      |           |       |           |   |
| 110.120 persistent hyaloid artery/remnant                           | 6                   | 0.2% | 0         |       |           |   |
| 110.320 vitreal degeneration  | 7                   | 0.2% | 1         | 0.2%  |           |   |
| <b>FUNDUS</b>   |                     |      |           |       |           |   |
| 97.110 choroidal hypoplasia   | 22                  | 0.6% | 0         |       |           |   |
| 97.120 coloboma   | 4                   | 0.1% | 0         |       |           |   |



|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 52 1.4%     | 0         |
| 120.180 retinal dysplasia, geographic                     | 1 0.0%      | 2 0.4%    |
| 120.310 generalized progressive retinal atrophy (PRA)     | 8 0.2%      | 0         |
| 120.960 retinopathy                                       | 2 0.1%      | 0         |
| <b>OPTIC NERVE</b>  |             |           |
| 130.150 optic disc coloboma                               | 1 0.0%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 37 1.0%     | 0         |
| 900.100 other, not inherited                              | 73 1.9%     | 1 0.2%    |
| 900.110 other. suspect not inherited/significance unknown | 26 0.7%     | 30 6.6%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 3,047 80.7% | 315 69.4% |

# BEAUCERON

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--|-------------|-----------|-------------------------|
| A. | Distichiasis   | Not defined | 1         | Breeder option          |
| B. | Persistent pupillary membranes<br>- lens pigment foci/no strands | Not defined | 1         | Passes with no notation |
| C. | Vitreous degeneration  | Not defined | 1         | Breeder option          |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPM)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### C. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Beauceron breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT BEAUCERON

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 0.110 microphthalmia  |                     | 1         | 0.8%  | 0         |       |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 25.110 distichiasis   |                     | 1         | 0.8%  | 5         | 1.3%  |
| <b>NICTITANS</b>  |                     |           |       |           |       |
| 51.100 third eyelid cartilage anomaly                               |                     | 0         |       | 1         | 0.3%  |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.210 corneal pannus   |                     | 1         | 0.8%  | 0         |       |
| 70.700 corneal dystrophy  |                     | 0         |       | 2         | 0.5%  |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 2         | 1.6%  | 12        | 3.0%  |
| 93.720 persistent pupillary membranes, iris to lens                 |                     | 0         |       | 1         | 0.3%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 6         | 4.7%  | 27        | 6.8%  |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 3         | 2.3%  | 14        | 3.5%  |
| 100.302 punctate cataract, posterior cortex                         |                     | 1         | 0.8%  | 1         | 0.3%  |
| 100.305 punctate cataract, posterior sutures                        |                     | 2         | 1.6%  | 0         |       |
| 100.307 punctate cataract, capsular                                 |                     | 1         | 0.8%  | 1         | 0.3%  |
| 100.311 incipient cataract, anterior cortex                         |                     | 0         |       | 2         | 0.5%  |
| 100.315 incipient cataract, posterior sutures                       |                     | 2         | 1.6%  | 0         |       |
| 100.316 incipient cataract, nucleus                                 |                     | 1         | 0.8%  | 1         | 0.3%  |
| 100.317 incipient cataract, capsular                                |                     | 0         |       | 1         | 0.3%  |
| 100.328 y-suture tip opacities                                      |                     | 1         | 0.8%  | 1         | 0.3%  |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 8         | 6.2%  | 7         | 1.8%  |
| 100.375 <i>subluxation/luxation, unspecified</i>                    |                     | 0         |       | 2         | 0.5%  |
| <b>VITREOUS</b>   |                     |           |       |           |       |
| 110.200 vitreous degeneration-anterior chamber                      |                     | 0         |       | 1         | 0.3%  |
| 110.320 vitreal degeneration  |                     | 6         | 4.7%  | 0         |       |
| <b>RETINA</b>   |                     |           |       |           |       |
| 120.170 retinal dysplasia, folds                                    |                     | 0         |       | 4         | 1.0%  |
| 120.180 retinal dysplasia, geographic                               |                     | 1         | 0.8%  | 0         |       |
| <b>OTHER</b>  |                     |           |       |           |       |
| 900.000 other, unspecified  |                     | 3         | 2.3%  | 0         |       |
| 900.110 other. suspect not inherited/significance unknown           |                     | 4         | 3.1%  | 16        | 4.0%  |
| <b>NORMAL</b>   |                     |           |       |           |       |
| 0.000 normal globe  |                     | 104       | 81.2% | 317       | 79.8% |

# BEDLINGTON TERRIER

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Distichiasis                                     | Not defined        | 1                | Breeder option         |
| B. | Imperforate lacrimal punctum                     | Not defined        | 1, 2             | Breeder option         |
| C. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |
| D. | Cataract   | Not defined        | 1                | NO                     |
| E. | Y suture tip opacity                             | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded. Breeding discretion is advised.

### B. Imperforate lacrimal punctum

A developmental anomaly resulting in failure of opening of the lacrimal duct located at the medial lid margins. The lower punctum is more frequently affected. This defect usually results in epiphora, an overflow of tears onto the face.

### C. Persistent pupillary membranes (PPMs)

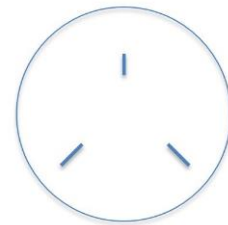
Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

#### D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

#### E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Barnett KC. Imperforate and micro-lachrymal puncta in the dog. *J Small Anim Pract.* 1979 Aug;20:481-490.

# OCULAR DISORDERS REPORT BEDLINGTON TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 0.110 microphthalmia  |                     | 4         | 0.3%  | 1         | 0.4%  |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 20.140 ectopic cilia  |                     | 2         | 0.1%  | 0         |       |
| 21.000 entropion, unspecified   |                     | 2         | 0.1%  | 0         |       |
| 25.110 distichiasis   |                     | 127       | 8.0%  | 12        | 4.6%  |
| <b>NASOLACRIMAL</b>   |                     |           |       |           |       |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 11        | 0.7%  | 4         | 1.5%  |
| <b>NICTITANS</b>  |                     |           |       |           |       |
| 52.110 prolapsed gland of the third eyelid                            |                     | 1         | 0.1%  | 0         |       |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.220 pigmentary keratitis   |                     | 1         | 0.1%  | 0         |       |
| 70.700 corneal dystrophy  |                     | 7         | 0.4%  | 0         |       |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 122       | 7.7%  | 39        | 14.9% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 2         | 0.1%  | 0         |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 5         | 0.3%  | 0         |       |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 3         | 0.2%  | 0         |       |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1         | 0.1%  | 0         |       |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.200 cataract, unspecified   |                     | 13        | 0.8%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 114       | 7.2%  | 20        | 7.7%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 7         | 0.4%  | 3         | 1.1%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 3         | 0.2%  | 1         | 0.4%  |
| 100.303 punctate cataract, equatorial cortex                          |                     | 8         | 0.5%  | 2         | 0.8%  |
| 100.304 punctate cataract, anterior sutures                           |                     | 2         | 0.1%  | 0         |       |
| 100.305 punctate cataract, posterior sutures                          |                     | 14        | 0.9%  | 4         | 1.5%  |
| 100.307 punctate cataract, capsular                                   |                     | 3         | 0.2%  | 2         | 0.8%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 38        | 2.4%  | 1         | 0.4%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 18        | 1.1%  | 0         |       |
| 100.313 incipient cataract, equatorial cortex                         |                     | 31        | 1.9%  | 0         |       |
| 100.314 incipient cataract, anterior sutures                          |                     | 4         | 0.3%  | 0         |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 9         | 0.6%  | 0         |       |
| 100.316 incipient cataract, nucleus                                   |                     | 3         | 0.2%  | 0         |       |
| 100.317 incipient cataract, capsular                                  |                     | 1         | 0.1%  | 0         |       |
| 100.321 incomplete cataract, anterior cortex                          |                     | 1         | 0.1%  | 0         |       |
| 100.322 incomplete cataract, posterior cortex                         |                     | 1         | 0.1%  | 0         |       |
| 100.328 y-suture tip opacities  |                     | 4         | 0.3%  | 2         | 0.8%  |
| 100.330 generalized/complete cataract                                 |                     | 14        | 0.9%  | 2         | 0.8%  |
| 100.345 significant cataracts (summary)                               |                     | 174       | 10.9% | 17        | 6.5%  |
| 100.375 subluxation/luxation, unspecified                             |                     | 1         | 0.1%  | 0         |       |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>VITREOUS</b>   |             |           |
| 110.320 vitreal degeneration                              | 7 0.4%      | 1 0.4%    |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 8 0.5%      | 2 0.8%    |
| 120.190 retinal dysplasia, detached                       | 1 0.1%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 3 0.2%      | 0         |
| 120.910 retinal detachment without dialysis               | 1 0.1%      | 0         |
| 120.960 retinopathy                                       | 1 0.1%      | 0         |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 0           | 2 0.8%    |
| 130.120 optic nerve hypoplasia                            | 1 0.1%      | 0         |
| 130.150 optic disc coloboma                               | 5 0.3%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 13 0.8%     | 0         |
| 900.100 other, not inherited                              | 34 2.1%     | 0         |
| 900.110 other. suspect not inherited/significance unknown | 16 1.0%     | 9 3.4%    |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,188 74.6% | 176 67.4% |



# **OCULAR DISORDERS REPORT BELGIAN LAEKENOIS**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the BELGIAN LAEKENOIS breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT BELGIAN LAEKENOIS

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|------------------|--|-----------|-------|-----------|-------|
|                  |  | #         | %     | #         | %     |
| <b>EYELIDS</b>   |  |           |       |           |       |
| 25.110           | distichiasis   | 5         | 3.3%  | 0         |       |
| <b>NICTITANS</b> |  |           |       |           |       |
| 52.110           | prolapsed gland of the third eyelid                          | 0         |       | 2         | 3.7%  |
| <b>CORNEA</b>    |  |           |       |           |       |
| 70.700           | corneal dystrophy  | 1         | 0.7%  | 0         |       |
| <b>UVEA</b>      |  |           |       |           |       |
| 93.710           | persistent pupillary membranes, iris to iris                 | 1         | 0.7%  | 2         | 3.7%  |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands | 0         |       | 1         | 1.9%  |
| <b>LENS</b>      |  |           |       |           |       |
| 100.210          | cataract. suspect not inherited/significance unknown         | 16        | 10.5% | 2         | 3.7%  |
| 100.302          | punctate cataract, posterior cortex                          | 0         |       | 1         | 1.9%  |
| 100.311          | incipient cataract, anterior cortex                          | 0         |       | 2         | 3.7%  |
| 100.312          | incipient cataract, posterior cortex                         | 0         |       | 1         | 1.9%  |
| 100.322          | incomplete cataract, posterior cortex                        | 0         |       | 1         | 1.9%  |
| 100.345          | <i>significant cataracts (summary)</i>                       | 0         |       | 5         | 9.3%  |
| <b>VITREOUS</b>  |  |           |       |           |       |
| 110.320          | vitreal degeneration   | 5         | 3.3%  | 0         |       |
| <b>RETINA</b>    |  |           |       |           |       |
| 120.170          | retinal dysplasia, folds                                     | 6         | 3.9%  | 0         |       |
| 120.310          | generalized progressive retinal atrophy (PRA)                | 0         |       | 1         | 1.9%  |
| <b>OTHER</b>     |  |           |       |           |       |
| 900.000          | other, unspecified   | 4         | 2.6%  | 0         |       |
| 900.100          | other, not inherited   | 4         | 2.6%  | 0         |       |
| 900.110          | other. suspect not inherited/significance unknown            | 2         | 1.3%  | 0         |       |
| <b>NORMAL</b>    |  |           |       |           |       |
| 0.000            | normal globe   | 125       | 81.7% | 45        | 83.3% |

## BELGIAN MALINOIS

There are 4 varieties of Belgian Shepherd- the Groenendael, Laekenois, Malinois and Tervuren. In Europe these varieties may be interbred and are not considered genetically distinct, thus it is likely that the same genetic diseases exist in all four. In the United States the Groenendael (known as the Belgian Sheepdog), Malinois, Tervuren and Laekenois are recognized as separate breeds.

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |
| B. | Cataract   | Not defined        | 1                | NO                     |
| C. | Y-suture tip opacity                             | Not defined        | 1                | Breeder option         |

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### Description and Comments

#### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

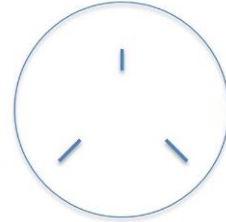
#### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Belgian Malinois, cataract most often occurs as a non-progressive, triangular opacity in the posterior cortex.

### C. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Belgian Malinois breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT BELGIAN MALINOIS

| Diagnostic Name  | TOTAL DOGS EXAMINED  |  | 1991-2015 |      | 2016-2020 |      |
|------------------|--|--|-----------|------|-----------|------|
|                  |  |  | 2,577     |      | 846       |      |
|                  |  |  | #         | %    | #         | %    |
| <b>GLOBE</b>     |  |  |           |      |           |      |
| 0.110            | microphthalmia   |  | 2         | 0.1% | 0         |      |
| <b>EYELIDS</b>   |  |  |           |      |           |      |
| 21.000           | entropion, unspecified   |  | 0         |      | 1         | 0.1% |
| 22.000           | ectropion, unspecified   |  | 1         | 0.0% | 0         |      |
| 25.110           | distichiasis   |  | 2         | 0.1% | 1         | 0.1% |
| <b>NICTITANS</b> |  |  |           |      |           |      |
| 50.210           | pannus of third eyelid   |  | 0         |      | 1         | 0.1% |
| 51.100           | third eyelid cartilage anomaly                                 |  | 0         |      | 3         | 0.4% |
| <b>CORNEA</b>    |  |  |           |      |           |      |
| 70.210           | corneal pannus   |  | 10        | 0.4% | 1         | 0.1% |
| 70.220           | pigmentary keratitis   |  | 1         | 0.0% | 0         |      |
| 70.700           | corneal dystrophy  |  | 16        | 0.6% | 2         | 0.2% |
| 70.730           | corneal endothelial degeneration                               |  | 2         | 0.1% | 0         |      |
| <b>UVEA</b>      |  |  |           |      |           |      |
| 93.710           | persistent pupillary membranes, iris to iris                   |  | 30        | 1.2% | 20        | 2.4% |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands   |  | 0         |      | 3         | 0.4% |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands |  | 1         | 0.0% | 0         |      |
| 93.999           | uveal cysts  |  | 10        | 0.4% | 0         |      |
| <b>LENS</b>      |  |  |           |      |           |      |
| 100.200          | cataract, unspecified  |  | 3         | 0.1% | 0         |      |
| 100.210          | cataract. suspect not inherited/significance unknown           |  | 112       | 4.3% | 46        | 5.4% |
| 100.301          | punctate cataract, anterior cortex                             |  | 12        | 0.5% | 3         | 0.4% |
| 100.302          | punctate cataract, posterior cortex                            |  | 8         | 0.3% | 1         | 0.1% |
| 100.303          | punctate cataract, equatorial cortex                           |  | 1         | 0.0% | 0         |      |
| 100.304          | punctate cataract, anterior sutures                            |  | 2         | 0.1% | 0         |      |
| 100.305          | punctate cataract, posterior sutures                           |  | 12        | 0.5% | 5         | 0.6% |
| 100.306          | punctate cataract, nucleus                                     |  | 2         | 0.1% | 0         |      |
| 100.307          | punctate cataract, capsular                                    |  | 1         | 0.0% | 2         | 0.2% |
| 100.311          | incipient cataract, anterior cortex                            |  | 12        | 0.5% | 6         | 0.7% |
| 100.312          | incipient cataract, posterior cortex                           |  | 21        | 0.8% | 7         | 0.8% |
| 100.313          | incipient cataract, equatorial cortex                          |  | 6         | 0.2% | 1         | 0.1% |
| 100.314          | incipient cataract, anterior sutures                           |  | 7         | 0.3% | 0         |      |
| 100.315          | incipient cataract, posterior sutures                          |  | 8         | 0.3% | 1         | 0.1% |
| 100.316          | incipient cataract, nucleus                                    |  | 15        | 0.6% | 2         | 0.2% |
| 100.317          | incipient cataract, capsular                                   |  | 2         | 0.1% | 2         | 0.2% |
| 100.321          | incomplete cataract, anterior cortex                           |  | 0         |      | 1         | 0.1% |
| 100.322          | incomplete cataract, posterior cortex                          |  | 0         |      | 1         | 0.1% |
| 100.324          | incomplete cataract, anterior sutures                          |  | 1         | 0.0% | 0         |      |
| 100.328          | y-suture tip opacities   |  | 2         | 0.1% | 6         | 0.7% |
| 100.330          | generalized/complete cataract                                  |  | 6         | 0.2% | 0         |      |
| 100.345          | significant cataracts (summary)                                |  | 121       | 4.7% | 38        | 4.5% |
| 100.375          | subluxation/luxation, unspecified                              |  | 1         | 0.0% | 0         |      |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>VITREOUS</b>   |             |           |
| 110.120 persistent hyaloid artery/remnant                 | 1 0.0%      | 2 0.2%    |
| 110.135 PHPV/PTVL   | 2 0.1%      | 0         |
| 110.320 vitreal degeneration                              | 18 0.7%     | 6 0.7%    |
| <b>FUNDUS</b>   |             |           |
| 97.120 coloboma   | 1 0.0%      | 0         |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 22 0.9%     | 4 0.5%    |
| 120.180 retinal dysplasia, geographic                     | 6 0.2%      | 1 0.1%    |
| 120.190 retinal dysplasia, detached                       | 1 0.0%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 13 0.5%     | 1 0.1%    |
| 120.910 retinal detachment without dialysis               | 4 0.2%      | 0         |
| 120.920 retinal detachment with dialysis                  | 1 0.0%      | 3 0.4%    |
| 120.960 retinopathy                                       | 1 0.0%      | 3 0.4%    |
| <b>OPTIC NERVE</b>  |             |           |
| 130.150 optic disc coloboma                               | 1 0.0%      | 1 0.1%    |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 21 0.8%     | 0         |
| 900.100 other, not inherited                              | 78 3.0%     | 1 0.1%    |
| 900.110 other. suspect not inherited/significance unknown | 24 0.9%     | 45 5.3%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 2,286 88.7% | 702 83.0% |

## **BELGIAN SHEEPDOG**

### **(BELGIAN SHEPHERD-GROENENDAEL)**

There are 4 varieties of Belgian Shepherd- the Groenendael, Laekenois, Malinois and Tervuren. In Europe these varieties may be interbred and are not considered genetically distinct, thus it is likely that the same genetic diseases exist in all four. In the United States the Groenendael (known as the Belgian Sheepdog), Malinois, Tervuren and Laekenois are recognized as separate breeds.

|    | <b>DISORDER</b>                       | <b>INHERITANCE</b>  | <b>REFERENCE</b> | <b>BREEDING ADVICE</b>  |
|----|---------------------------------------|---------------------|------------------|-------------------------|
| A. | Chronic superficial keratitis/pannus  | Not defined         | 1                | NO                      |
| B. | Persistent pupillary membranes        |                     |                  |                         |
|    | - iris to iris                        | Not defined         | 1                | Breeder option          |
|    | - lens pigment foci/no strands        | Not defined         | 1                | Passes with no notation |
| C. | Cataract                              | Not defined         | 1                | NO                      |
| D. | Achiasmic optic nerves with nystagmus | Autosomal recessive | 2                | NO                      |

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### **Description and Comments**

#### A. Chronic superficial keratitis/pannus

A bilateral inflammatory disease of the cornea which usually starts as a grayish haze to the ventral or ventrolateral cornea, followed by the formation of a vascularized subepithelial growth that begins to spread toward the central cornea; pigmentation follows the vascularization. If severe, vision impairment occurs. Pannus may be associated with plasma cell infiltration of the nictitans.

#### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Belgian Sheepdog, cataract most often occurs as a non-progressive, triangular opacity in the posterior cortex.

D. Achromatic optic nerves with nystagmus

Achromatic optic nerves with nystagmus have been described in a small family of black Belgian Sheepdogs. Congenital nystagmus is the clinical sign most commonly noted. All retinal ganglion cell axons extend directly into the ipsilateral optic disc with no chiasmal decussation. No optic nerve hypoplasia/micropapilla was noted in the animals studied and reported.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Hogan D and Williams RW. Analysis of the retinas and optic nerves of achromatic Belgian sheepdogs. *The Journal of comparative neurology*. 1995 Feb 13;352:367-380.



# OCULAR DISORDERS REPORT BELGIAN SHEEPDOG

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015<br>5,646 |      | 2016-2020<br>932 |      |
|------------------|--|--------------------|------|------------------|------|
|                  |  | #                  | %    | #                | %    |
| <b>GLOBE</b>     |  |                    |      |                  |      |
| 10.000           | glaucoma   | 1                  | 0.0% | 0                |      |
| <b>EYELIDS</b>   |  |                    |      |                  |      |
| 22.000           | ectropion, unspecified   | 1                  | 0.0% | 0                |      |
| 25.110           | distichiasis   | 11                 | 0.2% | 3                | 0.3% |
| <b>NICTITANS</b> |  |                    |      |                  |      |
| 50.210           | pannus of third eyelid   | 2                  | 0.0% | 7                | 0.8% |
| 51.100           | third eyelid cartilage anomaly                                 | 3                  | 0.1% | 1                | 0.1% |
| <b>CORNEA</b>    |  |                    |      |                  |      |
| 70.210           | corneal pannus   | 49                 | 0.9% | 18               | 1.9% |
| 70.220           | pigmentary keratitis   | 3                  | 0.1% | 4                | 0.4% |
| 70.700           | corneal dystrophy  | 32                 | 0.6% | 5                | 0.5% |
| 70.730           | corneal endothelial degeneration                               | 1                  | 0.0% | 0                |      |
| <b>UVEA</b>      |  |                    |      |                  |      |
| 93.140           | corneal endothelial pigment without PPM                        | 1                  | 0.0% | 0                |      |
| 93.710           | persistent pupillary membranes, iris to iris                   | 408                | 7.2% | 81               | 8.7% |
| 93.720           | persistent pupillary membranes, iris to lens                   | 3                  | 0.1% | 0                |      |
| 93.730           | persistent pupillary membranes, iris to cornea                 | 4                  | 0.1% | 2                | 0.2% |
| 93.740           | persistent pupillary membranes, iris sheets                    | 5                  | 0.1% | 0                |      |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands   | 9                  | 0.2% | 7                | 0.8% |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands | 3                  | 0.1% | 0                |      |
| 93.999           | uveal cysts  | 3                  | 0.1% | 3                | 0.3% |
| 97.150           | chorioretinal coloboma, congenital                             | 0                  |      | 1                | 0.1% |
| <b>LENS</b>      |  |                    |      |                  |      |
| 100.200          | cataract, unspecified  | 13                 | 0.2% | 0                |      |
| 100.210          | cataract. suspect not inherited/significance unknown           | 199                | 3.5% | 40               | 4.3% |
| 100.301          | punctate cataract, anterior cortex                             | 17                 | 0.3% | 3                | 0.3% |
| 100.302          | punctate cataract, posterior cortex                            | 39                 | 0.7% | 3                | 0.3% |
| 100.303          | punctate cataract, equatorial cortex                           | 5                  | 0.1% | 0                |      |
| 100.304          | punctate cataract, anterior sutures                            | 3                  | 0.1% | 0                |      |
| 100.305          | punctate cataract, posterior sutures                           | 15                 | 0.3% | 2                | 0.2% |
| 100.306          | punctate cataract, nucleus                                     | 4                  | 0.1% | 1                | 0.1% |
| 100.307          | punctate cataract, capsular                                    | 6                  | 0.1% | 5                | 0.5% |
| 100.311          | incipient cataract, anterior cortex                            | 25                 | 0.4% | 2                | 0.2% |
| 100.312          | incipient cataract, posterior cortex                           | 59                 | 1.0% | 6                | 0.6% |
| 100.313          | incipient cataract, equatorial cortex                          | 12                 | 0.2% | 1                | 0.1% |
| 100.314          | incipient cataract, anterior sutures                           | 4                  | 0.1% | 0                |      |
| 100.315          | incipient cataract, posterior sutures                          | 14                 | 0.2% | 0                |      |
| 100.316          | incipient cataract, nucleus                                    | 11                 | 0.2% | 0                |      |
| 100.317          | incipient cataract, capsular                                   | 6                  | 0.1% | 1                | 0.1% |
| 100.321          | incomplete cataract, anterior cortex                           | 0                  |      | 2                | 0.2% |
| 100.322          | incomplete cataract, posterior cortex                          | 0                  |      | 2                | 0.2% |
| 100.325          | incomplete cataract, posterior sutures                         | 1                  | 0.0% | 0                |      |
| 100.328          | y-suture tip opacities   | 4                  | 0.1% | 2                | 0.2% |
| 100.330          | generalized/complete cataract                                  | 7                  | 0.1% | 0                |      |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.345 significant cataracts (summary)                   | 245 4.3%         | 30 3.2%          |
| 100.375 subluxation/luxation, unspecified                 | 0                | 1 0.1%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 3 0.1%           | 0                |
| 110.320 vitreal degeneration                              | 3 0.1%           | 2 0.2%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.120 coloboma   | 2 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 37 0.7%          | 2 0.2%           |
| 120.180 retinal dysplasia, geographic                     | 6 0.1%           | 1 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 4 0.1%           | 0                |
| 120.910 retinal detachment without dialysis               | 2 0.0%           | 0                |
| 120.960 retinopathy                                       | 0                | 1 0.1%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 28 0.5%          | 6 0.6%           |
| 130.120 optic nerve hypoplasia                            | 12 0.2%          | 2 0.2%           |
| 130.150 optic disc coloboma                               | 5 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 54 1.0%          | 0                |
| 900.100 other, not inherited                              | 112 2.0%         | 1 0.1%           |
| 900.110 other. suspect not inherited/significance unknown | 44 0.8%          | 50 5.4%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 4,846 85.8%      | 691 74.1%        |

## BELGIAN TERVUREN

There are 4 varieties of Belgian Shepherd- the Groenendael, Laekenois, Malinois and Tervuren. In Europe these varieties may be interbred and are not considered genetically distinct, thus it is likely that the same genetic diseases exist in all four. In the United States the Groenendael (known as the Belgian Sheepdog), Malinois, Tervuren and Laekenois are recognized as separate breeds.

|    | DISORDER                             | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--------------------------------------|-------------|-----------|-------------------------|
| A. | Distichiasis                         | Not defined | 1         | Breeder option          |
| B. | Chronic superficial keratitis/pannus | Not defined | 1, 2      | NO                      |
| C. | Persistent pupillary membranes       |             |           |                         |
|    | - iris to iris                       | Not defined | 1         | Breeder option          |
|    | - lens pigment foci/no strands       | Not defined | 1         | Passes with no notation |
| D. | Cataract                             | Not defined | 1         | NO                      |
| E. | Micropapilla                         | Not defined | 1         | Breeder option          |

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### Description and Comments

#### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

#### B. Chronic superficial keratitis/pannus

A bilateral inflammatory disease of the cornea which usually starts as a grayish haze to the ventral or ventrolateral cornea, followed by the formation of a vascularized subepithelial growth that begins to spread toward the central cornea; pigmentation follows the vascularization. If severe, vision impairment occurs. Pannus may be associated with plasma cell infiltration of the nictitans.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Belgian Tervuren, cataract most often occurs as a non-progressive, triangular opacity in the posterior cortex.

E. Micropapilla

Micropapilla refers to a small optic disc which is not associated with vision impairment. Optic nerve hypoplasia refers to a congenital defect of the optic nerve which causes blindness and abnormal pupil response in the affected eye. It may be difficult to differentiate between micropapilla and optic nerve hypoplasia on a routine (dilated) screening ophthalmoscopic exam.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Chavkin MJ, Roberts SM, Salman MD, et al. Risk factors for development of chronic superficial keratitis in dogs. *J Am Vet Med Assoc.* 1994 May 15;204:1630-1634.

# OCULAR DISORDERS REPORT BELGIAN TERVUREN

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>12,526 |      | 2016-2020<br>1,798 |       |
|---------------------|--|---------------------|------|--------------------|-------|
|                     |  | #                   | %    | #                  | %     |
| <b>GLOBE</b>        |  |                     |      |                    |       |
| 0.110               | microphthalmia   | 4                   | 0.0% | 0                  |       |
| 10.000              | glaucoma   | 1                   | 0.0% | 0                  |       |
| <b>EYELIDS</b>      |  |                     |      |                    |       |
| 21.000              | entropion, unspecified                                       | 3                   | 0.0% | 0                  |       |
| 25.110              | distichiasis   | 115                 | 0.9% | 8                  | 0.4%  |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |       |
| 40.910              | keratoconjunctivitis sicca                                   | 2                   | 0.0% | 0                  |       |
| <b>NICTITANS</b>    |  |                     |      |                    |       |
| 50.210              | pannus of third eyelid                                       | 5                   | 0.0% | 6                  | 0.3%  |
| 51.100              | third eyelid cartilage anomaly                               | 18                  | 0.1% | 3                  | 0.2%  |
| 52.110              | prolapsed gland of the third eyelid                          | 1                   | 0.0% | 0                  |       |
| <b>CORNEA</b>       |  |                     |      |                    |       |
| 70.210              | corneal pannus   | 87                  | 0.7% | 25                 | 1.4%  |
| 70.220              | pigmentary keratitis   | 6                   | 0.0% | 3                  | 0.2%  |
| 70.700              | corneal dystrophy  | 69                  | 0.6% | 8                  | 0.4%  |
| 70.730              | corneal endothelial degeneration                             | 7                   | 0.1% | 0                  |       |
| <b>UVEA</b>         |  |                     |      |                    |       |
| 93.150              | iris coloboma  | 2                   | 0.0% | 0                  |       |
| 93.710              | persistent pupillary membranes, iris to iris                 | 933                 | 7.4% | 207                | 11.5% |
| 93.720              | persistent pupillary membranes, iris to lens                 | 12                  | 0.1% | 1                  | 0.1%  |
| 93.730              | persistent pupillary membranes, iris to cornea               | 5                   | 0.0% | 0                  |       |
| 93.740              | persistent pupillary membranes, iris sheets                  | 14                  | 0.1% | 0                  |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands | 28                  | 0.2% | 32                 | 1.8%  |
| 93.810              | uveal melanoma   | 0                   |      | 2                  | 0.1%  |
| 93.999              | uveal cysts  | 17                  | 0.1% | 1                  | 0.1%  |
| <b>LENS</b>         |  |                     |      |                    |       |
| 100.200             | cataract, unspecified  | 66                  | 0.5% | 0                  |       |
| 100.210             | cataract. suspect not inherited/significance unknown         | 650                 | 5.2% | 132                | 7.3%  |
| 100.301             | punctate cataract, anterior cortex                           | 59                  | 0.5% | 12                 | 0.7%  |
| 100.302             | punctate cataract, posterior cortex                          | 89                  | 0.7% | 12                 | 0.7%  |
| 100.303             | punctate cataract, equatorial cortex                         | 17                  | 0.1% | 0                  |       |
| 100.304             | punctate cataract, anterior sutures                          | 3                   | 0.0% | 5                  | 0.3%  |
| 100.305             | punctate cataract, posterior sutures                         | 31                  | 0.2% | 4                  | 0.2%  |
| 100.306             | punctate cataract, nucleus                                   | 3                   | 0.0% | 2                  | 0.1%  |
| 100.307             | punctate cataract, capsular                                  | 17                  | 0.1% | 10                 | 0.6%  |
| 100.311             | incipient cataract, anterior cortex                          | 59                  | 0.5% | 9                  | 0.5%  |
| 100.312             | incipient cataract, posterior cortex                         | 129                 | 1.0% | 31                 | 1.7%  |
| 100.313             | incipient cataract, equatorial cortex                        | 18                  | 0.1% | 6                  | 0.3%  |
| 100.314             | incipient cataract, anterior sutures                         | 6                   | 0.0% | 1                  | 0.1%  |
| 100.315             | incipient cataract, posterior sutures                        | 26                  | 0.2% | 5                  | 0.3%  |
| 100.316             | incipient cataract, nucleus                                  | 3                   | 0.0% | 2                  | 0.1%  |
| 100.317             | incipient cataract, capsular                                 | 15                  | 0.1% | 5                  | 0.3%  |
| 100.321             | incomplete cataract, anterior cortex                         | 0                   |      | 1                  | 0.1%  |
| 100.322             | incomplete cataract, posterior cortex                        | 0                   |      | 5                  | 0.3%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.326 incomplete cataract, nucleus                      | 0                | 1 0.1%           |
| 100.328 y-suture tip opacities                            | 4 0.0%           | 9 0.5%           |
| 100.330 generalized/complete cataract                     | 12 0.1%          | 0                |
| 100.340 resorbing/hypermature cataract                    | 1 0.0%           | 0                |
| 100.345 <i>significant cataracts (summary)</i>            | 558 4.5%         | 120 6.7%         |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 1 0.0%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 9 0.1%           | 5 0.3%           |
| 110.135 PHPV/PTVL   | 3 0.0%           | 0                |
| 110.320 vitreal degeneration                              | 29 0.2%          | 16 0.9%          |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 1 0.0%           | 0                |
| 97.120 coloboma   | 2 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 41 0.3%          | 2 0.1%           |
| 120.180 retinal dysplasia, geographic                     | 11 0.1%          | 2 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 23 0.2%          | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| 120.920 retinal detachment with dialysis                  | 1 0.0%           | 0                |
| 120.960 retinopathy                                       | 2 0.0%           | 4 0.2%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 120 1.0%         | 21 1.2%          |
| 130.120 optic nerve hypoplasia                            | 91 0.7%          | 5 0.3%           |
| 130.150 optic disc coloboma                               | 4 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 107 0.9%         | 0                |
| 900.100 other, not inherited                              | 250 2.0%         | 2 0.1%           |
| 900.110 other. suspect not inherited/significance unknown | 143 1.1%         | 115 6.4%         |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 10,438 83.3%     | 1,238 68.9%      |

## BERGER PICARD (PICARDY SHEPHERD, PICARDIE)

|    | DISORDER                                      | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|---|-------------|-----------|-----------------|
| A. | Distichiasis                                  | Not defined | 1         | Breeder option  |
| B. | Nictitans cartilage anomaly/eversion          | Not defined | 1         | Breeder option  |
| C. | Corneal dystrophy - epithelial/stromal        | Not defined | 1         | Breeder option  |
| D. | Persistent pupillary membranes - iris to iris | Not defined | 1         | Breeder option  |
| E. | Cataract                                      | Not defined | 1         | NO              |
| F. | Y suture tip opacity                          | Not defined | 1         | Breeder option  |
| G. | Retinal atrophy - generalized                 | Not defined | 1         | NO              |
| H. | Retinal dysplasia - folds                     | Not defined | 1         | Breeder option  |
| I. | Retinopathy                                   | Not defined | 1         | Breeder option  |

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### Description and Comments

#### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

#### B. Nictitans cartilage anomaly/eversion

A scroll-like curling of the cartilage of the third eyelid, usually everting the margin. This condition may occur in one or both eyes and may cause mild ocular irritation.

C. Corneal dystrophy- epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

D. Persistent pupillary membranes (PPMs)

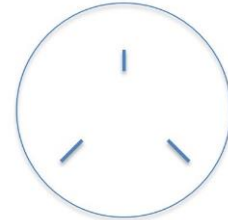
Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

F. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.



G. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality is also known as progressive retinal atrophy or PRA, and may be detected by electroretinogram (not part of a routine eye screening examination) before there are detectable fundusoscopic changes seen by ophthalmoscopy. There are multiple genetic types of PRA including the rod cone dysplasias described elsewhere.

H. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

I. Retinopathy

A lesion similar to canine multifocal retinopathy has been noted in the Berger Picard. The lesions initially appear as multifocal sub-retinal fluid elevations that over time may become hyper-reflective lesions.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT BERGER PICARD

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 0.110 microphthalmia  |                     | 0         |       | 1         | 0.1%  |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 25.110 distichiasis   |                     | 64        | 8.1%  | 46        | 6.0%  |
| <b>NASOLACRIMAL</b>   |                     |           |       |           |       |
| 40.910 keratoconjunctivitis sicca                                     |                     | 2         | 0.3%  | 0         |       |
| <b>NICTITANS</b>  |                     |           |       |           |       |
| 51.100 third eyelid cartilage anomaly                                 |                     | 14        | 1.8%  | 17        | 2.2%  |
| 52.110 prolapsed gland of the third eyelid                            |                     | 0         |       | 2         | 0.3%  |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.700 corneal dystrophy  |                     | 11        | 1.4%  | 18        | 2.4%  |
| <b>UVEA</b>   |                     |           |       |           |       |
| 90.250 pigmentary uveitis   |                     | 1         | 0.1%  | 0         |       |
| 93.150 iris coloboma  |                     | 0         |       | 1         | 0.1%  |
| 93.180 liris sphincter dysplasia                                      |                     | 0         |       | 1         | 0.1%  |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 199       | 25.2% | 94        | 12.3% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 0         |       | 1         | 0.1%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 1         | 0.1%  | 0         |       |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1         | 0.1%  | 0         |       |
| 93.810 uveal melanoma   |                     | 1         | 0.1%  | 0         |       |
| 93.999 uveal cysts  |                     | 2         | 0.3%  | 5         | 0.7%  |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 93        | 11.8% | 82        | 10.8% |
| 100.301 punctate cataract, anterior cortex                            |                     | 0         |       | 5         | 0.7%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 1         | 0.1%  | 4         | 0.5%  |
| 100.304 punctate cataract, anterior sutures                           |                     | 0         |       | 2         | 0.3%  |
| 100.305 punctate cataract, posterior sutures                          |                     | 18        | 2.3%  | 20        | 2.6%  |
| 100.307 punctate cataract, capsular                                   |                     | 3         | 0.4%  | 5         | 0.7%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 1         | 0.1%  | 4         | 0.5%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 5         | 0.6%  | 8         | 1.0%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 0         |       | 1         | 0.1%  |
| 100.314 incipient cataract, anterior sutures                          |                     | 1         | 0.1%  | 0         |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 7         | 0.9%  | 9         | 1.2%  |
| 100.316 incipient cataract, nucleus                                   |                     | 0         |       | 2         | 0.3%  |
| 100.317 incipient cataract, capsular                                  |                     | 0         |       | 1         | 0.1%  |
| 100.321 incomplete cataract, anterior cortex                          |                     | 0         |       | 3         | 0.4%  |
| 100.322 incomplete cataract, posterior cortex                         |                     | 3         | 0.4%  | 3         | 0.4%  |
| 100.325 incomplete cataract, posterior sutures                        |                     | 0         |       | 1         | 0.1%  |
| 100.326 incomplete cataract, nucleus                                  |                     | 1         | 0.1%  | 0         |       |
| 100.328 y-suture tip opacities  |                     | 12        | 1.5%  | 28        | 3.7%  |
| 100.330 generalized/complete cataract                                 |                     | 0         |       | 1         | 0.1%  |
| 100.345 significant cataracts (summary)                               |                     | 52        | 6.6%  | 97        | 12.7% |

|   | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 5 0.6%           | 4 0.5%           |
| 110.320 vitreal degeneration                              | 1 0.1%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 159 20.2%        | 80 10.5%         |
| 120.180 retinal dysplasia, geographic                     | 7 0.9%           | 4 0.5%           |
| 120.190 retinal dysplasia, detached                       | 0                | 1 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 14 1.8%          | 16 2.1%          |
| 120.960 retinopathy                                       | 34 4.3%          | 35 4.6%          |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 0                | 2 0.3%           |
| 130.150 optic disc coloboma                               | 1 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 25 3.2%          | 0                |
| 900.100 other, not inherited                              | 13 1.6%          | 10 1.3%          |
| 900.110 other. suspect not inherited/significance unknown | 35 4.4%          | 46 6.0%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 343 43.5%        | 393 51.6%        |

# BERNESE MOUNTAIN DOG

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Entropion  | Not defined | 1         | Breeder option  |
| B. | Distichiasis                                     | Not defined | 1         | Breeder option  |
| C. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| D. | Cataract   | Not defined | 1         | NO              |
| E. | Retinal atrophy -<br>generalized                 | Not defined | 1, 2      | NO              |

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## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. Except for X-linked PRA in the Siberian Husky, in all breeds studied to date, PRA is inherited as an autosomal recessive trait.

In the Bernese Mountain Dog, one French report found the early onset retinopathy to be functionally and electroretinographically similar to the congenital stationary night blindness (retinal dystrophy) seen in the Briard.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Chaudieu G and Molon-Noblot S. Early retinopathy in the Bernese Mountain Dog in France: preliminary observations. *Vet Ophthalmol.* 2004 May-Jun;7:175-184.

# OCULAR DISORDERS REPORT BERNESE MOUNTAIN DOG

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>15,937 |      | 2016-2020<br>3,328 |      |
|---|---------------------|---------------------|------|--------------------|------|
|   |                     | #                   | %    | #                  | %    |
| <b>GLOBE</b>  |                     |                     |      |                    |      |
| 0.110 microphthalmia  |                     | 6                   | 0.0% | 1                  | 0.0% |
| <b>EYELIDS</b>  |                     |                     |      |                    |      |
| 20.160 macropalpebral fissure   |                     | 25                  | 0.2% | 0                  |      |
| 21.000 entropion, unspecified   |                     | 239                 | 1.5% | 39                 | 1.2% |
| 22.000 ectropion, unspecified   |                     | 103                 | 0.6% | 16                 | 0.5% |
| 25.110 distichiasis   |                     | 141                 | 0.9% | 45                 | 1.4% |
| <b>NASOLACRIMAL</b>   |                     |                     |      |                    |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 1                   | 0.0% | 0                  |      |
| <b>NICTITANS</b>  |                     |                     |      |                    |      |
| 51.100 third eyelid cartilage anomaly                                 |                     | 40                  | 0.3% | 5                  | 0.2% |
| 52.110 prolapsed gland of the third eyelid                            |                     | 1                   | 0.0% | 2                  | 0.1% |
| <b>CORNEA</b>   |                     |                     |      |                    |      |
| 70.210 corneal pannus   |                     | 2                   | 0.0% | 1                  | 0.0% |
| 70.700 corneal dystrophy  |                     | 63                  | 0.4% | 13                 | 0.4% |
| 70.730 corneal endothelial degeneration                               |                     | 4                   | 0.0% | 0                  |      |
| <b>UVEA</b>   |                     |                     |      |                    |      |
| 93.110 iris hypoplasia  |                     | 1                   | 0.0% | 0                  |      |
| 93.150 iris coloboma  |                     | 8                   | 0.1% | 0                  |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 591                 | 3.7% | 156                | 4.7% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 14                  | 0.1% | 5                  | 0.2% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 6                   | 0.0% | 5                  | 0.2% |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 5                   | 0.0% | 0                  |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 16                  | 0.1% | 30                 | 0.9% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 9                   | 0.1% | 3                  | 0.1% |
| 93.999 uveal cysts  |                     | 52                  | 0.3% | 8                  | 0.2% |
| <b>LENS</b>   |                     |                     |      |                    |      |
| 100.200 cataract, unspecified   |                     | 6                   | 0.0% | 0                  |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 957                 | 6.0% | 155                | 4.7% |
| 100.301 punctate cataract, anterior cortex                            |                     | 77                  | 0.5% | 20                 | 0.6% |
| 100.302 punctate cataract, posterior cortex                           |                     | 80                  | 0.5% | 15                 | 0.5% |
| 100.303 punctate cataract, equatorial cortex                          |                     | 41                  | 0.3% | 10                 | 0.3% |
| 100.304 punctate cataract, anterior sutures                           |                     | 12                  | 0.1% | 5                  | 0.2% |
| 100.305 punctate cataract, posterior sutures                          |                     | 29                  | 0.2% | 7                  | 0.2% |
| 100.306 punctate cataract, nucleus                                    |                     | 21                  | 0.1% | 6                  | 0.2% |
| 100.307 punctate cataract, capsular                                   |                     | 14                  | 0.1% | 12                 | 0.4% |
| 100.311 incipient cataract, anterior cortex                           |                     | 53                  | 0.3% | 14                 | 0.4% |
| 100.312 incipient cataract, posterior cortex                          |                     | 173                 | 1.1% | 16                 | 0.5% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 99                  | 0.6% | 10                 | 0.3% |
| 100.314 incipient cataract, anterior sutures                          |                     | 8                   | 0.1% | 2                  | 0.1% |
| 100.315 incipient cataract, posterior sutures                         |                     | 30                  | 0.2% | 2                  | 0.1% |
| 100.316 incipient cataract, nucleus                                   |                     | 29                  | 0.2% | 9                  | 0.3% |
| 100.317 incipient cataract, capsular                                  |                     | 45                  | 0.3% | 9                  | 0.3% |
| 100.321 incomplete cataract, anterior cortex                          |                     | 0                   |      | 3                  | 0.1% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.322 incomplete cataract, posterior cortex             | 3                | 0.0%  | 1                | 0.0%  |
| 100.323 incomplete cataract, equatorial cortex            | 1                | 0.0%  | 1                | 0.0%  |
| 100.325 incomplete cataract, posterior sutures            | 0                |       | 1                | 0.0%  |
| 100.326 incomplete cataract, nucleus                      | 1                | 0.0%  | 8                | 0.2%  |
| 100.327 incomplete cataract, capsular                     | 0                |       | 3                | 0.1%  |
| 100.328 y-suture tip opacities                            | 2                | 0.0%  | 4                | 0.1%  |
| 100.330 generalized/complete cataract                     | 28               | 0.2%  | 1                | 0.0%  |
| 100.340 resorbing/hypermature cataract                    | 0                |       | 2                | 0.1%  |
| 100.345 <i>significant cataracts (summary)</i>            | 752              | 4.7%  | 161              | 4.8%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 9                | 0.1%  | 1                | 0.0%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 26               | 0.2%  | 8                | 0.2%  |
| 110.135 PHPV/PTVL   | 9                | 0.1%  | 3                | 0.1%  |
| 110.320 vitreal degeneration                              | 29               | 0.2%  | 1                | 0.0%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 1                | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 34               | 0.2%  | 10               | 0.3%  |
| 120.180 retinal dysplasia, geographic                     | 8                | 0.1%  | 1                | 0.0%  |
| 120.190 retinal dysplasia, detached                       | 3                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 51               | 0.3%  | 0                |       |
| 120.400 retinal hemorrhage                                | 2                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 3                | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 0                |       | 1                | 0.0%  |
| 120.960 retinopathy                                       | 3                | 0.0%  | 8                | 0.2%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 19               | 0.1%  | 9                | 0.3%  |
| 130.120 optic nerve hypoplasia                            | 30               | 0.2%  | 6                | 0.2%  |
| 130.150 optic disc coloboma                               | 21               | 0.1%  | 1                | 0.0%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 193              | 1.2%  | 0                |       |
| 900.100 other, not inherited                              | 454              | 2.8%  | 5                | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown | 148              | 0.9%  | 137              | 4.1%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 13,685           | 85.9% | 2,627            | 78.9% |

# BICHON FRISE

|    | DISORDER  | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|---|-------------|-----------|-----------------|
| A. | Entropion   | Not defined | 1         | Breeder option  |
| B. | Distichiasis  | Not defined | 1         | Breeder option  |
| C. | Corneal dystrophy<br>- epithelial/stromal           | Not defined | 1         | Breeder option  |
| D. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| E. | Cataract  | Not defined | 1-3       | NO              |
| F. | Y suture tip opacity                                | Not defined | 1         | Breeder option  |
| G. | Vitreous degeneration                               | Not defined | 1         | Breeder option  |

## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### C. Corneal dystrophy- epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.



D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

In the Bichon Frise, many of these strands bridge between the iris and cornea where they may be associated with corneal opacities and vision impairment.

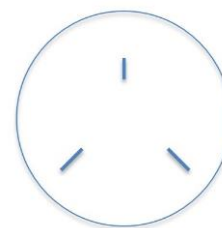
E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

The range in age of animals affected with cataracts in one study was 1-2 years to 9-10 years old, with the peak age of 3 years old. The cataracts involved all regions of the lens, but in age groups of 2-4 years old, the predominant regions affected were the posterior cortex, and the anterior and posterior cortices combined. The earliest abnormalities usually consisted of small punctate opacities in the paracentral posterior cortex, independent of the posterior lens sutures.

F. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts -

which would either be breeder option or failing.

G. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gelatt KN, Wallace MR, Andrew SE, et al. Cataracts in the Bichon Frise. *Vet Ophthalmol.* 2003 Mar;6:3-9.
3. Schmidt GM and Vainisi SJ. Retrospective study of prophylactic random transscleral retinopexy in the Bichon Frise with cataract. *Vet Ophthalmol.* 2004 Sep-Oct;7:307-310.

# OCULAR DISORDERS REPORT BICHON FRISE

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>9,835 |      | 2016-2020<br>1,663 |      |
|---------------------|--|--------------------|------|--------------------|------|
|                     |  | #                  | %    | #                  | %    |
| <b>GLOBE</b>        |  |                    |      |                    |      |
| 0.110               | microphthalmia   | 2                  | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                    |      |                    |      |
| 20.140              | ectopic cilia  | 2                  | 0.0% | 0                  |      |
| 21.000              | entropion, unspecified   | 7                  | 0.1% | 19                 | 1.1% |
| 22.000              | ectropion, unspecified   | 1                  | 0.0% | 0                  |      |
| 25.110              | distichiasis   | 347                | 3.5% | 79                 | 4.8% |
| <b>NASOLACRIMAL</b> |  |                    |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 0                  |      | 2                  | 0.1% |
| 40.910              | keratoconjunctivitis sicca                                     | 2                  | 0.0% | 0                  |      |
| <b>NICTITANS</b>    |  |                    |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 1                  | 0.0% | 0                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 0                  |      | 3                  | 0.2% |
| <b>CORNEA</b>       |  |                    |      |                    |      |
| 70.210              | corneal pannus   | 2                  | 0.0% | 0                  |      |
| 70.220              | pigmentary keratitis   | 2                  | 0.0% | 2                  | 0.1% |
| 70.700              | corneal dystrophy  | 339                | 3.4% | 63                 | 3.8% |
| 70.730              | corneal endothelial degeneration                               | 5                  | 0.1% | 2                  | 0.1% |
| <b>UVEA</b>         |  |                    |      |                    |      |
| 93.110              | iris hypoplasia  | 2                  | 0.0% | 0                  |      |
| 93.140              | corneal endothelial pigment without PPM                        | 2                  | 0.0% | 0                  |      |
| 93.150              | iris coloboma  | 4                  | 0.0% | 0                  |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 223                | 2.3% | 60                 | 3.6% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 13                 | 0.1% | 0                  |      |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 31                 | 0.3% | 1                  | 0.1% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 8                  | 0.1% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 1                  | 0.0% | 6                  | 0.4% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 8                  | 0.1% | 2                  | 0.1% |
| <b>LENS</b>         |  |                    |      |                    |      |
| 100.200             | cataract, unspecified  | 23                 | 0.2% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 534                | 5.4% | 79                 | 4.8% |
| 100.301             | punctate cataract, anterior cortex                             | 93                 | 0.9% | 12                 | 0.7% |
| 100.302             | punctate cataract, posterior cortex                            | 85                 | 0.9% | 6                  | 0.4% |
| 100.303             | punctate cataract, equatorial cortex                           | 11                 | 0.1% | 2                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 8                  | 0.1% | 0                  |      |
| 100.305             | punctate cataract, posterior sutures                           | 37                 | 0.4% | 8                  | 0.5% |
| 100.306             | punctate cataract, nucleus                                     | 9                  | 0.1% | 1                  | 0.1% |
| 100.307             | punctate cataract, capsular                                    | 8                  | 0.1% | 11                 | 0.7% |
| 100.311             | incipient cataract, anterior cortex                            | 84                 | 0.9% | 12                 | 0.7% |
| 100.312             | incipient cataract, posterior cortex                           | 214                | 2.2% | 16                 | 1.0% |
| 100.313             | incipient cataract, equatorial cortex                          | 32                 | 0.3% | 6                  | 0.4% |
| 100.314             | incipient cataract, anterior sutures                           | 2                  | 0.0% | 0                  |      |
| 100.315             | incipient cataract, posterior sutures                          | 46                 | 0.5% | 3                  | 0.2% |
| 100.316             | incipient cataract, nucleus                                    | 9                  | 0.1% | 2                  | 0.1% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.317 incipient cataract, capsular                      | 12               | 0.1%  | 3                | 0.2%  |
| 100.321 incomplete cataract, anterior cortex              | 0                |       | 4                | 0.2%  |
| 100.322 incomplete cataract, posterior cortex             | 3                | 0.0%  | 3                | 0.2%  |
| 100.328 y-suture tip opacities                            | 4                | 0.0%  | 22               | 1.3%  |
| 100.330 generalized/complete cataract                     | 147              | 1.5%  | 2                | 0.1%  |
| 100.345 <i>significant cataracts (summary)</i>            | 827              | 8.4%  | 113              | 6.8%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 4                | 0.0%  | 0                |       |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 20               | 0.2%  | 18               | 1.1%  |
| 110.135 PHPV/PTVL   | 3                | 0.0%  | 0                |       |
| 110.200 vitreous degeneration-anterior chamber            | 2                | 0.0%  | 8                | 0.5%  |
| 110.320 vitreal degeneration                              | 97               | 1.0%  | 34               | 2.0%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.120 coloboma   | 1                | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 67               | 0.7%  | 6                | 0.4%  |
| 120.180 retinal dysplasia, geographic                     | 3                | 0.0%  | 1                | 0.1%  |
| 120.310 generalized progressive retinal atrophy (PRA)     | 58               | 0.6%  | 2                | 0.1%  |
| 120.910 retinal detachment without dialysis               | 1                | 0.0%  | 0                |       |
| 120.960 retinopathy                                       | 2                | 0.0%  | 3                | 0.2%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 1                | 0.0%  | 1                | 0.1%  |
| 130.120 optic nerve hypoplasia                            | 1                | 0.0%  | 0                |       |
| 130.150 optic disc coloboma                               | 10               | 0.1%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 39               | 0.4%  | 0                |       |
| 900.100 other, not inherited                              | 145              | 1.5%  | 0                |       |
| 900.110 other. suspect not inherited/significance unknown | 64               | 0.7%  | 90               | 5.4%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 8,104            | 82.4% | 1,191            | 71.6% |

# **OCULAR DISORDERS REPORT BIEWER TERRIER**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the BIEWER TERRIER breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT BIEWER TERRIER

| Diagnostic Name    | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|--------------------|--|-----------|-------|-----------|-------|
|                    |  | #         | %     | #         | %     |
| <b>EYELIDS</b>     |  |           |       |           |       |
| 25.110             | distichiasis   | 1         | 1.7%  | 3         | 2.9%  |
| <b>CORNEA</b>      |  |           |       |           |       |
| 70.700             | corneal dystrophy  | 0         |       | 1         | 1.0%  |
| <b>UVEA</b>        |  |           |       |           |       |
| 93.710             | persistent pupillary membranes, iris to iris                   | 7         | 11.9% | 3         | 2.9%  |
| 93.720             | persistent pupillary membranes, iris to lens                   | 0         |       | 1         | 1.0%  |
| 93.750             | persistent pupillary membranes, lens pigment foci/no strands   | 2         | 3.4%  | 0         |       |
| 93.760             | persistent pupillary membranes, endothelial opacity/no strands | 0         |       | 1         | 1.0%  |
| <b>LENS</b>        |  |           |       |           |       |
| 100.210            | cataract. suspect not inherited/significance unknown           | 4         | 6.8%  | 0         |       |
| 100.302            | punctate cataract, posterior cortex                            | 0         |       | 1         | 1.0%  |
| 100.330            | generalized/complete cataract                                  | 0         |       | 1         | 1.0%  |
| 100.340            | resorbing/hypermature cataract                                 | 0         |       | 1         | 1.0%  |
| 100.345            | <i>significant cataracts (summary)</i>                         | 0         |       | 3         | 2.9%  |
| <b>VITREOUS</b>    |  |           |       |           |       |
| 110.120            | persistent hyaloid artery/remnant                              | 1         | 1.7%  | 0         |       |
| <b>FUNDUS</b>      |  |           |       |           |       |
| 97.110             | choroidal hypoplasia   | 1         | 1.7%  | 0         |       |
| <b>RETINA</b>      |  |           |       |           |       |
| 120.170            | retinal dysplasia, folds                                       | 0         |       | 2         | 1.9%  |
| <b>OPTIC NERVE</b> |  |           |       |           |       |
| 130.150            | optic disc coloboma  | 1         | 1.7%  | 0         |       |
| <b>OTHER</b>       |  |           |       |           |       |
| 900.000            | other, unspecified   | 1         | 1.7%  | 0         |       |
| <b>NORMAL</b>      |  |           |       |           |       |
| 0.000              | normal globe   | 49        | 83.1% | 93        | 88.6% |

# BLACK AND TAN COONHOUND

|    | DISORDER                     | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|------------------------------|-------------|-----------|-----------------|
| A. | Cataract                     | Not defined | 1         | NO              |
| B. | Retinal dysplasia<br>- folds | Not defined | 1         | Breeder option  |

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## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### B. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT BLACK AND TAN COONHOUND

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |       |
|---|---------------------|-----------|------|-----------|-------|
|   |                     | #         | %    | #         | %     |
| <b>GLOBE</b>  |                     |           |      |           |       |
| 0.110 microphthalmia  |                     | 1         | 0.2% | 0         |       |
| <b>EYELIDS</b>  |                     |           |      |           |       |
| 21.000 entropion, unspecified                                       |                     | 3         | 0.6% | 1         | 0.6%  |
| 22.000 ectropion, unspecified                                       |                     | 6         | 1.1% | 1         | 0.6%  |
| 25.110 distichiasis   |                     | 6         | 1.1% | 0         |       |
| <b>NICTITANS</b>  |                     |           |      |           |       |
| 51.100 third eyelid cartilage anomaly                               |                     | 2         | 0.4% | 0         |       |
| 52.110 prolapsed gland of the third eyelid                          |                     | 1         | 0.2% | 0         |       |
| <b>CORNEA</b>   |                     |           |      |           |       |
| 70.210 corneal pannus   |                     | 2         | 0.4% | 0         |       |
| <b>UVEA</b>   |                     |           |      |           |       |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 5         | 0.9% | 0         |       |
| 93.720 persistent pupillary membranes, iris to lens                 |                     | 3         | 0.6% | 0         |       |
| 93.730 persistent pupillary membranes, iris to cornea               |                     | 0         |      | 2         | 1.2%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 7         | 1.3% | 2         | 1.2%  |
| 93.999 uveal cysts  |                     | 0         |      | 1         | 0.6%  |
| <b>LENS</b>   |                     |           |      |           |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 42        | 7.7% | 8         | 4.6%  |
| 100.301 punctate cataract, anterior cortex                          |                     | 4         | 0.7% | 1         | 0.6%  |
| 100.302 punctate cataract, posterior cortex                         |                     | 1         | 0.2% | 0         |       |
| 100.304 punctate cataract, anterior sutures                         |                     | 0         |      | 3         | 1.7%  |
| 100.305 punctate cataract, posterior sutures                        |                     | 1         | 0.2% | 0         |       |
| 100.306 punctate cataract, nucleus                                  |                     | 6         | 1.1% | 0         |       |
| 100.307 punctate cataract, capsular                                 |                     | 1         | 0.2% | 2         | 1.2%  |
| 100.311 incipient cataract, anterior cortex                         |                     | 1         | 0.2% | 0         |       |
| 100.312 incipient cataract, posterior cortex                        |                     | 5         | 0.9% | 1         | 0.6%  |
| 100.314 incipient cataract, anterior sutures                        |                     | 1         | 0.2% | 1         | 0.6%  |
| 100.316 incipient cataract, nucleus                                 |                     | 3         | 0.6% | 0         |       |
| 100.323 incomplete cataract, equatorial cortex                      |                     | 1         | 0.2% | 0         |       |
| 100.330 generalized/complete cataract                               |                     | 3         | 0.6% | 0         |       |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 27        | 5.0% | 8         | 4.6%  |
| <b>VITREOUS</b>   |                     |           |      |           |       |
| 110.120 persistent hyaloid artery/remnant                           |                     | 0         |      | 1         | 0.6%  |
| 110.135 PHPV/PTVL   |                     | 1         | 0.2% | 0         |       |
| 110.320 vitreal degeneration  |                     | 1         | 0.2% | 0         |       |
| <b>FUNDUS</b>   |                     |           |      |           |       |
| 97.110 choroidal hypoplasia   |                     | 1         | 0.2% | 0         |       |
| <b>RETINA</b>   |                     |           |      |           |       |
| 120.170 retinal dysplasia, folds                                    |                     | 25        | 4.6% | 47        | 27.2% |
| 120.180 retinal dysplasia, geographic                               |                     | 0         |      | 1         | 0.6%  |



|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 2 0.4%    | 0         |
| 900.100 other, not inherited                              | 11 2.0%   | 0         |
| 900.110 other. suspect not inherited/significance unknown | 3 0.6%    | 7 4.0%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 430 79.2% | 105 60.7% |

# BLACK RUSSIAN TERRIER

|    | DISORDER  | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE                                    |
|----|---|---------------------|-----------|-----------------|--|
| A. | Distichiasis  | Not defined         | 1         | Breeder option  |  |
| B. | Persistent pupillary membranes<br>- iris to iris  | Not defined         | 1         | Breeder option  |  |
| C. | Cataract  | Not defined         | 1         | NO              |  |
| D. | POANV<br>(polyneuropathy, ocular abnormalities neuronal vacuolation) -<br>Microphthalmia -<br>Cataracts -PPM (iris to iris) | Autosomal recessive | 2         | NO              | Mutation in the <i>RAB3GAP1</i> :<br><i>c.743delC</i> gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. POANV- Polyneuropathy with ocular abnormalities and neuronal vacuolation

An autosomal recessive condition resulting in juvenile polyneuropathy that presents as laryngeal paralysis and weakness. Patients have concurrent ophthalmic abnormalities including microphthalmia, incomplete cataracts (primarily nuclear) and iris-to-iris PPMs. Neuronal vacuolation was identified on histopathology. Affected dogs were found to be homozygous for the RAB3GAP1: c.743delC mutation. Patients with this variant are not reported to survive past 6 months.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Mhlanga-Mutangadura T, Johnson GJ, Schnabel RD, et al. A mutation in the Warburg syndrome gene, RAB3GAP1, causes a similar syndrome with polyneuropathy and neuronal vacuolation in Black Russian Terrier dogs. *Neurobiology of Disease*. 2016;86:75-85.

# OCULAR DISORDERS REPORT BLACK RUSSIAN TERRIER

| Diagnostic Name    | TOTAL DOGS EXAMINED  | 1991-2015 |      | 2016-2020 |      |
|--------------------|--|-----------|------|-----------|------|
|                    |  | #         | %    | #         | %    |
| <b>EYELIDS</b>     |  |           |      |           |      |
| 21.000             | entropion, unspecified   | 5         | 0.9% | 3         | 1.0% |
| 22.000             | ectropion, unspecified   | 4         | 0.7% | 0         |      |
| 25.110             | distichiasis   | 5         | 0.9% | 6         | 2.0% |
| <b>NICTITANS</b>   |  |           |      |           |      |
| 51.100             | third eyelid cartilage anomaly                                 | 1         | 0.2% | 0         |      |
| 52.110             | prolapsed gland of the third eyelid                            | 1         | 0.2% | 0         |      |
| <b>CORNEA</b>      |  |           |      |           |      |
| 70.700             | corneal dystrophy  | 2         | 0.4% | 3         | 1.0% |
| <b>UVEA</b>        |  |           |      |           |      |
| 93.110             | iris hypoplasia  | 1         | 0.2% | 0         |      |
| 93.150             | iris coloboma  | 1         | 0.2% | 0         |      |
| 93.710             | persistent pupillary membranes, iris to iris                   | 10        | 1.8% | 10        | 3.3% |
| 93.720             | persistent pupillary membranes, iris to lens                   | 1         | 0.2% | 1         | 0.3% |
| 93.750             | persistent pupillary membranes, lens pigment foci/no strands   | 1         | 0.2% | 3         | 1.0% |
| 93.760             | persistent pupillary membranes, endothelial opacity/no strands | 1         | 0.2% | 0         |      |
| 93.810             | uveal melanoma   | 0         |      | 1         | 0.3% |
| 93.999             | uveal cysts  | 3         | 0.5% | 1         | 0.3% |
| <b>LENS</b>        |  |           |      |           |      |
| 100.210            | cataract. suspect not inherited/significance unknown           | 27        | 4.8% | 24        | 7.9% |
| 100.301            | punctate cataract, anterior cortex                             | 3         | 0.5% | 5         | 1.6% |
| 100.302            | punctate cataract, posterior cortex                            | 7         | 1.2% | 2         | 0.7% |
| 100.304            | punctate cataract, anterior sutures                            | 1         | 0.2% | 0         |      |
| 100.305            | punctate cataract, posterior sutures                           | 2         | 0.4% | 0         |      |
| 100.307            | punctate cataract, capsular                                    | 1         | 0.2% | 0         |      |
| 100.311            | incipient cataract, anterior cortex                            | 0         |      | 5         | 1.6% |
| 100.312            | incipient cataract, posterior cortex                           | 9         | 1.6% | 5         | 1.6% |
| 100.315            | incipient cataract, posterior sutures                          | 1         | 0.2% | 0         |      |
| 100.316            | incipient cataract, nucleus                                    | 1         | 0.2% | 0         |      |
| 100.317            | incipient cataract, capsular                                   | 0         |      | 1         | 0.3% |
| 100.326            | incomplete cataract, nucleus                                   | 0         |      | 2         | 0.7% |
| 100.328            | y-suture tip opacities   | 0         |      | 1         | 0.3% |
| 100.345            | <i>significant cataracts (summary)</i>                         | 25        | 4.4% | 21        | 6.9% |
| <b>VITREOUS</b>    |  |           |      |           |      |
| 110.120            | persistent hyaloid artery/remnant                              | 0         |      | 1         | 0.3% |
| 110.320            | vitreal degeneration   | 2         | 0.4% | 0         |      |
| <b>RETINA</b>      |  |           |      |           |      |
| 120.170            | retinal dysplasia, folds                                       | 1         | 0.2% | 2         | 0.7% |
| 120.920            | retinal detachment with dialysis                               | 0         |      | 1         | 0.3% |
| <b>OPTIC NERVE</b> |  |           |      |           |      |
| 130.110            | micropapilla   | 1         | 0.2% | 0         |      |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 12 2.1%   | 0         |
| 900.100 other, not inherited                              | 8 1.4%    | 0         |
| 900.110 other. suspect not inherited/significance unknown | 7 1.2%    | 6 2.0%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 491 87.4% | 234 76.7% |

# BLOODHOUND

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Ectropion  | Not defined | 1         | Breeder option  |
| B. | Entropion  | Not defined | 1         | Breeder option  |
| C. | Persistent pupillary membranes<br>- iris to cornea | Not defined | 1         | NO              |
| D. | Cataract   | Not defined | 1         | NO              |
| E. | Retinal dysplasia<br>- folds                       | Not defined | 1         | Breeder option  |

## Description and Comment

### A. Ectropion

A conformational defect resulting in eversion of the eyelid(s), which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely

(diffuse) or in a localized region.

E. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT BLOODHOUND

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 0.110 microphthalmia  |                     | 1         | 0.2%  | 0         |       |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 20.160 macropalpebral fissure   |                     | 75        | 13.1% | 0         |       |
| 21.000 entropion, unspecified   |                     | 128       | 22.3% | 4         | 7.3%  |
| 22.000 ectropion, unspecified   |                     | 149       | 26.0% | 9         | 16.4% |
| 25.110 distichiasis   |                     | 9         | 1.6%  | 2         | 3.6%  |
| <b>NASOLACRIMAL</b>   |                     |           |       |           |       |
| 40.910 keratoconjunctivitis sicca                                     |                     | 3         | 0.5%  | 0         |       |
| <b>NICTITANS</b>  |                     |           |       |           |       |
| 51.100 third eyelid cartilage anomaly                                 |                     | 1         | 0.2%  | 0         |       |
| 52.110 prolapsed gland of the third eyelid                            |                     | 6         | 1.0%  | 0         |       |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.210 corneal pannus   |                     | 5         | 0.9%  | 0         |       |
| 70.220 pigmentary keratitis   |                     | 3         | 0.5%  | 0         |       |
| 70.730 corneal endothelial degeneration                               |                     | 2         | 0.3%  | 1         | 1.8%  |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 17        | 3.0%  | 1         | 1.8%  |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 5         | 0.9%  | 0         |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 38        | 6.6%  | 1         | 1.8%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 3         | 0.5%  | 0         |       |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1         | 0.2%  | 0         |       |
| 93.999 uveal cysts  |                     | 1         | 0.2%  | 0         |       |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.200 cataract, unspecified   |                     | 1         | 0.2%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 14        | 2.4%  | 2         | 3.6%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 10        | 1.7%  | 0         |       |
| 100.302 punctate cataract, posterior cortex                           |                     | 1         | 0.2%  | 0         |       |
| 100.306 punctate cataract, nucleus                                    |                     | 1         | 0.2%  | 0         |       |
| 100.307 punctate cataract, capsular                                   |                     | 2         | 0.3%  | 0         |       |
| 100.311 incipient cataract, anterior cortex                           |                     | 15        | 2.6%  | 0         |       |
| 100.312 incipient cataract, posterior cortex                          |                     | 6         | 1.0%  | 0         |       |
| 100.314 incipient cataract, anterior sutures                          |                     | 3         | 0.5%  | 0         |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 1         | 0.2%  | 0         |       |
| 100.316 incipient cataract, nucleus                                   |                     | 4         | 0.7%  | 0         |       |
| 100.317 incipient cataract, capsular                                  |                     | 4         | 0.7%  | 1         | 1.8%  |
| 100.321 incomplete cataract, anterior cortex                          |                     | 1         | 0.2%  | 0         |       |
| 100.322 incomplete cataract, posterior cortex                         |                     | 2         | 0.3%  | 0         |       |
| 100.330 generalized/complete cataract                                 |                     | 1         | 0.2%  | 0         |       |
| 100.340 resorbing/hypermature cataract                                |                     | 1         | 0.2%  | 0         |       |
| 100.345 significant cataracts (summary)                               |                     | 53        | 9.2%  | 1         | 1.8%  |



|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>VITREOUS</b>   |           |           |
| 110.120 persistent hyaloid artery/remnant                 | 1 0.2%    | 0         |
| 110.135 PHPV/PTVL   | 1 0.2%    | 0         |
| 110.320 vitreal degeneration                              | 1 0.2%    | 0         |
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 33 5.8%   | 1 1.8%    |
| 120.310 generalized progressive retinal atrophy (PRA)     | 1 0.2%    | 0         |
| 120.910 retinal detachment without dialysis               | 1 0.2%    | 0         |
| <b>OPTIC NERVE</b>  |           |           |
| 130.150 optic disc coloboma                               | 1 0.2%    | 0         |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 5 0.9%    | 0         |
| 900.100 other, not inherited                              | 12 2.1%   | 0         |
| 900.110 other. suspect not inherited/significance unknown | 11 1.9%   | 3 5.5%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 258 45.0% | 35 63.6%  |

# BOERBOEL

|    | DISORDER                     | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC<br>TEST                  |
|----|------------------------------|------------------------|-----------|--------------------|----------------------------------|
| A. | Multifocal<br>retinopathy    | Autosomal<br>recessive | 2         | Breeder option     | Mutation in<br><i>BEST1</i> gene |
| B. | Retinal<br>dysplasia - folds | Not defined            | 1         | Breeder option     |                                  |

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## Description and Comments

### A. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (*cmr1*) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous sub-retinal fluid or accumulation of sub-retinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff.

### B. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

2. Zangerl B, Wickstrom K, Slavik J, et al. Assessment of canine BEST1 variations identifies new mutations and establishes an independent bestrophinopathy model (cmr3). *Mol Vis.* 2010;16:2791-2804.

# OCULAR DISORDERS REPORT BOERBOEL

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 20.160          | macropalpebral fissure   | 1         | 4.5%  | 0         |       |
| 21.000          | entropion, unspecified   | 0         |       | 3         | 4.5%  |
| 22.000          | ectropion, unspecified   | 1         | 4.5%  | 0         |       |
| 25.110          | distichiasis   | 3         | 13.6% | 1         | 1.5%  |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.220          | pigmentary keratitis   | 0         |       | 1         | 1.5%  |
| 70.700          | corneal dystrophy  | 0         |       | 1         | 1.5%  |
| 70.730          | corneal endothelial degeneration                               | 0         |       | 1         | 1.5%  |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                   | 1         | 4.5%  | 0         |       |
| 93.720          | persistent pupillary membranes, iris to lens                   | 0         |       | 1         | 1.5%  |
| 93.730          | persistent pupillary membranes, iris to cornea                 | 0         |       | 2         | 3.0%  |
| 93.760          | persistent pupillary membranes, endothelial opacity/no strands | 1         | 4.5%  | 1         | 1.5%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown           | 2         | 9.1%  | 2         | 3.0%  |
| 100.301         | punctate cataract, anterior cortex                             | 0         |       | 1         | 1.5%  |
| 100.302         | punctate cataract, posterior cortex                            | 0         |       | 1         | 1.5%  |
| 100.312         | incipient cataract, posterior cortex                           | 0         |       | 1         | 1.5%  |
| 100.315         | incipient cataract, posterior sutures                          | 0         |       | 1         | 1.5%  |
| 100.345         | <i>significant cataracts (summary)</i>                         | 0         |       | 4         | 6.1%  |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.120         | persistent hyaloid artery/remnant                              | 0         |       | 2         | 3.0%  |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                                       | 0         |       | 5         | 7.6%  |
| 120.180         | retinal dysplasia, geographic                                  | 1         | 4.5%  | 0         |       |
| 120.960         | retinopathy  | 0         |       | 1         | 1.5%  |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.110         | other. suspect not inherited/significance unknown              | 0         |       | 5         | 7.6%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 15        | 68.2% | 46        | 69.7% |

# BOLOGNESE

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Distichiasis                                     | Not defined | 1         | Breeder option  |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Bolognese breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT BOLOGNESE

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|---------------------|--|-----------|-------|-----------|-------|
|                     |  | #         | %     | #         | %     |
| <b>EYELIDS</b>      |  |           |       |           |       |
| 21.000              | entropion, unspecified   | 3         | 0.4%  | 0         |       |
| 25.110              | distichiasis   | 106       | 15.0% | 4         | 3.6%  |
| <b>NASOLACRIMAL</b> |  |           |       |           |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 2         | 0.3%  | 0         |       |
| 40.910              | keratoconjunctivitis sicca                                     | 2         | 0.3%  | 0         |       |
| <b>NICTITANS</b>    |  |           |       |           |       |
| 52.110              | prolapsed gland of the third eyelid                            | 2         | 0.3%  | 0         |       |
| <b>CORNEA</b>       |  |           |       |           |       |
| 70.700              | corneal dystrophy  | 14        | 2.0%  | 2         | 1.8%  |
| <b>UVEA</b>         |  |           |       |           |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 99        | 14.0% | 24        | 21.6% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 6         | 0.8%  | 0         |       |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 4         | 0.6%  | 0         |       |
| <b>LENS</b>         |  |           |       |           |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 18        | 2.5%  | 2         | 1.8%  |
| 100.305             | punctate cataract, posterior sutures                           | 1         | 0.1%  | 0         |       |
| 100.311             | incipient cataract, anterior cortex                            | 2         | 0.3%  | 0         |       |
| 100.312             | incipient cataract, posterior cortex                           | 2         | 0.3%  | 0         |       |
| 100.313             | incipient cataract, equatorial cortex                          | 3         | 0.4%  | 0         |       |
| 100.314             | incipient cataract, anterior sutures                           | 0         |       | 1         | 0.9%  |
| 100.315             | incipient cataract, posterior sutures                          | 7         | 1.0%  | 0         |       |
| 100.317             | incipient cataract, capsular                                   | 1         | 0.1%  | 0         |       |
| 100.330             | generalized/complete cataract                                  | 4         | 0.6%  | 0         |       |
| 100.345             | significant cataracts (summary)                                | 20        | 2.8%  | 1         | 0.9%  |
| <b>VITREOUS</b>     |  |           |       |           |       |
| 110.120             | persistent hyaloid artery/remnant                              | 0         |       | 1         | 0.9%  |
| 110.135             | PHPV/PTVL  | 0         |       | 1         | 0.9%  |
| 110.200             | vitreous degeneration-anterior chamber                         | 2         | 0.3%  | 0         |       |
| 110.320             | vitreal degeneration   | 12        | 1.7%  | 0         |       |
| <b>RETINA</b>       |  |           |       |           |       |
| 120.170             | retinal dysplasia, folds                                       | 6         | 0.8%  | 0         |       |
| 120.180             | retinal dysplasia, geographic                                  | 0         |       | 1         | 0.9%  |
| 120.190             | retinal dysplasia, detached                                    | 1         | 0.1%  | 0         |       |
| 120.310             | generalized progressive retinal atrophy (PRA)                  | 1         | 0.1%  | 0         |       |
| 120.910             | retinal detachment without dialysis                            | 1         | 0.1%  | 0         |       |
| <b>OPTIC NERVE</b>  |  |           |       |           |       |
| 130.110             | micropapilla   | 1         | 0.1%  | 0         |       |
| <b>OTHER</b>        |  |           |       |           |       |
| 900.000             | other, unspecified   | 19        | 2.7%  | 0         |       |
| 900.100             | other, not inherited   | 20        | 2.8%  | 0         |       |

| <b>OTHER CONTINUED</b>                                    | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 900.110 other. suspect not inherited/significance unknown | 7 1.0%           | 1 0.9%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 515 72.7%        | 75 67.6%         |

## BORDER COLLIE

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|--|---------------------|-----------|-----------------|--------------------------------------|
| A. | Glaucoma – POAG  | Autosomal recessive | 2, 3      | NO              | Mutation in the <i>ADAMTS17</i> gene |
| B. | Corneal dystrophy<br>- epithelial/stromal  | Not defined         | 1         | Breeder option  |                                      |
| C. | Persistent pupillary membranes<br>- iris to iris   | Not defined         | 1         | Breeder option  |                                      |
| D. | Cataract   | Not defined         | 1         | NO              |                                      |
| E. | Y suture tip opacity   | Not defined         | 1         | Breeder option  |                                      |
| F. | Lens luxation  | Autosomal recessive | 1, 3      | NO              | Mutation in the <i>ADAMTS17</i> gene |
| G. | Vitreous degeneration  | Not defined         | 1         | Breeder option  |                                      |
| H. | Retinal atrophy<br>- generalized   | Suggested X-linked  | 1, 4      | NO              |                                      |
| I. | Choroidal hypoplasia (Collie Eye Anomaly)<br>- optic Nerve coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- staphyloma/coloboma | Autosomal recessive | 5-7       | NO              | Mutation in the <i>NHEJ1</i> gene    |

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### Description and Comments

#### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure which, when sustained even for a brief period of time, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening exam.

POAG in the Basset Hound is caused by a 19 base pair deletion in exon 2 of *ADAMTS17*. This deletion alters the reading frame and is suspected to cause a truncated protein. The



trait shows an autosomal recessive mode of inheritance. A DNA test is available.

B. Corneal Dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

C. Persistent pupillary membranes (PPMs)

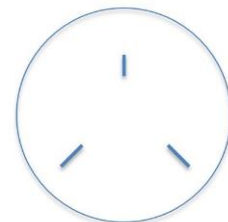
Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

F. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness.

G. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

H. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

I. Choroidal hypoplasia (Collie Eye Anomaly)

- Staphyloma/coloboma
- Retinal detachment
- Retinal hemorrhage
- Optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

**Historical Note:**

Central progressive retinal atrophy was previously a condition listed for this breed. However as the condition is no longer identified in the breed, the condition has been removed. Central progressive retinal atrophy was a progressive retinal degeneration in which photoreceptor death occurred secondary to disease of the underlying pigment epithelium. Progression was slow and some animals never lost vision. CPRA occurred in England, but was uncommon elsewhere.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Foster SJ, Curtis R, Barnett KC. Primary lens luxation in the Border Collie. *J Small Anim Pract.* 1986;27:1-6.
3. Gould D, Pettitt L, McLaughlin B, et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Veterinary Ophthalmology* 2011;14:378-384.
4. Vilboux T, Chaudieu G, Jeannin P, et al. Progressive retinal atrophy in the Border Collie: a new XLPRA. *BMC Vet Res.* 2008;4:10.
5. Bedford PG. Collie eye anomaly in the Border Collie. *Vet Rec.* 1982;111:34-35.
6. Parker HG, Kukekova AV, Akey DT, et al. Breed relationships facilitate fine-mapping studies: a 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. *Gen Res.* 2007;17:1562-1571.
7. Lowe JK, Kukekova AV, Kirkness EF, et al. Linkage mapping of the primary disease locus for collie eye anomaly. *Genomics.* 2003;82:86-95.

# OCULAR DISORDERS REPORT BORDER COLLIE

| Diagnostic Name  | TOTAL DOGS EXAMINED  |       | 1991-2015<br>25,576 |     | 2016-2020<br>2,893 |   |
|------------------|--|-------|---------------------|-----|--------------------|---|
|                  | #  | %     | #                   | %   | #                  | % |
| <b>GLOBE</b>     |  |       |                     |     |                    |   |
| 0.110            | microphthalmia   | 13    | 0.1%                | 1   | 0.0%               |   |
| 10.000           | glaucoma   | 1     | 0.0%                | 0   |                    |   |
| <b>EYELIDS</b>   |  |       |                     |     |                    |   |
| 21.000           | entropion, unspecified   | 2     | 0.0%                | 0   |                    |   |
| 25.110           | distichiasis   | 122   | 0.5%                | 17  | 0.6%               |   |
| <b>NICTITANS</b> |  |       |                     |     |                    |   |
| 50.210           | pannus of third eyelid   | 0     |                     | 1   | 0.0%               |   |
| 51.100           | third eyelid cartilage anomaly                                 | 5     | 0.0%                | 2   | 0.1%               |   |
| <b>CORNEA</b>    |  |       |                     |     |                    |   |
| 70.210           | corneal pannus   | 17    | 0.1%                | 5   | 0.2%               |   |
| 70.220           | pigmentary keratitis   | 1     | 0.0%                | 0   |                    |   |
| 70.700           | corneal dystrophy  | 197   | 0.8%                | 48  | 1.7%               |   |
| 70.730           | corneal endothelial degeneration                               | 4     | 0.0%                | 1   | 0.0%               |   |
| <b>UVEA</b>      |  |       |                     |     |                    |   |
| 90.250           | pigmentary uveitis   | 1     | 0.0%                | 0   |                    |   |
| 93.110           | iris hypoplasia  | 1     | 0.0%                | 1   | 0.0%               |   |
| 93.140           | corneal endothelial pigment without PPM                        | 2     | 0.0%                | 0   |                    |   |
| 93.150           | iris coloboma  | 8     | 0.0%                | 0   |                    |   |
| 93.710           | persistent pupillary membranes, iris to iris                   | 1,591 | 6.2%                | 215 | 7.4%               |   |
| 93.720           | persistent pupillary membranes, iris to lens                   | 31    | 0.1%                | 8   | 0.3%               |   |
| 93.730           | persistent pupillary membranes, iris to cornea                 | 34    | 0.1%                | 1   | 0.0%               |   |
| 93.740           | persistent pupillary membranes, iris sheets                    | 15    | 0.1%                | 1   | 0.0%               |   |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands   | 8     | 0.0%                | 12  | 0.4%               |   |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands | 3     | 0.0%                | 2   | 0.1%               |   |
| 93.810           | uveal melanoma   | 0     |                     | 1   | 0.0%               |   |
| 93.999           | uveal cysts  | 10    | 0.0%                | 2   | 0.1%               |   |
| 97.150           | chorioretinal coloboma, congenital                             | 2     | 0.0%                | 1   | 0.0%               |   |
| <b>LENS</b>      |  |       |                     |     |                    |   |
| 100.200          | cataract, unspecified  | 57    | 0.2%                | 0   |                    |   |
| 100.210          | cataract, suspect not inherited/significance unknown           | 1,198 | 4.7%                | 162 | 5.6%               |   |
| 100.301          | punctate cataract, anterior cortex                             | 108   | 0.4%                | 17  | 0.6%               |   |
| 100.302          | punctate cataract, posterior cortex                            | 63    | 0.2%                | 3   | 0.1%               |   |
| 100.303          | punctate cataract, equatorial cortex                           | 46    | 0.2%                | 6   | 0.2%               |   |
| 100.304          | punctate cataract, anterior sutures                            | 5     | 0.0%                | 2   | 0.1%               |   |
| 100.305          | punctate cataract, posterior sutures                           | 109   | 0.4%                | 49  | 1.7%               |   |
| 100.306          | punctate cataract, nucleus                                     | 25    | 0.1%                | 7   | 0.2%               |   |
| 100.307          | punctate cataract, capsular                                    | 27    | 0.1%                | 13  | 0.4%               |   |
| 100.311          | incipient cataract, anterior cortex                            | 138   | 0.5%                | 12  | 0.4%               |   |
| 100.312          | incipient cataract, posterior cortex                           | 101   | 0.4%                | 13  | 0.4%               |   |
| 100.313          | incipient cataract, equatorial cortex                          | 115   | 0.4%                | 19  | 0.7%               |   |
| 100.314          | incipient cataract, anterior sutures                           | 12    | 0.0%                | 0   |                    |   |
| 100.315          | incipient cataract, posterior sutures                          | 52    | 0.2%                | 17  | 0.6%               |   |
| 100.316          | incipient cataract, nucleus                                    | 25    | 0.1%                | 12  | 0.4%               |   |
| 100.317          | incipient cataract, capsular                                   | 25    | 0.1%                | 3   | 0.1%               |   |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.321 incomplete cataract, anterior cortex              | 4                | 0.0%  | 7                | 0.2%  |
| 100.322 incomplete cataract, posterior cortex             | 1                | 0.0%  | 5                | 0.2%  |
| 100.323 incomplete cataract, equatorial cortex            | 2                | 0.0%  | 2                | 0.1%  |
| 100.326 incomplete cataract, nucleus                      | 0                |       | 2                | 0.1%  |
| 100.327 incomplete cataract, capsular                     | 1                | 0.0%  | 0                |       |
| 100.328 y-suture tip opacities                            | 19               | 0.1%  | 65               | 2.2%  |
| 100.330 generalized/complete cataract                     | 29               | 0.1%  | 1                | 0.0%  |
| 100.340 resorbing/hypermature cataract                    | 0                |       | 1                | 0.0%  |
| 100.345 <i>significant cataracts (summary)</i>            | 964              | 3.8%  | 256              | 8.8%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 14               | 0.1%  | 0                |       |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 66               | 0.3%  | 3                | 0.1%  |
| 110.135 PHPV/PTVL   | 19               | 0.1%  | 1                | 0.0%  |
| 110.200 vitreous degeneration-anterior chamber            | 3                | 0.0%  | 6                | 0.2%  |
| 110.320 vitreal degeneration                              | 162              | 0.6%  | 21               | 0.7%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 431              | 1.7%  | 40               | 1.4%  |
| 97.120 coloboma   | 48               | 0.2%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 196              | 0.8%  | 16               | 0.6%  |
| 120.180 retinal dysplasia, geographic                     | 16               | 0.1%  | 1                | 0.0%  |
| 120.310 generalized progressive retinal atrophy (PRA)     | 226              | 0.9%  | 17               | 0.6%  |
| 120.400 retinal hemorrhage                                | 6                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 18               | 0.1%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 0                |       | 1                | 0.0%  |
| 120.960 retinopathy                                       | 15               | 0.1%  | 8                | 0.3%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 19               | 0.1%  | 4                | 0.1%  |
| 130.120 optic nerve hypoplasia                            | 18               | 0.1%  | 1                | 0.0%  |
| 130.150 optic disc coloboma                               | 91               | 0.4%  | 6                | 0.2%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 214              | 0.8%  | 0                |       |
| 900.100 other, not inherited                              | 607              | 2.4%  | 1                | 0.0%  |
| 900.110 other. suspect not inherited/significance unknown | 212              | 0.8%  | 140              | 4.8%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 21,384           | 83.6% | 2,078            | 71.8% |

# BORDER TERRIER

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| B. | Cataract   | Not defined | 1         | NO              |
| C. | Y suture tip opacity                             | Not defined | 1         | Breeder option  |

## Description and Comments

### A. Persistent pupillary membranes (PPMs)

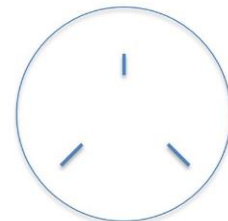
Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Border Terrier breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT BORDER TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>5,864 |      | 2016-2020<br>1,618 |       |
|---------------------|--|--------------------|------|--------------------|-------|
|                     |  | #                  | %    | #                  | %     |
| <b>EYELIDS</b>      |  |                    |      |                    |       |
| 21.000              | entropion, unspecified   | 3                  | 0.1% | 0                  |       |
| 25.110              | distichiasis   | 44                 | 0.8% | 11                 | 0.7%  |
| <b>NASOLACRIMAL</b> |  |                    |      |                    |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 0                  |      | 2                  | 0.1%  |
| <b>NICTITANS</b>    |  |                    |      |                    |       |
| 52.110              | prolapsed gland of the third eyelid                            | 1                  | 0.0% | 0                  |       |
| <b>CORNEA</b>       |  |                    |      |                    |       |
| 70.700              | corneal dystrophy  | 12                 | 0.2% | 4                  | 0.2%  |
| <b>UVEA</b>         |  |                    |      |                    |       |
| 93.140              | corneal endothelial pigment without PPM                        | 1                  | 0.0% | 0                  |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 154                | 2.6% | 69                 | 4.3%  |
| 93.720              | persistent pupillary membranes, iris to lens                   | 1                  | 0.0% | 0                  |       |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 3                  | 0.1% | 0                  |       |
| 93.740              | persistent pupillary membranes, iris sheets                    | 2                  | 0.0% | 0                  |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 1                  | 0.0% | 1                  | 0.1%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 1                  | 0.0% | 0                  |       |
| 93.999              | uveal cysts  | 1                  | 0.0% | 0                  |       |
| <b>LENS</b>         |  |                    |      |                    |       |
| 100.200             | cataract, unspecified  | 9                  | 0.2% | 0                  |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 359                | 6.1% | 150                | 9.3%  |
| 100.301             | punctate cataract, anterior cortex                             | 31                 | 0.5% | 11                 | 0.7%  |
| 100.302             | punctate cataract, posterior cortex                            | 19                 | 0.3% | 5                  | 0.3%  |
| 100.303             | punctate cataract, equatorial cortex                           | 17                 | 0.3% | 4                  | 0.2%  |
| 100.304             | punctate cataract, anterior sutures                            | 2                  | 0.0% | 1                  | 0.1%  |
| 100.305             | punctate cataract, posterior sutures                           | 12                 | 0.2% | 16                 | 1.0%  |
| 100.306             | punctate cataract, nucleus                                     | 5                  | 0.1% | 0                  |       |
| 100.307             | punctate cataract, capsular                                    | 5                  | 0.1% | 9                  | 0.6%  |
| 100.311             | incipient cataract, anterior cortex                            | 55                 | 0.9% | 16                 | 1.0%  |
| 100.312             | incipient cataract, posterior cortex                           | 46                 | 0.8% | 15                 | 0.9%  |
| 100.313             | incipient cataract, equatorial cortex                          | 63                 | 1.1% | 22                 | 1.4%  |
| 100.314             | incipient cataract, anterior sutures                           | 2                  | 0.0% | 2                  | 0.1%  |
| 100.315             | incipient cataract, posterior sutures                          | 12                 | 0.2% | 8                  | 0.5%  |
| 100.316             | incipient cataract, nucleus                                    | 13                 | 0.2% | 0                  |       |
| 100.317             | incipient cataract, capsular                                   | 8                  | 0.1% | 3                  | 0.2%  |
| 100.321             | incomplete cataract, anterior cortex                           | 3                  | 0.1% | 6                  | 0.4%  |
| 100.322             | incomplete cataract, posterior cortex                          | 4                  | 0.1% | 7                  | 0.4%  |
| 100.323             | incomplete cataract, equatorial cortex                         | 2                  | 0.0% | 3                  | 0.2%  |
| 100.326             | incomplete cataract, nucleus                                   | 1                  | 0.0% | 0                  |       |
| 100.327             | incomplete cataract, capsular                                  | 1                  | 0.0% | 0                  |       |
| 100.328             | y-suture tip opacities   | 10                 | 0.2% | 45                 | 2.8%  |
| 100.330             | generalized/complete cataract                                  | 19                 | 0.3% | 3                  | 0.2%  |
| 100.340             | resorbing/hypermature cataract                                 | 1                  | 0.0% | 2                  | 0.1%  |
| 100.345             | significant cataracts (summary)                                | 340                | 5.8% | 178                | 11.0% |
| 100.375             | subluxation/luxation, unspecified                              | 1                  | 0.0% | 0                  |       |



|   | 1991-2015   | 2016-2020   |
|---|-------------|-------------|
| <b>VITREOUS</b>   |             |             |
| 110.120 persistent hyaloid artery/remnant                 | 5 0.1%      | 5 0.3%      |
| 110.200 vitreous degeneration-anterior chamber            | 5 0.1%      | 5 0.3%      |
| 110.320 vitreal degeneration                              | 56 1.0%     | 14 0.9%     |
| <b>FUNDUS</b>   |             |             |
| 97.110 choroidal hypoplasia                               | 1 0.0%      | 0           |
| 97.120 coloboma   | 1 0.0%      | 0           |
| <b>RETINA</b>   |             |             |
| 120.170 retinal dysplasia, folds                          | 12 0.2%     | 5 0.3%      |
| 120.180 retinal dysplasia, geographic                     | 8 0.1%      | 0           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 11 0.2%     | 2 0.1%      |
| 120.910 retinal detachment without dialysis               | 1 0.0%      | 0           |
| 120.960 retinopathy                                       | 0           | 5 0.3%      |
| <b>OPTIC NERVE</b>  |             |             |
| 130.110 micropapilla                                      | 0           | 1 0.1%      |
| 130.120 optic nerve hypoplasia                            | 1 0.0%      | 0           |
| <b>OTHER</b>  |             |             |
| 900.000 other, unspecified                                | 56 1.0%     | 0           |
| 900.100 other, not inherited                              | 125 2.1%    | 5 0.3%      |
| 900.110 other. suspect not inherited/significance unknown | 52 0.9%     | 76 4.7%     |
| <b>NORMAL</b>   |             |             |
| 0.000 normal globe  | 5,207 88.8% | 1,208 74.7% |

# BORZOI

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |
| B. | Cataract   | Not defined        | 1                | NO                     |
| C. | Retinopathy                                      | Not defined        | 2                | Breeder option         |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Retinopathy

Patchy focal unilateral or bilateral hyper reflective tapetal lesions most frequently peripheral but occasionally central around a pigmented spot, usually non progressive. Not usually present prior to 3 months of age but usually present by 18 months of age.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Storey ES, Grahn BH and Alcorn J. Multifocal chorioretinal lesions in Borzoi dogs. *Vet Ophthalmol.* 2005 Sep-Oct;8:337-347.

# OCULAR DISORDERS REPORT BORZOI

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015<br>3,277 |      | 2016-2020<br>962 |      |
|------------------|--|--------------------|------|------------------|------|
|                  |  | #                  | %    | #                | %    |
| <b>GLOBE</b>     |  |                    |      |                  |      |
| 0.110            | microphthalmia   | 7                  | 0.2% | 0                |      |
| <b>EYELIDS</b>   |  |                    |      |                  |      |
| 20.160           | macropalpebral fissure   | 1                  | 0.0% | 0                |      |
| 25.110           | distichiasis   | 9                  | 0.3% | 2                | 0.2% |
| <b>NICTITANS</b> |  |                    |      |                  |      |
| 50.210           | pannus of third eyelid   | 0                  |      | 1                | 0.1% |
| 51.100           | third eyelid cartilage anomaly                                 | 1                  | 0.0% | 2                | 0.2% |
| <b>CORNEA</b>    |  |                    |      |                  |      |
| 70.210           | corneal pannus   | 17                 | 0.5% | 2                | 0.2% |
| 70.220           | pigmentary keratitis   | 0                  |      | 2                | 0.2% |
| 70.700           | corneal dystrophy  | 16                 | 0.5% | 0                |      |
| 70.730           | corneal endothelial degeneration                               | 1                  | 0.0% | 0                |      |
| <b>UVEA</b>      |  |                    |      |                  |      |
| 93.710           | persistent pupillary membranes, iris to iris                   | 71                 | 2.2% | 10               | 1.0% |
| 93.720           | persistent pupillary membranes, iris to lens                   | 6                  | 0.2% | 0                |      |
| 93.730           | persistent pupillary membranes, iris to cornea                 | 11                 | 0.3% | 0                |      |
| 93.740           | persistent pupillary membranes, iris sheets                    | 1                  | 0.0% | 0                |      |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands   | 0                  |      | 3                | 0.3% |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands | 1                  | 0.0% | 1                | 0.1% |
| 93.810           | uveal melanoma   | 0                  |      | 5                | 0.5% |
| 93.999           | uveal cysts  | 6                  | 0.2% | 4                | 0.4% |
| <b>LENS</b>      |  |                    |      |                  |      |
| 100.200          | cataract, unspecified  | 2                  | 0.1% | 0                |      |
| 100.210          | cataract. suspect not inherited/significance unknown           | 102                | 3.1% | 24               | 2.5% |
| 100.301          | punctate cataract, anterior cortex                             | 5                  | 0.2% | 3                | 0.3% |
| 100.302          | punctate cataract, posterior cortex                            | 9                  | 0.3% | 3                | 0.3% |
| 100.303          | punctate cataract, equatorial cortex                           | 0                  |      | 1                | 0.1% |
| 100.304          | punctate cataract, anterior sutures                            | 2                  | 0.1% | 0                |      |
| 100.305          | punctate cataract, posterior sutures                           | 8                  | 0.2% | 2                | 0.2% |
| 100.306          | punctate cataract, nucleus                                     | 1                  | 0.0% | 0                |      |
| 100.307          | punctate cataract, capsular                                    | 4                  | 0.1% | 1                | 0.1% |
| 100.311          | incipient cataract, anterior cortex                            | 11                 | 0.3% | 2                | 0.2% |
| 100.312          | incipient cataract, posterior cortex                           | 18                 | 0.5% | 1                | 0.1% |
| 100.313          | incipient cataract, equatorial cortex                          | 2                  | 0.1% | 2                | 0.2% |
| 100.314          | incipient cataract, anterior sutures                           | 2                  | 0.1% | 0                |      |
| 100.315          | incipient cataract, posterior sutures                          | 1                  | 0.0% | 0                |      |
| 100.316          | incipient cataract, nucleus                                    | 1                  | 0.0% | 0                |      |
| 100.317          | incipient cataract, capsular                                   | 4                  | 0.1% | 3                | 0.3% |
| 100.324          | incomplete cataract, anterior sutures                          | 1                  | 0.0% | 0                |      |
| 100.328          | y-suture tip opacities   | 3                  | 0.1% | 2                | 0.2% |
| 100.330          | generalized/complete cataract                                  | 7                  | 0.2% | 1                | 0.1% |
| 100.340          | resorbing/hypermature cataract                                 | 0                  |      | 1                | 0.1% |
| 100.345          | significant cataracts (summary)                                | 81                 | 2.5% | 22               | 2.3% |
| 100.375          | subluxation/luxation, unspecified                              | 4                  | 0.1% | 0                |      |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>VITREOUS</b>   |             |           |
| 110.120 persistent hyaloid artery/remnant                 | 12 0.4%     | 2 0.2%    |
| 110.135 PHPV/PTVL   | 11 0.3%     | 0         |
| 110.200 vitreous degeneration-anterior chamber            | 0           | 4 0.4%    |
| 110.320 vitreal degeneration                              | 9 0.3%      | 3 0.3%    |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 8 0.2%      | 2 0.2%    |
| 120.180 retinal dysplasia, geographic                     | 8 0.2%      | 2 0.2%    |
| 120.190 retinal dysplasia, detached                       | 1 0.0%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 25 0.8%     | 3 0.3%    |
| 120.400 retinal hemorrhage                                | 2 0.1%      | 0         |
| 120.910 retinal detachment without dialysis               | 5 0.2%      | 0         |
| 120.920 retinal detachment with dialysis                  | 2 0.1%      | 0         |
| 120.960 retinopathy                                       | 18 0.5%     | 24 2.5%   |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 10 0.3%     | 8 0.8%    |
| 130.120 optic nerve hypoplasia                            | 14 0.4%     | 2 0.2%    |
| 130.150 optic disc coloboma                               | 3 0.1%      | 1 0.1%    |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 44 1.3%     | 0         |
| 900.100 other, not inherited                              | 109 3.3%    | 1 0.1%    |
| 900.110 other. suspect not inherited/significance unknown | 68 2.1%     | 74 7.7%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 2,849 86.9% | 787 81.8% |

# BOSTON TERRIER

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE                               |
|----|---|------------------------|-----------|--------------------|--|
| A. | Glaucoma  | Not defined            | 1-3       | NO                 |  |
| B. | Distichiasis  | Not defined            | 1         | Breeder option     |  |
| C. | Imperforate<br>lacrimal punctum                     | Not defined            | 1         | Breeder option     |  |
| D. | Corneal dystrophy<br>- epithelial/stromal           | Not defined            | 1         | Breeder option     |  |
| E. | Corneal dystrophy<br>- endothelial                  | Not defined            | 1, 4      | NO                 |  |
| F. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined            | 1         | Breeder option     |  |
| G. | Cataract  | Autosomal<br>recessive | 1, 5-9    | NO                 | Mutation in the<br><i>HSF4</i> gene<br>( <i>HSF4-1</i> ) |
| H. | Vitreous<br>degeneration                            | Not defined            | 1         | Breeder option     |  |

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## Description and Comments

### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of the IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine screening exam for certification.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

C. Imperforate lacrimal punctum

A developmental anomaly resulting in failure of opening of the lacrimal duct located at the medial lid margins. The lower punctum is more frequently affected. This defect usually results in epiphora, an overflow of tears onto the face.

D. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral

E. Corneal dystrophy – endothelial

Corneal endothelial dystrophy is an abnormal loss of the inner lining of the cornea that causes progressive fluid retention (edema). With time the edema results in keratitis and decreased vision. This usually does not occur until the animal is older.

In the Boston Terrier, this is a primary degenerative endothelial disease leading to progressive and permanent corneal edema. It is not known if this disease is an inherited disorder. There is no sex predilection. The condition is observed in older dogs, 6 to 13 years of age with a mean of 9.5 years. The corneal edema starts asymptotically in the dorsal temporal corneal quadrant of one eye and slowly progresses medially, eventually involving the entire cornea. Typically, it becomes bilateral. In the later stages, discomfort, intracorneal bullae with subsequent ulceration and keratoconus may develop.

F. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally during the first three months of life. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

G. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

The Boston Terrier has at least two distinct forms of inherited cataract. One type has an onset before 6 months of age with rapid progression to complete opacity prior to 2 years old. The early onset cataract is inherited as an autosomal recessive mutation in the *HSF4* gene (*HSF4-1*). A DNA test is available. A second type of cataract occurs after 4-5 years of age with variable progression. The genetic mutation responsible for this cataract is not yet known.

H. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Slater MR, Erb HN. Effects of risk factors and prophylactic treatment on primary glaucoma in the dog. *J Am Vet Med Assoc.* 1986;188:1028-1030.
3. Gelatt KN, MacKay EO. Prevalence of the breed-related glaucomas in pure-bred dogs in North America. *Vet Ophthalmol.* 2004;7:97-111.
4. Martin CL, Dice PF. Corneal Endothelial Dystrophy in the Dog. *J Am Anim Hosp Assoc.* 1982;18:327-336.
5. Curtis R. Late-onset cataract in the Boston terrier. *Vet Rec.* 1984;115:577-578.
6. Barnett KC. The diagnosis and differential diagnosis of cataract in the dog. *J Small Anim Pract.* 1985;26:305-316.
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8. Mellersh CS, Graves KT, McLaughlin B, et al. Mutation in HSF4 associated with early but not late-onset hereditary cataract in the Boston Terrier. *J Hered.* 2007;98:531-533.
9. Mellersh CS, Pettitt L, Forman OP, et al. Identification of mutations in HSF4 in dogs of three different breeds with hereditary cataracts. *Vet Ophthalmol.* 2006;9:369-378.

# OCULAR DISORDERS REPORT BOSTON TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>13,491 |      | 2016-2020<br>3,575 |      |
|---------------------|--|---------------------|------|--------------------|------|
|                     |  | #                   | %    | #                  | %    |
| <b>GLOBE</b>        |  |                     |      |                    |      |
| 0.110               | microphthalmia   | 2                   | 0.0% | 3                  | 0.1% |
| 10.000              | glaucoma   | 1                   | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                     |      |                    |      |
| 20.140              | ectopic cilia  | 5                   | 0.0% | 0                  |      |
| 20.160              | macropalpebral fissure                                       | 12                  | 0.1% | 0                  |      |
| 21.000              | entropion, unspecified                                       | 41                  | 0.3% | 16                 | 0.4% |
| 22.000              | ectropion, unspecified                                       | 2                   | 0.0% | 0                  |      |
| 25.110              | distichiasis   | 462                 | 3.4% | 108                | 3.0% |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                       | 29                  | 0.2% | 65                 | 1.8% |
| 40.910              | keratoconjunctivitis sicca                                   | 9                   | 0.1% | 5                  | 0.1% |
| <b>NICTITANS</b>    |  |                     |      |                    |      |
| 50.210              | pannus of third eyelid                                       | 0                   |      | 3                  | 0.1% |
| 51.100              | third eyelid cartilage anomaly                               | 1                   | 0.0% | 0                  |      |
| 52.110              | prolapsed gland of the third eyelid                          | 9                   | 0.1% | 4                  | 0.1% |
| <b>CORNEA</b>       |  |                     |      |                    |      |
| 70.210              | corneal pannus   | 1                   | 0.0% | 0                  |      |
| 70.220              | pigmentary keratitis   | 18                  | 0.1% | 8                  | 0.2% |
| 70.700              | corneal dystrophy  | 322                 | 2.4% | 66                 | 1.8% |
| 70.730              | corneal endothelial degeneration                             | 26                  | 0.2% | 3                  | 0.1% |
| <b>UVEA</b>         |  |                     |      |                    |      |
| 93.110              | iris hypoplasia  | 5                   | 0.0% | 3                  | 0.1% |
| 93.150              | iris coloboma  | 7                   | 0.1% | 1                  | 0.0% |
| 93.710              | persistent pupillary membranes, iris to iris                 | 498                 | 3.7% | 153                | 4.3% |
| 93.720              | persistent pupillary membranes, iris to lens                 | 12                  | 0.1% | 4                  | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea               | 6                   | 0.0% | 1                  | 0.0% |
| 93.740              | persistent pupillary membranes, iris sheets                  | 8                   | 0.1% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands | 1                   | 0.0% | 0                  |      |
| 93.810              | uveal melanoma   | 1                   | 0.0% | 0                  |      |
| 93.999              | uveal cysts  | 27                  | 0.2% | 18                 | 0.5% |
| <b>LENS</b>         |  |                     |      |                    |      |
| 100.200             | cataract, unspecified  | 81                  | 0.6% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown         | 310                 | 2.3% | 72                 | 2.0% |
| 100.301             | punctate cataract, anterior cortex                           | 160                 | 1.2% | 44                 | 1.2% |
| 100.302             | punctate cataract, posterior cortex                          | 46                  | 0.3% | 14                 | 0.4% |
| 100.303             | punctate cataract, equatorial cortex                         | 70                  | 0.5% | 16                 | 0.4% |
| 100.304             | punctate cataract, anterior sutures                          | 33                  | 0.2% | 13                 | 0.4% |
| 100.305             | punctate cataract, posterior sutures                         | 24                  | 0.2% | 5                  | 0.1% |
| 100.306             | punctate cataract, nucleus                                   | 9                   | 0.1% | 1                  | 0.0% |
| 100.307             | punctate cataract, capsular                                  | 20                  | 0.1% | 20                 | 0.6% |
| 100.311             | incipient cataract, anterior cortex                          | 631                 | 4.7% | 114                | 3.2% |
| 100.312             | incipient cataract, posterior cortex                         | 156                 | 1.2% | 26                 | 0.7% |
| 100.313             | incipient cataract, equatorial cortex                        | 292                 | 2.2% | 35                 | 1.0% |
| 100.314             | incipient cataract, anterior sutures                         | 83                  | 0.6% | 13                 | 0.4% |



| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.315 incipient cataract, posterior sutures             | 34               | 0.3%  | 5                | 0.1%  |
| 100.316 incipient cataract, nucleus                       | 18               | 0.1%  | 5                | 0.1%  |
| 100.317 incipient cataract, capsular                      | 17               | 0.1%  | 1                | 0.0%  |
| 100.321 incomplete cataract, anterior cortex              | 25               | 0.2%  | 51               | 1.4%  |
| 100.322 incomplete cataract, posterior cortex             | 11               | 0.1%  | 18               | 0.5%  |
| 100.323 incomplete cataract, equatorial cortex            | 15               | 0.1%  | 9                | 0.3%  |
| 100.324 incomplete cataract, anterior sutures             | 2                | 0.0%  | 1                | 0.0%  |
| 100.325 incomplete cataract, posterior sutures            | 0                |       | 1                | 0.0%  |
| 100.326 incomplete cataract, nucleus                      | 0                |       | 2                | 0.1%  |
| 100.328 y-suture tip opacities                            | 7                | 0.1%  | 7                | 0.2%  |
| 100.330 generalized/complete cataract                     | 92               | 0.7%  | 8                | 0.2%  |
| 100.340 resorbing/hypermature cataract                    | 1                | 0.0%  | 0                |       |
| 100.345 <i>significant cataracts (summary)</i>            | 1,827            | 13.5% | 409              | 11.4% |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 13               | 0.1%  | 5                | 0.1%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 44               | 0.3%  | 20               | 0.6%  |
| 110.135 PHPV/PTVL   | 9                | 0.1%  | 4                | 0.1%  |
| 110.200 vitreous degeneration-anterior chamber            | 6                | 0.0%  | 7                | 0.2%  |
| 110.320 vitreal degeneration                              | 176              | 1.3%  | 25               | 0.7%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 2                | 0.0%  | 2                | 0.1%  |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 34               | 0.3%  | 5                | 0.1%  |
| 120.180 retinal dysplasia, geographic                     | 12               | 0.1%  | 5                | 0.1%  |
| 120.190 retinal dysplasia, detached                       | 4                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 11               | 0.1%  | 0                |       |
| 120.400 retinal hemorrhage                                | 3                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 1                | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 1                | 0.0%  | 0                |       |
| 120.960 retinopathy                                       | 3                | 0.0%  | 2                | 0.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 1                | 0.0%  | 0                |       |
| 130.120 optic nerve hypoplasia                            | 2                | 0.0%  | 1                | 0.0%  |
| 130.150 optic disc coloboma                               | 0                |       | 1                | 0.0%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 165              | 1.2%  | 0                |       |
| 900.100 other, not inherited                              | 380              | 2.8%  | 3                | 0.1%  |
| 900.110 other. suspect not inherited/significance unknown | 174              | 1.3%  | 199              | 5.6%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 10,916           | 80.9% | 2,667            | 74.6% |

# BOUVIER DES FLANDRES

|    | DISORDER  | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|---|-------------|-----------|-------------------------|
| A. | Glaucoma  | Not defined | 1-3       | NO                      |
| B. | Persistent pupillary membranes  |             |           |                         |
|    | - iris to iris  | Not defined | 1         | Breeder option          |
|    | - lens pigment foci/no strands  | Not defined | 1         | Passes with no notation |
| C. | Cataract  | Not defined | 1         | NO                      |
| D. | Y-suture tip opacity  | Not defined | 1         | Breeder option          |
| E. | Persistent hyperplastic primary vitreous/Persistent hyperplastic tunica vasculosa lentis (PHPV/PHTVL) | Not defined | 1, 4      | NO                      |

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## Description and Comments

### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of the IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine screening exam for certification.

In this breed, primary glaucoma is associated with narrowed iridocorneal angles and various degrees of congenital angle malformations varying from mild to severe. Dysplastic pectinate ligaments and subsequent narrowed angles are similar to those described in the Basset Hound and American and English Cocker Spaniels. The occurrence of glaucoma is related to the most severe abnormalities of the pectinate ligaments. The relationship between glaucoma development and the anomaly of the pectinate ligament is not clear.

A recent study evaluated risk factors for development of glaucoma in the Bouvier des Flandres. A narrow angle with dysplastic pectinate ligaments on gonioscopy and/or presence of a narrow or closed ciliary cleft on high resolution ultrasound were associated with development of primary glaucoma in the breed.

B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

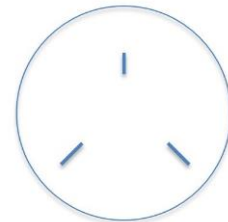
Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

E. Persistent hyperplastic primary vitreous (PHPV)/Persistent hyperplastic tunica vasculosa lentis (PHTVL)

Persistent hyperplastic primary vitreous is a congenital defect resulting from abnormalities in the development and regression of the hyaloid artery (the primary vitreous) and the

interaction of this blood vessel with the posterior lens capsule/cortex during embryogenesis. This condition is often associated with persistent hyperplastic tunica vasculosa lentis which results from failure of regression of the embryologic vascular network which surrounds the developing lens.

In the Bouvier des Flandres, the condition is associated with retinal dysplasia and detachment, optic nerve hypoplasia, lenticonus, cataract and congenital blindness.

## References

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2. van der Linde-Sipman JS. Dysplasia of the pectinate ligament and primary glaucoma in the Bouvier des Flandres dog. *Vet Pathol.* 1987;24:201-206.
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# OCULAR DISORDERS REPORT BOUVIER DES FLANDRES

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |       |
|---|---------------------|-----------|------|-----------|-------|
|   |                     | #         | %    | #         | %     |
| <b>GLOBE</b>  |                     |           |      |           |       |
| 10.000 glaucoma   |                     | 1         | 0.0% | 1         | 0.1%  |
| <b>EYELIDS</b>  |                     |           |      |           |       |
| 20.160 macropalpebral fissure   |                     | 1         | 0.0% | 0         |       |
| 21.000 entropion, unspecified   |                     | 28        | 0.6% | 4         | 0.5%  |
| 22.000 ectropion, unspecified   |                     | 6         | 0.1% | 0         |       |
| 25.110 distichiasis   |                     | 42        | 0.8% | 6         | 0.7%  |
| <b>CORNEA</b>   |                     |           |      |           |       |
| 70.210 corneal pannus   |                     | 1         | 0.0% | 0         |       |
| 70.220 pigmentary keratitis   |                     | 1         | 0.0% | 1         | 0.1%  |
| 70.700 corneal dystrophy  |                     | 31        | 0.6% | 2         | 0.2%  |
| 70.730 corneal endothelial degeneration                               |                     | 4         | 0.1% | 0         |       |
| <b>UVEA</b>   |                     |           |      |           |       |
| 93.180 liris sphincter dysplasia                                      |                     | 0         |      | 1         | 0.1%  |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 417       | 8.3% | 80        | 9.6%  |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 11        | 0.2% | 0         |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 6         | 0.1% | 1         | 0.1%  |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 7         | 0.1% | 1         | 0.1%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 16        | 0.3% | 13        | 1.6%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 2         | 0.0% | 0         |       |
| 93.810 uveal melanoma   |                     | 1         | 0.0% | 0         |       |
| 93.999 uveal cysts  |                     | 13        | 0.3% | 8         | 1.0%  |
| <b>LENS</b>   |                     |           |      |           |       |
| 100.200 cataract, unspecified   |                     | 5         | 0.1% | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 414       | 8.2% | 96        | 11.5% |
| 100.301 punctate cataract, anterior cortex                            |                     | 30        | 0.6% | 9         | 1.1%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 38        | 0.8% | 5         | 0.6%  |
| 100.303 punctate cataract, equatorial cortex                          |                     | 4         | 0.1% | 2         | 0.2%  |
| 100.304 punctate cataract, anterior sutures                           |                     | 5         | 0.1% | 3         | 0.4%  |
| 100.305 punctate cataract, posterior sutures                          |                     | 31        | 0.6% | 8         | 1.0%  |
| 100.306 punctate cataract, nucleus                                    |                     | 9         | 0.2% | 2         | 0.2%  |
| 100.307 punctate cataract, capsular                                   |                     | 19        | 0.4% | 5         | 0.6%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 18        | 0.4% | 5         | 0.6%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 97        | 1.9% | 10        | 1.2%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 20        | 0.4% | 3         | 0.4%  |
| 100.314 incipient cataract, anterior sutures                          |                     | 0         |      | 1         | 0.1%  |
| 100.315 incipient cataract, posterior sutures                         |                     | 23        | 0.5% | 4         | 0.5%  |
| 100.316 incipient cataract, nucleus                                   |                     | 32        | 0.6% | 5         | 0.6%  |
| 100.317 incipient cataract, capsular                                  |                     | 10        | 0.2% | 4         | 0.5%  |
| 100.321 incomplete cataract, anterior cortex                          |                     | 1         | 0.0% | 3         | 0.4%  |
| 100.322 incomplete cataract, posterior cortex                         |                     | 2         | 0.0% | 2         | 0.2%  |
| 100.326 incomplete cataract, nucleus                                  |                     | 1         | 0.0% | 1         | 0.1%  |
| 100.328 y-suture tip opacities  |                     | 9         | 0.2% | 27        | 3.2%  |
| 100.330 generalized/complete cataract                                 |                     | 31        | 0.6% | 0         |       |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 385       | 7.7% | 99        | 11.9% |
| 100.375 <i>subluxation/luxation, unspecified</i>                      |                     | 2         | 0.0% | 0         |       |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>VITREOUS</b>   |             |           |
| 110.120 persistent hyaloid artery/remnant                 | 8 0.2%      | 7 0.8%    |
| 110.135 PHPV/PTVL   | 6 0.1%      | 0         |
| 110.200 vitreous degeneration-anterior chamber            | 0           | 1 0.1%    |
| 110.320 vitreal degeneration                              | 11 0.2%     | 2 0.2%    |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 35 0.7%     | 1 0.1%    |
| 120.180 retinal dysplasia, geographic                     | 3 0.1%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 14 0.3%     | 0         |
| 120.960 retinopathy                                       | 0           | 1 0.1%    |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 2 0.0%      | 1 0.1%    |
| 130.120 optic nerve hypoplasia                            | 1 0.0%      | 0         |
| 130.150 optic disc coloboma                               | 3 0.1%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 64 1.3%     | 0         |
| 900.100 other, not inherited                              | 136 2.7%    | 2 0.2%    |
| 900.110 other. suspect not inherited/significance unknown | 145 2.9%    | 44 5.3%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 3,909 77.9% | 545 65.3% |

# BOXER

|    | <b>DISORDER</b>                           | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|---|--------------------|------------------|------------------------|
| A. | Distichiasis                              | Not defined        | 1                | Breeder option         |
| B. | Entropion                                 | Not Defined        | 1                | Breeder option         |
| C. | Ectropion                                 | Not defined        | 1                | Breeder option         |
| D. | Corneal dystrophy<br>- epithelial/stromal | Not defined        | 1                | Breeder option         |
| E. | Corneal dystrophy<br>- epithelial erosion | Not defined        | 1-4              | Breeder option         |
| F. | Cataract                                  | Not defined        | 1                | NO                     |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

In the Boxer, because there is significant clinical disease associated with the abnormal hairs, breeding affected animals should be discouraged.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### C. Ectropion

A conformational defect resulting in eversion of the eyelid(s), which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

D. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

E. Corneal dystrophy - epithelial erosion

A general group of corneal ulcerative conditions (e.g. erosions, indolent or persistent ulcers, epithelial bonding defects) is recognized as a common problem in older Boxers (as well as other older animals). It has been commonly referred to as Boxer corneal ulceration. Animals that are affected are usually 7-8 years of age or older. The ulceration can be a very difficult lesion to heal, and it is often recurrent. The chronic form stimulates eventual scarring, with vascularization, fibrosis and pigmentation of the lesion site. The lesion can cause vision impairment.

F. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Roberts SR. Superficial indolent ulcer in the cornea of Boxer dogs. *J Small Anim Pract.* 1965;6:111.
3. Gelatt KN and Samuelson DA. Recurrent corneal erosions and epithelial dystrophy in the Boxer dog. *J Am Anim Hosp Assoc.* 1982;18:453.
4. Kirschner SE, Niyo Y and Betts DM. Idiopathic persistent corneal erosions: clinical and pathological findings in 18 dogs. *J Am Anim Hosp Assoc.* 1989;25:84.



# OCULAR DISORDERS REPORT BOXER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>1,664 |       | 2016-2020<br>238 |       |
|---|---------------------|--------------------|-------|------------------|-------|
|   |                     | #                  | %     | #                | %     |
| <b>GLOBE</b>  |                     |                    |       |                  |       |
| 0.110 microphthalmia  |                     | 5                  | 0.3%  | 0                |       |
| <b>EYELIDS</b>  |                     |                    |       |                  |       |
| 20.140 ectopic cilia  |                     | 3                  | 0.2%  | 0                |       |
| 20.160 macropalpebral fissure   |                     | 9                  | 0.5%  | 0                |       |
| 21.000 entropion, unspecified   |                     | 4                  | 0.2%  | 3                | 1.3%  |
| 22.000 ectropion, unspecified   |                     | 61                 | 3.7%  | 9                | 3.8%  |
| 25.110 distichiasis   |                     | 198                | 11.9% | 32               | 13.4% |
| <b>NASOLACRIMAL</b>   |                     |                    |       |                  |       |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 1                  | 0.1%  | 0                |       |
| <b>CORNEA</b>   |                     |                    |       |                  |       |
| 70.210 corneal pannus   |                     | 1                  | 0.1%  | 0                |       |
| 70.220 pigmentary keratitis   |                     | 1                  | 0.1%  | 0                |       |
| 70.700 corneal dystrophy  |                     | 141                | 8.5%  | 14               | 5.9%  |
| 70.730 corneal endothelial degeneration                               |                     | 2                  | 0.1%  | 1                | 0.4%  |
| <b>UVEA</b>   |                     |                    |       |                  |       |
| 93.150 iris coloboma  |                     | 1                  | 0.1%  | 0                |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 4                  | 0.2%  | 0                |       |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 3                  | 0.2%  | 1                | 0.4%  |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 8                  | 0.5%  | 4                | 1.7%  |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 1                  | 0.1%  | 0                |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 1                  | 0.1%  | 4                | 1.7%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 3                  | 0.2%  | 1                | 0.4%  |
| 93.999 uveal cysts  |                     | 1                  | 0.1%  | 1                | 0.4%  |
| <b>LENS</b>   |                     |                    |       |                  |       |
| 100.200 cataract, unspecified   |                     | 4                  | 0.2%  | 0                |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 40                 | 2.4%  | 12               | 5.0%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 2                  | 0.1%  | 1                | 0.4%  |
| 100.303 punctate cataract, equatorial cortex                          |                     | 2                  | 0.1%  | 0                |       |
| 100.304 punctate cataract, anterior sutures                           |                     | 3                  | 0.2%  | 0                |       |
| 100.305 punctate cataract, posterior sutures                          |                     | 1                  | 0.1%  | 1                | 0.4%  |
| 100.306 punctate cataract, nucleus                                    |                     | 1                  | 0.1%  | 0                |       |
| 100.307 punctate cataract, capsular                                   |                     | 2                  | 0.1%  | 0                |       |
| 100.311 incipient cataract, anterior cortex                           |                     | 15                 | 0.9%  | 4                | 1.7%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 2                  | 0.1%  | 1                | 0.4%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 7                  | 0.4%  | 0                |       |
| 100.314 incipient cataract, anterior sutures                          |                     | 2                  | 0.1%  | 0                |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 2                  | 0.1%  | 0                |       |
| 100.316 incipient cataract, nucleus                                   |                     | 2                  | 0.1%  | 2                | 0.8%  |
| 100.317 incipient cataract, capsular                                  |                     | 1                  | 0.1%  | 1                | 0.4%  |
| 100.321 incomplete cataract, anterior cortex                          |                     | 1                  | 0.1%  | 1                | 0.4%  |
| 100.326 incomplete cataract, nucleus                                  |                     | 1                  | 0.1%  | 0                |       |
| 100.328 y-suture tip opacities  |                     | 1                  | 0.1%  | 1                | 0.4%  |
| 100.330 generalized/complete cataract                                 |                     | 7                  | 0.4%  | 0                |       |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 56                 | 3.4%  | 12               | 5.0%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.375 subluxation/luxation, unspecified                 | 2 0.1%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 2 0.1%           | 1 0.4%           |
| 110.135 PHPV/PTVL   | 1 0.1%           | 0                |
| 110.320 vitreal degeneration                              | 11 0.7%          | 1 0.4%           |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 5 0.3%           | 0                |
| 120.180 retinal dysplasia, geographic                     | 0                | 2 0.8%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 3 0.2%           | 0                |
| 120.400 retinal hemorrhage                                | 1 0.1%           | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.1%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 1 0.1%           | 0                |
| 130.120 optic nerve hypoplasia                            | 1 0.1%           | 0                |
| 130.150 optic disc coloboma                               | 3 0.2%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 13 0.8%          | 0                |
| 900.100 other, not inherited                              | 44 2.6%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 18 1.1%          | 13 5.5%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,217 73.1%      | 161 67.6%        |

# BOYKIN SPANIEL

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE         | GENETIC TESTS<br>AVAILABLE           |
|----|---|------------------------|-----------|----------------------------|--------------------------------------|
| A. | Distichiasis  | Not defined            | 1         | Breeder option             |                                      |
| B. | Corneal dystrophy<br>- epithelial/stromal   | Not defined            | 1         | Breeder option             |                                      |
| C. | Persistent pupillary<br>membranes   | Not defined            | 1         | Breeder option             |                                      |
|    | - iris to iris<br>- lens pigment foci/no<br>strands   | Not defined            | 1         | Passes with no<br>notation |                                      |
| D. | Cataract  | Not defined            | 1         | NO                         |                                      |
| E. | Y-suture tip opacity  | Not defined            | 1         | Breeder option             |                                      |
| F. | Persistent hyaloid artery   | Not defined            | 1         | Breeder option             |                                      |
| G. | Retinal atrophy<br>- generalized  | Not defined            | 1         | NO                         |                                      |
| H. | Retinal dysplasia<br>- folds  | Not defined            | 1         | Breeder option             |                                      |
| I. | Choroidal hypoplasia<br>(Collie Eye Anomaly)<br>- staphyloma/coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- optic nerve coloboma | Autosomal<br>recessive | 1, 2      | NO                         | Mutation in the<br><i>NHEJ1</i> gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

B. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

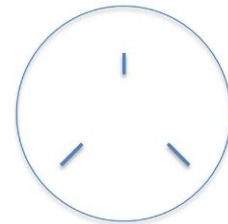
Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

F. Persistent hyaloid artery (PHA)

Congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small patent vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

G. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram before it is apparent clinically. In most breeds studied to date, retinal atrophy is recessively inherited.

H. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

I. Choroidal hypoplasia (Collie Eye Anomaly)

- Staphyloma/coloboma
- Retinal detachment
- Retinal hemorrhage
- Optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

## References

1. ACVO Genetics Committee, 1999 and/or Data from OFA All-Breeds Report, 1991-1998.
2. Parker HG, Kukekova AV, Akey DT, et al. Breed relationships facilitate fine-mapping studies: a 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. *Genome Res.* 2007 Nov;17:1562-1571.

# OCULAR DISORDERS REPORT BOYKIN SPANIEL

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>3,413 |       | 2016-2020<br>1,640 |       |
|---|---------------------|--------------------|-------|--------------------|-------|
|   |                     | #                  | %     | #                  | %     |
| <b>GLOBE</b>  |                     |                    |       |                    |       |
| 0.110 microphthalmia  |                     | 1                  | 0.0%  | 1                  | 0.1%  |
| <b>EYELIDS</b>  |                     |                    |       |                    |       |
| 20.160 macropalpebral fissure   |                     | 2                  | 0.1%  | 0                  |       |
| 21.000 entropion, unspecified   |                     | 1                  | 0.0%  | 2                  | 0.1%  |
| 25.110 distichiasis   |                     | 436                | 12.8% | 247                | 15.1% |
| <b>NASOLACRIMAL</b>   |                     |                    |       |                    |       |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 0                  |       | 3                  | 0.2%  |
| <b>NICTITANS</b>  |                     |                    |       |                    |       |
| 51.100 third eyelid cartilage anomaly                                 |                     | 1                  | 0.0%  | 1                  | 0.1%  |
| 52.110 prolapsed gland of the third eyelid                            |                     | 1                  | 0.0%  | 0                  |       |
| <b>CORNEA</b>   |                     |                    |       |                    |       |
| 70.210 corneal pannus   |                     | 1                  | 0.0%  | 0                  |       |
| 70.220 pigmentary keratitis   |                     | 4                  | 0.1%  | 0                  |       |
| 70.700 corneal dystrophy  |                     | 53                 | 1.6%  | 17                 | 1.0%  |
| 70.730 corneal endothelial degeneration                               |                     | 1                  | 0.0%  | 0                  |       |
| <b>UVEA</b>   |                     |                    |       |                    |       |
| 93.110 iris hypoplasia  |                     | 2                  | 0.1%  | 4                  | 0.2%  |
| 93.150 iris coloboma  |                     | 1                  | 0.0%  | 0                  |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 85                 | 2.5%  | 48                 | 2.9%  |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 2                  | 0.1%  | 0                  |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 5                  | 0.1%  | 1                  | 0.1%  |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 2                  | 0.1%  | 0                  |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 10                 | 0.3%  | 28                 | 1.7%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 0                  |       | 1                  | 0.1%  |
| 93.999 uveal cysts  |                     | 1                  | 0.0%  | 0                  |       |
| 97.150 chorioretinal coloboma, congenital                             |                     | 1                  | 0.0%  | 1                  | 0.1%  |
| <b>LENS</b>   |                     |                    |       |                    |       |
| 100.200 cataract, unspecified   |                     | 7                  | 0.2%  | 0                  |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 185                | 5.4%  | 151                | 9.2%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 15                 | 0.4%  | 16                 | 1.0%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 43                 | 1.3%  | 10                 | 0.6%  |
| 100.303 punctate cataract, equatorial cortex                          |                     | 7                  | 0.2%  | 0                  |       |
| 100.304 punctate cataract, anterior sutures                           |                     | 4                  | 0.1%  | 3                  | 0.2%  |
| 100.305 punctate cataract, posterior sutures                          |                     | 17                 | 0.5%  | 7                  | 0.4%  |
| 100.306 punctate cataract, nucleus                                    |                     | 9                  | 0.3%  | 9                  | 0.5%  |
| 100.307 punctate cataract, capsular                                   |                     | 6                  | 0.2%  | 13                 | 0.8%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 17                 | 0.5%  | 9                  | 0.5%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 39                 | 1.1%  | 35                 | 2.1%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 6                  | 0.2%  | 4                  | 0.2%  |
| 100.314 incipient cataract, anterior sutures                          |                     | 1                  | 0.0%  | 0                  |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 5                  | 0.1%  | 5                  | 0.3%  |
| 100.316 incipient cataract, nucleus                                   |                     | 10                 | 0.3%  | 5                  | 0.3%  |
| 100.317 incipient cataract, capsular                                  |                     | 7                  | 0.2%  | 16                 | 1.0%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.321 incomplete cataract, anterior cortex              | 0                | 3 0.2%           |
| 100.322 incomplete cataract, posterior cortex             | 0                | 3 0.2%           |
| 100.323 incomplete cataract, equatorial cortex            | 1 0.0%           | 3 0.2%           |
| 100.327 incomplete cataract, capsular                     | 0                | 1 0.1%           |
| 100.328 y-suture tip opacities                            | 4 0.1%           | 7 0.4%           |
| 100.330 generalized/complete cataract                     | 10 0.3%          | 1 0.1%           |
| 100.345 <i>significant cataracts (summary)</i>            | 208 6.1%         | 150 9.1%         |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 26 0.8%          | 36 2.2%          |
| 110.135 PHPV/PTVL   | 3 0.1%           | 3 0.2%           |
| 110.320 vitreal degeneration                              | 5 0.1%           | 5 0.3%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 48 1.4%          | 8 0.5%           |
| 97.120 coloboma   | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 63 1.8%          | 17 1.0%          |
| 120.180 retinal dysplasia, geographic                     | 9 0.3%           | 0                |
| 120.190 retinal dysplasia, detached                       | 1 0.0%           | 1 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 30 0.9%          | 3 0.2%           |
| 120.400 retinal hemorrhage                                | 2 0.1%           | 0                |
| 120.910 retinal detachment without dialysis               | 2 0.1%           | 0                |
| 120.920 retinal detachment with dialysis                  | 1 0.0%           | 2 0.1%           |
| 120.960 retinopathy                                       | 13 0.4%          | 3 0.2%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 1 0.0%           | 0                |
| 130.120 optic nerve hypoplasia                            | 4 0.1%           | 0                |
| 130.150 optic disc coloboma                               | 23 0.7%          | 15 0.9%          |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 73 2.1%          | 0                |
| 900.100 other, not inherited                              | 79 2.3%          | 10 0.6%          |
| 900.110 other. suspect not inherited/significance unknown | 46 1.3%          | 98 6.0%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 2,601 76.2%      | 1,006 61.3%      |

# BRACCO ITALIANO

|    | DISORDER     | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--------------|-------------|-----------|-----------------|
| A. | Distichiasis | Not defined | 1         | Breeder option  |
| B. | Cataract     | Not defined | 1         | NO              |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Bracco Italiano breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and Data from OFA All-Breeds Report.



# OCULAR DISORDERS REPORT BRACCO ITALIANO

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|------------------|--|-----------|-------|-----------|-------|
|                  |  | #         | %     | #         | %     |
| <b>EYELIDS</b>   |  |           |       |           |       |
| 20.160           | macropalpebral fissure                                       | 1         | 0.9%  | 0         |       |
| 21.000           | entropion, unspecified                                       | 4         | 3.5%  | 7         | 8.1%  |
| 25.110           | distichiasis   | 9         | 7.8%  | 10        | 11.6% |
| <b>NICTITANS</b> |  |           |       |           |       |
| 51.100           | third eyelid cartilage anomaly                               | 1         | 0.9%  | 1         | 1.2%  |
| 52.110           | prolapsed gland of the third eyelid                          | 1         | 0.9%  | 0         |       |
| <b>UVEA</b>      |  |           |       |           |       |
| 93.710           | persistent pupillary membranes, iris to iris                 | 2         | 1.7%  | 0         |       |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands | 0         |       | 2         | 2.3%  |
| <b>LENS</b>      |  |           |       |           |       |
| 100.210          | cataract. suspect not inherited/significance unknown         | 10        | 8.7%  | 4         | 4.7%  |
| 100.301          | punctate cataract, anterior cortex                           | 2         | 1.7%  | 0         |       |
| 100.302          | punctate cataract, posterior cortex                          | 3         | 2.6%  | 0         |       |
| 100.311          | incipient cataract, anterior cortex                          | 1         | 0.9%  | 2         | 2.3%  |
| 100.312          | incipient cataract, posterior cortex                         | 8         | 7.0%  | 1         | 1.2%  |
| 100.313          | incipient cataract, equatorial cortex                        | 3         | 2.6%  | 1         | 1.2%  |
| 100.316          | incipient cataract, nucleus                                  | 2         | 1.7%  | 0         |       |
| 100.317          | incipient cataract, capsular                                 | 2         | 1.7%  | 0         |       |
| 100.328          | y-suture tip opacities                                       | 0         |       | 1         | 1.2%  |
| 100.345          | significant cataracts (summary)                              | 21        | 18.3% | 5         | 5.8%  |
| <b>VITREOUS</b>  |  |           |       |           |       |
| 110.135          | PHPV/PTVL  | 1         | 0.9%  | 1         | 1.2%  |
| 110.320          | vitreal degeneration   | 0         |       | 2         | 2.3%  |
| <b>RETINA</b>    |  |           |       |           |       |
| 120.170          | retinal dysplasia, folds                                     | 6         | 5.2%  | 4         | 4.7%  |
| 120.960          | retinopathy  | 1         | 0.9%  | 1         | 1.2%  |
| <b>OTHER</b>     |  |           |       |           |       |
| 900.000          | other, unspecified   | 2         | 1.7%  | 0         |       |
| 900.100          | other, not inherited   | 3         | 2.6%  | 0         |       |
| 900.110          | other. suspect not inherited/significance unknown            | 1         | 0.9%  | 4         | 4.7%  |
| <b>NORMAL</b>    |  |           |       |           |       |
| 0.000            | normal globe   | 75        | 65.2% | 54        | 62.8% |

# **OCULAR DISORDERS REPORT BRAQUE D'Auvergne**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the BRAQUE D'Auvergne breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT BRAQUE D'AUVERGNE

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|--|---------------------|-----------|-------|-----------|-------|
|  |                     | #         | %     | #         | %     |
| <b>GLOBE</b>   |                     |           |       |           |       |
| 0.110 microphthalmia   |                     | 0         |       | 1         | 3.6%  |
| <b>EYELIDS</b>   |                     |           |       |           |       |
| 25.110 distichiasis  |                     | 0         |       | 1         | 3.6%  |
| <b>UVEA</b>  |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris          |                     | 1         | 5.6%  | 4         | 14.3% |
| 93.720 persistent pupillary membranes, iris to lens          |                     | 0         |       | 1         | 3.6%  |
| 93.730 persistent pupillary membranes, iris to cornea        |                     | 0         |       | 1         | 3.6%  |
| <b>LENS</b>  |                     |           |       |           |       |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 3         | 16.7% | 4         | 14.3% |
| 100.303 punctate cataract, equatorial cortex                 |                     | 0         |       | 1         | 3.6%  |
| 100.312 incipient cataract, posterior cortex                 |                     | 0         |       | 1         | 3.6%  |
| 100.317 incipient cataract, capsular                         |                     | 0         |       | 1         | 3.6%  |
| 100.345 <i>significant cataracts (summary)</i>               |                     | 0         |       | 3         | 10.7% |
| <b>RETINA</b>  |                     |           |       |           |       |
| 120.170 retinal dysplasia, folds                             |                     | 0         |       | 1         | 3.6%  |
| <b>OTHER</b>   |                     |           |       |           |       |
| 900.100 other, not inherited                                 |                     | 0         |       | 1         | 3.6%  |
| 900.110 other. suspect not inherited/significance unknown    |                     | 0         |       | 4         | 14.3% |
| <b>NORMAL</b>  |                     |           |       |           |       |
| 0.000 normal globe   |                     | 14        | 77.8% | 13        | 46.4% |

# BRAQUE FRANCAIS

| DISORDER    | INHERITANCE | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE |
|-------------|-------------|-----------|--------------------|----------------------------|
| A. Cataract | Not defined | 1         | NO                 |                            |

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## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT BRAQUE FRANCAIS

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 2         | 6.2%  | 1         | 1.5%  |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 0         |       | 2         | 3.0%  |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 0         |       | 3         | 4.5%  |
| 93.999          | uveal cysts  | 0         |       | 1         | 1.5%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 4         | 12.5% | 4         | 6.1%  |
| 100.301         | punctate cataract, anterior cortex                           | 0         |       | 1         | 1.5%  |
| 100.311         | incipient cataract, anterior cortex                          | 0         |       | 1         | 1.5%  |
| 100.312         | incipient cataract, posterior cortex                         | 1         | 3.1%  | 0         |       |
| 100.317         | incipient cataract, capsular                                 | 0         |       | 1         | 1.5%  |
| 100.322         | incomplete cataract, posterior cortex                        | 0         |       | 2         | 3.0%  |
| 100.323         | incomplete cataract, equatorial cortex                       | 0         |       | 1         | 1.5%  |
| 100.326         | incomplete cataract, nucleus                                 | 0         |       | 1         | 1.5%  |
| 100.345         | <i>significant cataracts (summary)</i>                       | 1         | 3.1%  | 7         | 10.6% |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.110         | other. suspect not inherited/significance unknown            | 3         | 9.4%  | 6         | 9.1%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 24        | 75.0% | 48        | 72.7% |

# BRAZILIAN TERRIER

|    | DISORDER                  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE           |
|----|---------------------------|------------------------|-----------|--------------------|--------------------------------------|
| A. | Multifocal<br>retinopathy | Autosomal<br>recessive | 1         | Breeder option     | Mutation in the <i>BEST1</i><br>gene |

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## Description and Comments

### A. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (cmr1) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (BEST1) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Brazilian Terrier. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Zangerl B, Wickstrom K, Slavik J, et al. Assessment of canine BEST1 variations identifies new mutations and establishes an independent bestrophinopathy model (cmr3). *Mol Vis.* 2010;16:2791-2804.

# BRIARD

|    | DISORDER  | INHERITANCE                | REFERENCE | BREEDING<br>ADVICE                           | GENETIC TESTS<br>AVAILABLE           |
|----|---|----------------------------|-----------|--|--------------------------------------|
| A. | Corneal dystrophy<br>- epithelial/stromal   | Not defined                | 1         | Breeder option                               |                                      |
| B. | Persistent pupillary<br>membranes<br>- iris to iris<br>- lens pigment foci/no<br>strands  | Not defined<br>Not defined | 1<br>1    | Breeder option<br>Passes with no<br>notation |                                      |
| C. | Cataract  | Not defined                | 1         | NO   |                                      |
| D. | Retinal dystrophy<br>formerly Congenital<br>stationary night blindness<br>( <i>CSNB</i> ) | Autosomal<br>recessive     | 1-3       | NO   | Mutation in the<br><i>LRIT3</i> gene |

## Description and Comments

### A. Corneal dystrophy- epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Retinal dystrophy formerly Congenital stationary night blindness (*CSNB*)

A non-progressive retinal function defect characterized primarily by night blindness; day vision is normal to severely compromised. CSNB is an autosomal recessive trait caused by a mutation in the RPE65 gene. The condition is detected by 5-6 weeks of age, after the postnatal maturation of the retina is completed. Nystagmus is present in some dogs, particularly in those having night blindness and severely compromised day vision. Ophthalmoscopic examination shows no abnormalities. Abnormalities in serum lipids (mild hypercholesterolemia) and elevated arachidonic acid have been noted in some animals. The ERG results are specific and diagnostic for the disorder. ERG testing is essential to distinguish this disorder from more central visual pathway defects which may appear clinically similar.

The gene mutation RPE65 has been identified. This is the same mutation as causes Leber's congenital amaurosis, also sometimes called juvenile retinitis pigmentosa (RP), in humans. A DNA test is available.

**Historical Note:**

Central progressive retinal atrophy was previously a condition listed for this breed. However as the condition is no longer identified in the breed, the condition has been removed. Central progressive retinal atrophy was a progressive retinal degeneration in which photoreceptor death occurred secondary to disease of the underlying pigment epithelium. Progression was slow and some animals never lost vision. CPRA occurred in England, but was uncommon elsewhere.

**References**

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Das, R. G., et al. (2019). "Genome-wide association study and whole-genome sequencing identify a deletion in LRIT3 associated with canine congenital stationary night blindness." Sci Rep 9(1): 14166. PMID: 31578364
3. Oh, A., et al. (2018). "Phenotypic characterization of complete CSNB in the inbred research beagle: how common is CSNB in research and companion dogs?" Doc Ophthalmol 137(2): 87-101. PMID: 30051304



# OCULAR DISORDERS REPORT BRIARD

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>2,221 |      | 2016-2020<br>200 |      |
|---|---------------------|--------------------|------|------------------|------|
|   |                     | #                  | %    | #                | %    |
| <b>GLOBE</b>  |                     |                    |      |                  |      |
| 10.000 glaucoma   |                     | 1                  | 0.0% | 0                |      |
| <b>EYELIDS</b>  |                     |                    |      |                  |      |
| 20.140 ectopic cilia  |                     | 1                  | 0.0% | 0                |      |
| 21.000 entropion, unspecified                                       |                     | 1                  | 0.0% | 0                |      |
| 25.110 distichiasis   |                     | 9                  | 0.4% | 1                | 0.5% |
| <b>NASOLACRIMAL</b>   |                     |                    |      |                  |      |
| 32.110 imperforate lower nasolacrimal punctum                       |                     | 2                  | 0.1% | 0                |      |
| <b>NICTITANS</b>  |                     |                    |      |                  |      |
| 51.100 third eyelid cartilage anomaly                               |                     | 2                  | 0.1% | 1                | 0.5% |
| 52.110 prolapsed gland of the third eyelid                          |                     | 2                  | 0.1% | 0                |      |
| <b>CORNEA</b>   |                     |                    |      |                  |      |
| 70.210 corneal pannus   |                     | 1                  | 0.0% | 0                |      |
| 70.700 corneal dystrophy  |                     | 31                 | 1.4% | 6                | 3.0% |
| <b>UVEA</b>   |                     |                    |      |                  |      |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 20                 | 0.9% | 9                | 4.5% |
| 93.720 persistent pupillary membranes, iris to lens                 |                     | 2                  | 0.1% | 1                | 0.5% |
| 93.730 persistent pupillary membranes, iris to cornea               |                     | 2                  | 0.1% | 0                |      |
| 93.740 persistent pupillary membranes, iris sheets                  |                     | 2                  | 0.1% | 0                |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 7                  | 0.3% | 7                | 3.5% |
| 93.999 uveal cysts  |                     | 10                 | 0.5% | 0                |      |
| <b>LENS</b>   |                     |                    |      |                  |      |
| 100.200 cataract, unspecified                                       |                     | 9                  | 0.4% | 0                |      |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 71                 | 3.2% | 10               | 5.0% |
| 100.301 punctate cataract, anterior cortex                          |                     | 6                  | 0.3% | 0                |      |
| 100.302 punctate cataract, posterior cortex                         |                     | 1                  | 0.0% | 0                |      |
| 100.303 punctate cataract, equatorial cortex                        |                     | 0                  |      | 1                | 0.5% |
| 100.305 punctate cataract, posterior sutures                        |                     | 2                  | 0.1% | 3                | 1.5% |
| 100.306 punctate cataract, nucleus                                  |                     | 5                  | 0.2% | 1                | 0.5% |
| 100.307 punctate cataract, capsular                                 |                     | 4                  | 0.2% | 2                | 1.0% |
| 100.311 incipient cataract, anterior cortex                         |                     | 6                  | 0.3% | 0                |      |
| 100.312 incipient cataract, posterior cortex                        |                     | 9                  | 0.4% | 0                |      |
| 100.313 incipient cataract, equatorial cortex                       |                     | 2                  | 0.1% | 0                |      |
| 100.315 incipient cataract, posterior sutures                       |                     | 1                  | 0.0% | 0                |      |
| 100.316 incipient cataract, nucleus                                 |                     | 3                  | 0.1% | 1                | 0.5% |
| 100.317 incipient cataract, capsular                                |                     | 2                  | 0.1% | 0                |      |
| 100.323 incomplete cataract, equatorial cortex                      |                     | 1                  | 0.0% | 0                |      |
| 100.328 y-suture tip opacities                                      |                     | 0                  |      | 2                | 1.0% |
| 100.330 generalized/complete cataract                               |                     | 3                  | 0.1% | 0                |      |
| 100.345 significant cataracts (summary)                             |                     | 54                 | 2.4% | 10               | 5.0% |
| <b>VITREOUS</b>   |                     |                    |      |                  |      |
| 110.120 persistent hyaloid artery/remnant                           |                     | 1                  | 0.0% | 0                |      |
| 110.135 PHPV/PTVL   |                     | 3                  | 0.1% | 0                |      |
| 110.320 vitreal degeneration  |                     | 2                  | 0.1% | 1                | 0.5% |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>FUNDUS</b>   |             |           |
| 97.120 coloboma   | 1 0.0%      | 0         |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 7 0.3%      | 0         |
| 120.180 retinal dysplasia, geographic                     | 1 0.0%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 1 0.0%      | 0         |
| 120.400 retinal hemorrhage                                | 1 0.0%      | 0         |
| 120.910 retinal detachment without dialysis               | 2 0.1%      | 0         |
| <b>OPTIC NERVE</b>  |             |           |
| 130.120 optic nerve hypoplasia                            | 1 0.0%      | 0         |
| 130.150 optic disc coloboma                               | 3 0.1%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 37 1.7%     | 0         |
| 900.100 other, not inherited                              | 58 2.6%     | 0         |
| 900.110 other. suspect not inherited/significance unknown | 32 1.4%     | 8 4.0%    |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 2,035 91.6% | 161 80.5% |

# BRITTANY

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--------------------------------|-------------|-----------|-------------------------|
| A. | Distichiasis                   | Not defined | 1         | Breeder option          |
| B. | Persistent pupillary membrane  |             |           |                         |
|    | - iris to iris                 | Not defined | 1         | Breeder option          |
|    | - lens pigment foci/no strands | Not defined | 1         | Passes with no notation |
| C. | Cataract                       | Not defined | 1         | NO                      |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membrane (PPM)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

The exact frequency and significance of cataracts in the Brittany is not known, although it is probably low.

## References

There are no references providing detailed descriptions of hereditary conditions of the Brittany breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT BRITTANY

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 25.110 distichiasis   |                     | 54        | 2.4% | 14        | 1.6% |
| <b>NASOLACRIMAL</b>   |                     |           |      |           |      |
| 40.910 keratoconjunctivitis sicca                                   |                     | 1         | 0.0% | 0         |      |
| <b>NICTITANS</b>  |                     |           |      |           |      |
| 52.110 prolapsed gland of the third eyelid                          |                     | 2         | 0.1% | 0         |      |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.700 corneal dystrophy  |                     | 5         | 0.2% | 2         | 0.2% |
| 70.730 corneal endothelial degeneration                             |                     | 3         | 0.1% | 0         |      |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 33        | 1.5% | 15        | 1.8% |
| 93.720 persistent pupillary membranes, iris to lens                 |                     | 2         | 0.1% | 1         | 0.1% |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 8         | 0.4% | 18        | 2.1% |
| 93.999 uveal cysts  |                     | 1         | 0.0% | 1         | 0.1% |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.200 cataract, unspecified                                       |                     | 10        | 0.4% | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 94        | 4.2% | 42        | 4.9% |
| 100.301 punctate cataract, anterior cortex                          |                     | 13        | 0.6% | 3         | 0.4% |
| 100.302 punctate cataract, posterior cortex                         |                     | 27        | 1.2% | 9         | 1.1% |
| 100.303 punctate cataract, equatorial cortex                        |                     | 2         | 0.1% | 1         | 0.1% |
| 100.304 punctate cataract, anterior sutures                         |                     | 1         | 0.0% | 0         |      |
| 100.305 punctate cataract, posterior sutures                        |                     | 6         | 0.3% | 0         |      |
| 100.306 punctate cataract, nucleus                                  |                     | 1         | 0.0% | 2         | 0.2% |
| 100.307 punctate cataract, capsular                                 |                     | 9         | 0.4% | 2         | 0.2% |
| 100.311 incipient cataract, anterior cortex                         |                     | 10        | 0.4% | 4         | 0.5% |
| 100.312 incipient cataract, posterior cortex                        |                     | 36        | 1.6% | 20        | 2.3% |
| 100.313 incipient cataract, equatorial cortex                       |                     | 13        | 0.6% | 0         |      |
| 100.314 incipient cataract, anterior sutures                        |                     | 2         | 0.1% | 0         |      |
| 100.315 incipient cataract, posterior sutures                       |                     | 9         | 0.4% | 3         | 0.4% |
| 100.316 incipient cataract, nucleus                                 |                     | 7         | 0.3% | 1         | 0.1% |
| 100.317 incipient cataract, capsular                                |                     | 4         | 0.2% | 4         | 0.5% |
| 100.321 incomplete cataract, anterior cortex                        |                     | 1         | 0.0% | 0         |      |
| 100.322 incomplete cataract, posterior cortex                       |                     | 1         | 0.0% | 0         |      |
| 100.323 incomplete cataract, equatorial cortex                      |                     | 1         | 0.0% | 0         |      |
| 100.327 incomplete cataract, capsular                               |                     | 1         | 0.0% | 3         | 0.4% |
| 100.328 y-suture tip opacities                                      |                     | 1         | 0.0% | 1         | 0.1% |
| 100.330 generalized/complete cataract                               |                     | 4         | 0.2% | 0         |      |
| 100.340 resorbing/hypermature cataract                              |                     | 0         |      | 1         | 0.1% |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 159       | 7.1% | 54        | 6.3% |
| 100.375 <i>subluxation/luxation, unspecified</i>                    |                     | 3         | 0.1% | 1         | 0.1% |
| <b>VITREOUS</b>   |                     |           |      |           |      |
| 110.120 persistent hyaloid artery/remnant                           |                     | 3         | 0.1% | 7         | 0.8% |
| 110.135 PHPV/PTVL   |                     | 1         | 0.0% | 1         | 0.1% |
| 110.200 vitreous degeneration-anterior chamber                      |                     | 0         |      | 4         | 0.5% |
| 110.320 vitreal degeneration  |                     | 14        | 0.6% | 5         | 0.6% |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 7 0.3%      | 3 0.4%    |
| 120.180 retinal dysplasia, geographic                     | 6 0.3%      | 3 0.4%    |
| 120.310 generalized progressive retinal atrophy (PRA)     | 21 0.9%     | 0         |
| 120.910 retinal detachment without dialysis               | 1 0.0%      | 0         |
| 120.920 retinal detachment with dialysis                  | 1 0.0%      | 0         |
| 120.960 retinopathy                                       | 2 0.1%      | 1 0.1%    |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 1 0.0%      | 0         |
| 130.120 optic nerve hypoplasia                            | 1 0.0%      | 0         |
| 130.150 optic disc coloboma                               | 1 0.0%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 17 0.8%     | 0         |
| 900.100 other, not inherited                              | 61 2.7%     | 2 0.2%    |
| 900.110 other. suspect not inherited/significance unknown | 17 0.8%     | 47 5.5%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,946 86.8% | 666 77.8% |

# BRUSSELS GRIFFON

|    | DISORDER   | INHERITANCE                | REFERENCES | BREEDING ADVICE                           |
|----|--|----------------------------|------------|---|
| A. | Exposure keratopathy syndrome  | Not defined                | 1          | Breeder option                            |
| B. | Distichiasis   | Not defined                | 1          | Breeder option                            |
| C. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined | 1<br>1     | Breeder option<br>Passes with no notation |
| D. | Cataract   | Not defined                | 1          | NO  |
| E. | Lens luxation  | Autosomal recessive        | 1          | NO  |
| F. | Vitreous degeneration  | Not defined                | 1, 2       | Breeder option                            |
| G. | Retinal atrophy<br>- generalized   | Not defined                | 1          | NO  |
| H. | Retinal dysplasia<br>- folds   | Not defined                | 1          | Breeder option                            |

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## Description and Comments

### A. Exposure keratopathy syndrome/macrolepharon

A condition characterized by variable degrees of superficial vascularization, fibrosis and/or pigmentation of the cornea. May be associated with excessive exposure/irritation of the globe due to shallow orbits, lower eyelid medial entropion, lagophthalmos and macropalpebral fissure.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness.

F. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

G. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as Progressive Retinal Atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. Except for X-linked PRA in the Siberian Husky, in all breeds studied to date, PRA is inherited as an autosomal recessive trait.

H. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

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1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.



2. Krishnan, H., et al. (2020). "Vitreous degeneration and associated ocular abnormalities in the dog." Vet Ophthalmol 23(2): 219-224. PMID: 31464365.

# OCULAR DISORDERS REPORT BRUSSELS GRIFFON

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|---------------------|--|-----------|-------|-----------|-------|
|                     |  | #         | %     | #         | %     |
| <b>EYELIDS</b>      |  |           |       |           |       |
| 20.140              | ectopic cilia  | 8         | 0.6%  | 0         |       |
| 21.000              | entropion, unspecified   | 3         | 0.2%  | 3         | 0.9%  |
| 25.110              | distichiasis   | 30        | 2.2%  | 6         | 1.8%  |
| <b>NASOLACRIMAL</b> |  |           |       |           |       |
| 40.910              | keratoconjunctivitis sicca                                     | 3         | 0.2%  | 0         |       |
| <b>CORNEA</b>       |  |           |       |           |       |
| 70.210              | corneal pannus   | 1         | 0.1%  | 0         |       |
| 70.220              | pigmentary keratitis   | 23        | 1.7%  | 5         | 1.5%  |
| 70.700              | corneal dystrophy  | 10        | 0.7%  | 3         | 0.9%  |
| <b>UVEA</b>         |  |           |       |           |       |
| 93.110              | iris hypoplasia  | 2         | 0.1%  | 0         |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 121       | 8.8%  | 35        | 10.4% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 1         | 0.1%  | 0         |       |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 2         | 0.1%  | 0         |       |
| 93.740              | persistent pupillary membranes, iris sheets                    | 1         | 0.1%  | 0         |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 9         | 0.7%  | 12        | 3.6%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 4         | 0.3%  | 1         | 0.3%  |
| 93.999              | uveal cysts  | 2         | 0.1%  | 0         |       |
| 97.150              | chorioretinal coloboma, congenital                             | 1         | 0.1%  | 0         |       |
| <b>LENS</b>         |  |           |       |           |       |
| 100.200             | cataract, unspecified  | 8         | 0.6%  | 0         |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 53        | 3.9%  | 8         | 2.4%  |
| 100.301             | punctate cataract, anterior cortex                             | 23        | 1.7%  | 2         | 0.6%  |
| 100.302             | punctate cataract, posterior cortex                            | 11        | 0.8%  | 1         | 0.3%  |
| 100.303             | punctate cataract, equatorial cortex                           | 5         | 0.4%  | 0         |       |
| 100.304             | punctate cataract, anterior sutures                            | 3         | 0.2%  | 1         | 0.3%  |
| 100.305             | punctate cataract, posterior sutures                           | 1         | 0.1%  | 0         |       |
| 100.307             | punctate cataract, capsular                                    | 4         | 0.3%  | 0         |       |
| 100.311             | incipient cataract, anterior cortex                            | 78        | 5.7%  | 6         | 1.8%  |
| 100.312             | incipient cataract, posterior cortex                           | 35        | 2.5%  | 2         | 0.6%  |
| 100.313             | incipient cataract, equatorial cortex                          | 43        | 3.1%  | 1         | 0.3%  |
| 100.314             | incipient cataract, anterior sutures                           | 7         | 0.5%  | 0         |       |
| 100.315             | incipient cataract, posterior sutures                          | 5         | 0.4%  | 0         |       |
| 100.316             | incipient cataract, nucleus                                    | 5         | 0.4%  | 0         |       |
| 100.317             | incipient cataract, capsular                                   | 2         | 0.1%  | 0         |       |
| 100.321             | incomplete cataract, anterior cortex                           | 1         | 0.1%  | 4         | 1.2%  |
| 100.322             | incomplete cataract, posterior cortex                          | 0         |       | 1         | 0.3%  |
| 100.330             | generalized/complete cataract                                  | 29        | 2.1%  | 0         |       |
| 100.345             | significant cataracts (summary)                                | 260       | 18.9% | 18        | 5.4%  |
| 100.375             | subluxation/luxation, unspecified                              | 8         | 0.6%  | 0         |       |
| <b>VITREOUS</b>     |  |           |       |           |       |
| 110.120             | persistent hyaloid artery/remnant                              | 9         | 0.7%  | 2         | 0.6%  |
| 110.135             | PHPV/PTVL  | 2         | 0.1%  | 0         |       |
| 110.200             | vitreous degeneration-anterior chamber                         | 15        | 1.1%  | 11        | 3.3%  |

| <b>VITREOUS CONTINUED</b>                                 | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 110.320 vitreal degeneration                              | 320 23.3%        | 22 6.5%          |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 2 0.1%           | 0                |
| 97.120 coloboma   | 2 0.1%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 21 1.5%          | 14 4.2%          |
| 120.180 retinal dysplasia, geographic                     | 13 0.9%          | 3 0.9%           |
| 120.190 retinal dysplasia, detached                       | 2 0.1%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 23 1.7%          | 0                |
| 120.400 retinal hemorrhage                                | 2 0.1%           | 0                |
| 120.910 retinal detachment without dialysis               | 2 0.1%           | 0                |
| 120.960 retinopathy                                       | 0                | 1 0.3%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.120 optic nerve hypoplasia                            | 3 0.2%           | 0                |
| 130.150 optic disc coloboma                               | 19 1.4%          | 1 0.3%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 26 1.9%          | 0                |
| 900.100 other, not inherited                              | 27 2.0%          | 2 0.6%           |
| 900.110 other. suspect not inherited/significance unknown | 21 1.5%          | 17 5.1%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 825 60.0%        | 219 65.2%        |

# **OCULAR DISORDERS REPORT BULL TERRIER**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the BULL TERRIER breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT BULL TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |       |
|---|---------------------|-----------|------|-----------|-------|
|   |                     | #         | %    | #         | %     |
| <b>GLOBE</b>  |                     |           |      |           |       |
| 0.110 microphthalmia  |                     | 3         | 1.2% | 0         |       |
| <b>EYELIDS</b>  |                     |           |      |           |       |
| 21.000 entropion, unspecified   |                     | 2         | 0.8% | 0         |       |
| 22.000 ectropion, unspecified   |                     | 1         | 0.4% | 0         |       |
| 25.110 distichiasis   |                     | 5         | 2.0% | 0         |       |
| <b>CORNEA</b>   |                     |           |      |           |       |
| 70.700 corneal dystrophy  |                     | 1         | 0.4% | 0         |       |
| 70.730 corneal endothelial degeneration                               |                     | 5         | 2.0% | 0         |       |
| <b>UVEA</b>   |                     |           |      |           |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 8         | 3.3% | 0         |       |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 4         | 1.6% | 0         |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 12        | 4.9% | 0         |       |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 1         | 0.4% | 0         |       |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1         | 0.4% | 0         |       |
| <b>LENS</b>   |                     |           |      |           |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 6         | 2.4% | 0         |       |
| 100.301 punctate cataract, anterior cortex                            |                     | 2         | 0.8% | 1         | 6.7%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 2         | 0.8% | 0         |       |
| 100.303 punctate cataract, equatorial cortex                          |                     | 2         | 0.8% | 0         |       |
| 100.304 punctate cataract, anterior sutures                           |                     | 1         | 0.4% | 0         |       |
| 100.306 punctate cataract, nucleus                                    |                     | 1         | 0.4% | 0         |       |
| 100.307 punctate cataract, capsular                                   |                     | 1         | 0.4% | 0         |       |
| 100.311 incipient cataract, anterior cortex                           |                     | 1         | 0.4% | 0         |       |
| 100.312 incipient cataract, posterior cortex                          |                     | 1         | 0.4% | 0         |       |
| 100.313 incipient cataract, equatorial cortex                         |                     | 2         | 0.8% | 2         | 13.3% |
| 100.314 incipient cataract, anterior sutures                          |                     | 1         | 0.4% | 0         |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 1         | 0.4% | 0         |       |
| 100.330 generalized/complete cataract                                 |                     | 3         | 1.2% | 0         |       |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 18        | 7.3% | 3         | 20.0% |
| 100.375 <i>subluxation/luxation, unspecified</i>                      |                     | 7         | 2.8% | 0         |       |
| <b>VITREOUS</b>   |                     |           |      |           |       |
| 110.320 vitreal degeneration  |                     | 5         | 2.0% | 0         |       |
| <b>RETINA</b>   |                     |           |      |           |       |
| 120.170 retinal dysplasia, folds                                      |                     | 1         | 0.4% | 0         |       |
| 120.180 retinal dysplasia, geographic                                 |                     | 0         |      | 1         | 6.7%  |
| 120.310 generalized progressive retinal atrophy (PRA)                 |                     | 1         | 0.4% | 0         |       |
| 120.910 retinal detachment without dialysis                           |                     | 2         | 0.8% | 0         |       |
| <b>OPTIC NERVE</b>  |                     |           |      |           |       |
| 130.110 micropapilla  |                     | 3         | 1.2% | 0         |       |
| 130.120 optic nerve hypoplasia  |                     | 3         | 1.2% | 0         |       |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 5 2.0%    | 0         |
| 900.100 other, not inherited                              | 8 3.3%    | 0         |
| 900.110 other. suspect not inherited/significance unknown | 3 1.2%    | 0         |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 194 78.9% | 12 80.0%  |

# BULLDOG

|    | DISORDER                             | INHERITANCE         | REFERENCE | BREEDING ADVICE         | GENETIC TESTS AVAILABLE           |
|----|--------------------------------------|---------------------|-----------|-------------------------|-----------------------------------|
| A. | Keratoconjunctivitis sicca           | Not defined         | 1, 5, 6   | NO                      |                                   |
| B. | Entropion                            | Not defined         | 1         | Breeder option          |                                   |
| C. | Ectropion                            | Not defined         | 1         | Breeder option          |                                   |
| D. | Distichiasis                         | Not defined         | 1         | Breeder option          |                                   |
| E. | Ectopic cilia                        | Not defined         | 1         | Breeder option          |                                   |
| F. | Prolapsed gland of third eyelid      | Not defined         | 1, 2-4    | Breeder option          |                                   |
| G. | Exposure/Pigmentary Keratitis        | Not defined         | 1         | Breeder option          |                                   |
| H. | Secondary keratitis - chronic        | Not defined         | 1         | Passes with no notation |                                   |
| I. | Persistent pupillary membranes       |                     |           |                         |                                   |
|    | - iris to iris                       | Not defined         | 1         | Breeder option          |                                   |
|    | - iris to cornea                     | Not defined         | 1         | NO                      |                                   |
| J. | Cataract                             | Not defined         | 1         | NO                      |                                   |
| K. | Retinal dysplasia - folds            | Not defined         | 1         | Breeder option          |                                   |
| L. | Multifocal retinopathy - <i>cmr1</i> | Autosomal recessive | 7, 8      | Breeder option          | Mutation in the <i>BEST1</i> gene |

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## Description and Comments

### A. Keratoconjunctivitis sicca

An abnormality of the tear film, most commonly a deficiency of the aqueous portion, although the mucin and/or lipid layers may be affected; results in ocular irritation and/or vision impairment.

B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

C. Ectropion

A conformational defect resulting in eversion of the eyelids which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

In the Bulldog, ectropion is associated with an exceptionally large palpebral fissure and laxity of the canthal structures. Central lower lid ectropion is often associated with entropion of the adjacent lid. This causes severe ocular irritation.

D. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

In the Bulldog, these abnormal eyelashes may be associated with significant clinical disease and breeding of affected animals should be discouraged.

E. Ectopic cilia

Hair emerging through the eyelid conjunctiva. Ectopic cilia occur more frequently in younger dogs and cause discomfort and corneal disease.

F. Prolapse of the gland of the third eyelid

Protrusion of the tear gland associated with the third eyelid. The mode of inheritance of this disorder is unknown. The exposed gland may become irritated and severe chronic inflammation or keratoconjunctivitis sicca/dry eye syndrome may ensue. Commonly referred to as "cherry eye."

Bulldogs were overrepresented in a study of prolapsed gland of the third eyelid. In the study, 100% of the prolapsed glands in Bulldogs occurred before 1 year of age. Bulldogs were also more likely to develop bilateral prolapsed glands that occurred either simultaneously with the first prolapse or with a short time interval between prolapses.

G. Exposure/pigmentary keratitis

A condition characterized by variable degrees of superficial vascularization, fibrosis and/or pigmentation of the cornea. May be associated with excessive exposure/irritation of the



globe due to shallow orbits, lower eyelid medial entropion, lagophthalmos and macropalpebral fissure.

H. Secondary keratitis - chronic

A specific designation does not exist on the CAER form for this condition. We ask examiners to mark other – unlisted conditions suspected as inherited. Then in the comments box please write secondary keratitis – chronic.

A condition characterized by variable degrees of superficial vascularization, fibrosis and/or pigmentation of the cornea. Often associated with entropion or a combination of entropion and ectropion.

I. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

J. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

K. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

L. Multifocal Retinopathy

A specific designation does not exist on the CAER form for this condition. We ask examiners to mark other – unlisted conditions suspected as inherited. Then in the comments box please write multifocal retinopathy.

Canine Multifocal Retinopathy type 1 (cmr1) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog. The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal

thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas. Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff. A DNA test is available.

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1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
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# OCULAR DISORDERS REPORT BULLDOG

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 0.110 microphthalmia  |                     | 1         | 0.1%  | 0         |       |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 20.140 ectopic cilia  |                     | 8         | 0.7%  | 5         | 1.2%  |
| 20.160 macropalpebral fissure   |                     | 16        | 1.4%  | 0         |       |
| 21.000 entropion, unspecified   |                     | 172       | 15.1% | 63        | 15.1% |
| 22.000 ectropion, unspecified   |                     | 63        | 5.5%  | 13        | 3.1%  |
| 25.110 distichiasis   |                     | 258       | 22.6% | 108       | 26.0% |
| <b>NASOLACRIMAL</b>   |                     |           |       |           |       |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 4         | 0.4%  | 2         | 0.5%  |
| 40.910 keratoconjunctivitis sicca                                     |                     | 2         | 0.2%  | 10        | 2.4%  |
| <b>NICTITANS</b>  |                     |           |       |           |       |
| 52.110 prolapsed gland of the third eyelid                            |                     | 16        | 1.4%  | 7         | 1.7%  |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.210 corneal pannus   |                     | 9         | 0.8%  | 4         | 1.0%  |
| 70.220 pigmentary keratitis   |                     | 24        | 2.1%  | 8         | 1.9%  |
| 70.700 corneal dystrophy  |                     | 10        | 0.9%  | 2         | 0.5%  |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 7         | 0.6%  | 6         | 1.4%  |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 1         | 0.1%  | 0         |       |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 2         | 0.2%  | 0         |       |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1         | 0.1%  | 0         |       |
| 93.999 uveal cysts  |                     | 11        | 1.0%  | 4         | 1.0%  |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.200 cataract, unspecified   |                     | 1         | 0.1%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 32        | 2.8%  | 4         | 1.0%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 4         | 0.4%  | 0         |       |
| 100.302 punctate cataract, posterior cortex                           |                     | 2         | 0.2%  | 0         |       |
| 100.303 punctate cataract, equatorial cortex                          |                     | 0         |       | 1         | 0.2%  |
| 100.305 punctate cataract, posterior sutures                          |                     | 1         | 0.1%  | 1         | 0.2%  |
| 100.306 punctate cataract, nucleus                                    |                     | 0         |       | 1         | 0.2%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 5         | 0.4%  | 1         | 0.2%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 2         | 0.2%  | 0         |       |
| 100.313 incipient cataract, equatorial cortex                         |                     | 3         | 0.3%  | 1         | 0.2%  |
| 100.314 incipient cataract, anterior sutures                          |                     | 1         | 0.1%  | 0         |       |
| 100.316 incipient cataract, nucleus                                   |                     | 3         | 0.3%  | 1         | 0.2%  |
| 100.317 incipient cataract, capsular                                  |                     | 1         | 0.1%  | 0         |       |
| 100.328 y-suture tip opacities  |                     | 0         |       | 1         | 0.2%  |
| 100.330 generalized/complete cataract                                 |                     | 5         | 0.4%  | 0         |       |
| 100.345 significant cataracts (summary)                               |                     | 28        | 2.5%  | 7         | 1.7%  |
| 100.375 subluxation/luxation, unspecified                             |                     | 2         | 0.2%  | 1         | 0.2%  |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>VITREOUS</b>   |           |           |
| 110.120 persistent hyaloid artery/remnant                 | 1 0.1%    | 0         |
| 110.320 vitreal degeneration                              | 2 0.2%    | 0         |
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 69 6.0%   | 18 4.3%   |
| 120.180 retinal dysplasia, geographic                     | 3 0.3%    | 1 0.2%    |
| 120.190 retinal dysplasia, detached                       | 2 0.2%    | 0         |
| 120.960 retinopathy                                       | 1 0.1%    | 0         |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 7 0.6%    | 0         |
| 900.100 other, not inherited                              | 39 3.4%   | 2 0.5%    |
| 900.110 other. suspect not inherited/significance unknown | 36 3.2%   | 37 8.9%   |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 665 58.2% | 198 47.6% |

# BULLMASTIFF

|    | DISORDER                             | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|--------------------------------------|---------------------|-----------|-----------------|-----------------------------------|
| A. | Entropion                            | Not defined         | 1         | Breeder option  |                                   |
| B. | Ectropion                            | Not defined         | 1         | Breeder option  |                                   |
| C. | Distichiasis                         | Not defined         | 1         | Breeder option  |                                   |
| D. | Persistent pupillary membranes       |                     |           |                 |                                   |
|    | - iris to iris                       | Not defined         | 1         | Breeder option  |                                   |
|    | - iris to cornea                     | Not defined         | 1         | NO              |                                   |
| E. | Cataract                             | Not defined         | 1         | NO              |                                   |
| F. | Retinal atrophy ( <i>RHO</i> )       | Autosomal dominant  | 2         | NO              | Mutation in the <i>RHO</i> gene   |
| G. | Retinal dysplasia - folds            | Not defined         | 1         | Breeder option  |                                   |
| H. | Multifocal retinopathy - <i>cmr1</i> | Autosomal recessive | 3         | Breeder option  | Mutation in the <i>BEST1</i> gene |

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## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

In the Bullmastiff, the palpebral fissures may become vertical and/or shaped like a "pagoda." Entropion in the Bullmastiff is severe and may require multiple surgical corrections.

### B. Ectropion

A conformational defect resulting in eversion (rolling-out) of the eyelids, which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the

conformation of the skull.

C. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

F. Retinal atrophy - *RHO*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. PRA in the Bullmastiff is inherited as an autosomal dominant trait. A DNA test is available.

G. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

H. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (*cmr1*) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous sub-retinal fluid, or accumulation of sub-retinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat

like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Kijas JW, Cideciyan AV, Aleman TS, et al. Naturally occurring rhodopsin mutation in the dog causes retinal dysfunction and degeneration mimicking human dominant retinitis pigmentosa. *Proc Natl Acad Sci U S A*. 2002 Apr 30;99:6328-6333.
3. Guziewicz KE, Zangerl B, Lindauer SJ, et al. Bestrophin gene mutations cause canine multifocal retinopathy: a novel animal model for best disease. *Invest Ophthalmol Vis Sci*. 2007 May;48:1959-1967.

# OCULAR DISORDERS REPORT BULLMASTIFF

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 0.110 microphthalmia  |                     | 5         | 0.3% | 0         |      |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 20.160 macropalpebral fissure   |                     | 16        | 1.0% | 0         |      |
| 21.000 entropion, unspecified   |                     | 96        | 5.9% | 33        | 4.9% |
| 22.000 ectropion, unspecified   |                     | 24        | 1.5% | 10        | 1.5% |
| 25.110 distichiasis   |                     | 42        | 2.6% | 16        | 2.4% |
| <b>NASOLACRIMAL</b>   |                     |           |      |           |      |
| 40.910 keratoconjunctivitis sicca                                     |                     | 0         |      | 1         | 0.1% |
| <b>NICTITANS</b>  |                     |           |      |           |      |
| 51.100 third eyelid cartilage anomaly                                 |                     | 1         | 0.1% | 2         | 0.3% |
| 52.110 prolapsed gland of the third eyelid                            |                     | 1         | 0.1% | 0         |      |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.210 corneal pannus   |                     | 2         | 0.1% | 0         |      |
| 70.220 pigmentary keratitis   |                     | 4         | 0.2% | 3         | 0.4% |
| 70.700 corneal dystrophy  |                     | 2         | 0.1% | 1         | 0.1% |
| 70.730 corneal endothelial degeneration                               |                     | 1         | 0.1% | 0         |      |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.140 corneal endothelial pigment without PPM                        |                     | 1         | 0.1% | 0         |      |
| 93.150 iris coloboma  |                     | 3         | 0.2% | 1         | 0.1% |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 54        | 3.3% | 39        | 5.8% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 9         | 0.6% | 1         | 0.1% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 23        | 1.4% | 5         | 0.7% |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 1         | 0.1% | 0         |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 6         | 0.4% | 1         | 0.1% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 3         | 0.2% | 5         | 0.7% |
| 93.999 uveal cysts  |                     | 9         | 0.6% | 5         | 0.7% |
| 97.150 chorioretinal coloboma, congenital                             |                     | 1         | 0.1% | 0         |      |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.200 cataract, unspecified   |                     | 1         | 0.1% | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 51        | 3.1% | 17        | 2.5% |
| 100.301 punctate cataract, anterior cortex                            |                     | 5         | 0.3% | 2         | 0.3% |
| 100.302 punctate cataract, posterior cortex                           |                     | 4         | 0.2% | 1         | 0.1% |
| 100.303 punctate cataract, equatorial cortex                          |                     | 1         | 0.1% | 0         |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 1         | 0.1% | 1         | 0.1% |
| 100.307 punctate cataract, capsular                                   |                     | 2         | 0.1% | 0         |      |
| 100.311 incipient cataract, anterior cortex                           |                     | 11        | 0.7% | 4         | 0.6% |
| 100.312 incipient cataract, posterior cortex                          |                     | 13        | 0.8% | 4         | 0.6% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 9         | 0.6% | 4         | 0.6% |
| 100.315 incipient cataract, posterior sutures                         |                     | 2         | 0.1% | 0         |      |
| 100.316 incipient cataract, nucleus                                   |                     | 4         | 0.2% | 0         |      |
| 100.321 incomplete cataract, anterior cortex                          |                     | 1         | 0.1% | 0         |      |
| 100.322 incomplete cataract, posterior cortex                         |                     | 3         | 0.2% | 1         | 0.1% |
| 100.323 incomplete cataract, equatorial cortex                        |                     | 1         | 0.1% | 0         |      |



| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.326 incomplete cataract, nucleus                      | 1 0.1%           | 0                |
| 100.328 y-suture tip opacities                            | 1 0.1%           | 2 0.3%           |
| 100.330 generalized/complete cataract                     | 7 0.4%           | 1 0.1%           |
| 100.345 <i>significant cataracts (summary)</i>            | 67 4.1%          | 20 3.0%          |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 0                | 2 0.3%           |
| 110.135 PHPV/PTVL   | 1 0.1%           | 0                |
| 110.320 vitreal degeneration                              | 3 0.2%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 74 4.5%          | 46 6.9%          |
| 120.180 retinal dysplasia, geographic                     | 3 0.2%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 3 0.2%           | 0                |
| 120.960 retinopathy                                       | 4 0.2%           | 3 0.4%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 7 0.4%           | 1 0.1%           |
| 130.120 optic nerve hypoplasia                            | 6 0.4%           | 2 0.3%           |
| 130.150 optic disc coloboma                               | 2 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 25 1.5%          | 0                |
| 900.100 other, not inherited                              | 42 2.6%          | 3 0.4%           |
| 900.110 other. suspect not inherited/significance unknown | 19 1.2%          | 28 4.2%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,248 76.6%      | 476 71.2%        |

# CAIRN TERRIER

|    | DISORDER                                   | INHERITANCE                 | REFERENCE | BREEDING ADVICE         |
|----|--|-----------------------------|-----------|-------------------------|
| A. | Ocular melanosis with and without glaucoma | Presumed autosomal dominant | 1, 2      | NO                      |
| B. | Persistent pupillary membranes             |                             |           |                         |
|    | - iris to iris                             | Not defined                 | 1         | Breeder option          |
|    | - lens pigment foci/no strands             | Not defined                 | 1         | Passes with no notation |
|    | - endothelial opacity/no strands           | Not defined                 | 1         | NO                      |
| C. | Cataract                                   | Not defined                 | 1         | NO                      |
| D. | Vitreous degeneration                      | Not defined                 | 1         | Breeder option          |
| E. | Persistent hyaloid artery                  | Not defined                 | 1         | Breeder option          |

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## Description and Comments

- A. Ocular melanosis with and without glaucoma  
(Previously ocular melanosis with secondary glaucoma, previously pigmentary glaucoma)

A proliferation of melanocytes within the uveal tract associated with an elevation in intraocular pressure. Obstruction of the aqueous outflow pathways occurs resulting in glaucoma. This condition has been identified most commonly in the Cairn Terrier. The condition is familial but the exact mode of inheritance is unknown (pedigree analysis has ruled out a sex-linked disorder). In the Cairn Terrier, the disease is very slowly progressive and blindness ultimately results. Some dogs develop episodes of anterior uveitis associated with the shedding of large amounts of pigment from the iris surface. There is a long pre-glaucomatous phase of the disease in which diagnosis of the condition is possible. Age of onset varies from 2-14 years.

- B. Persistent pupillary membranes (PPM)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

E. Persistent hyaloid artery (PHA)

Congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small patent vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Petersen-Jones SM, Forcier J, Mentzer AL. Ocular melanosis in the Cairn Terrier: clinical description and investigation of mode of inheritance. *Vet Ophthalmol.* 2007;10 Suppl 1:63-69.

# OCULAR DISORDERS REPORT CAIRN TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>3,840 |      | 2016-2020<br>812 |       |
|---------------------|--|--------------------|------|------------------|-------|
|                     |  | #                  | %    | #                | %     |
| <b>GLOBE</b>        |  |                    |      |                  |       |
| 0.110               | microphthalmia   | 1                  | 0.0% | 1                | 0.1%  |
| 10.000              | glaucoma   | 3                  | 0.1% | 0                |       |
| <b>EYELIDS</b>      |  |                    |      |                  |       |
| 25.110              | distichiasis   | 14                 | 0.4% | 7                | 0.9%  |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 1                  | 0.0% | 1                | 0.1%  |
| 40.910              | keratoconjunctivitis sicca                                     | 6                  | 0.2% | 2                | 0.2%  |
| <b>NICTITANS</b>    |  |                    |      |                  |       |
| 51.100              | third eyelid cartilage anomaly                                 | 1                  | 0.0% | 0                |       |
| 52.110              | prolapsed gland of the third eyelid                            | 1                  | 0.0% | 1                | 0.1%  |
| <b>CORNEA</b>       |  |                    |      |                  |       |
| 70.210              | corneal pannus   | 1                  | 0.0% | 0                |       |
| 70.220              | pigmentary keratitis   | 7                  | 0.2% | 0                |       |
| 70.700              | corneal dystrophy  | 25                 | 0.7% | 3                | 0.4%  |
| 70.730              | corneal endothelial degeneration                               | 3                  | 0.1% | 0                |       |
| <b>UVEA</b>         |  |                    |      |                  |       |
| 93.140              | corneal endothelial pigment without PPM                        | 1                  | 0.0% | 0                |       |
| 93.150              | iris coloboma  | 1                  | 0.0% | 1                | 0.1%  |
| 93.710              | persistent pupillary membranes, iris to iris                   | 323                | 8.4% | 129              | 15.9% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 10                 | 0.3% | 5                | 0.6%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 5                  | 0.1% | 0                |       |
| 93.740              | persistent pupillary membranes, iris sheets                    | 2                  | 0.1% | 0                |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 23                 | 0.6% | 31               | 3.8%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 7                  | 0.2% | 9                | 1.1%  |
| 93.810              | uveal melanoma   | 0                  |      | 2                | 0.2%  |
| 93.930              | ocular melanocytosis   | 9                  | 0.2% | 0                |       |
| 93.999              | uveal cysts  | 1                  | 0.0% | 0                |       |
| <b>LENS</b>         |  |                    |      |                  |       |
| 100.200             | cataract, unspecified  | 11                 | 0.3% | 0                |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 224                | 5.8% | 61               | 7.5%  |
| 100.301             | punctate cataract, anterior cortex                             | 25                 | 0.7% | 19               | 2.3%  |
| 100.302             | punctate cataract, posterior cortex                            | 24                 | 0.6% | 11               | 1.4%  |
| 100.303             | punctate cataract, equatorial cortex                           | 14                 | 0.4% | 4                | 0.5%  |
| 100.305             | punctate cataract, posterior sutures                           | 5                  | 0.1% | 0                |       |
| 100.306             | punctate cataract, nucleus                                     | 1                  | 0.0% | 2                | 0.2%  |
| 100.307             | punctate cataract, capsular                                    | 5                  | 0.1% | 7                | 0.9%  |
| 100.311             | incipient cataract, anterior cortex                            | 35                 | 0.9% | 5                | 0.6%  |
| 100.312             | incipient cataract, posterior cortex                           | 59                 | 1.5% | 7                | 0.9%  |
| 100.313             | incipient cataract, equatorial cortex                          | 28                 | 0.7% | 5                | 0.6%  |
| 100.315             | incipient cataract, posterior sutures                          | 10                 | 0.3% | 0                |       |
| 100.316             | incipient cataract, nucleus                                    | 3                  | 0.1% | 2                | 0.2%  |
| 100.317             | incipient cataract, capsular                                   | 5                  | 0.1% | 0                |       |
| 100.321             | incomplete cataract, anterior cortex                           | 10                 | 0.3% | 2                | 0.2%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.322 incomplete cataract, posterior cortex             | 9 0.2%           | 5 0.6%           |
| 100.323 incomplete cataract, equatorial cortex            | 3 0.1%           | 1 0.1%           |
| 100.324 incomplete cataract, anterior sutures             | 0                | 1 0.1%           |
| 100.326 incomplete cataract, nucleus                      | 2 0.1%           | 1 0.1%           |
| 100.328 y-suture tip opacities                            | 1 0.0%           | 1 0.1%           |
| 100.330 generalized/complete cataract                     | 32 0.8%          | 1 0.1%           |
| 100.340 resorbing/hypermature cataract                    | 2 0.1%           | 2 0.2%           |
| 100.345 <i>significant cataracts (summary)</i>            | 284 7.4%         | 76 9.4%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 1 0.0%           | 1 0.1%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 33 0.9%          | 21 2.6%          |
| 110.135 PHPV/PTVL   | 6 0.2%           | 0                |
| 110.320 vitreal degeneration                              | 44 1.1%          | 16 2.0%          |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 2 0.1%           | 0                |
| 97.120 coloboma   | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 20 0.5%          | 2 0.2%           |
| 120.180 retinal dysplasia, geographic                     | 8 0.2%           | 1 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 22 0.6%          | 1 0.1%           |
| 120.960 retinopathy                                       | 1 0.0%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 3 0.1%           | 0                |
| 130.120 optic nerve hypoplasia                            | 8 0.2%           | 0                |
| 130.150 optic disc coloboma                               | 11 0.3%          | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 76 2.0%          | 0                |
| 900.100 other, not inherited                              | 123 3.2%         | 8 1.0%           |
| 900.110 other. suspect not inherited/significance unknown | 121 3.2%         | 30 3.7%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 2,984 77.7%      | 487 60.0%        |

# CANAAN DOG

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| B. | Cataract   | Not defined | 1         | NO              |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary conditions of the Canaan Dog breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT CANAAN DOG

| Diagnostic Name | TOTAL DOGS EXAMINED                                  | 1991-2015<br>509 |       | 2016-2020<br>82 |       |
|-----------------|--|------------------|-------|-----------------|-------|
|                 |  | #                | %     | #               | %     |
| <b>EYELIDS</b>  |  |                  |       |                 |       |
| 25.110          | distichiasis   | 15               | 2.9%  | 1               | 1.2%  |
| <b>CORNEA</b>   |  |                  |       |                 |       |
| 70.700          | corneal dystrophy                                    | 4                | 0.8%  | 0               |       |
| <b>UVEA</b>     |  |                  |       |                 |       |
| 93.710          | persistent pupillary membranes, iris to iris         | 21               | 4.1%  | 2               | 2.4%  |
| 93.740          | persistent pupillary membranes, iris sheets          | 1                | 0.2%  | 0               |       |
| 93.999          | uveal cysts  | 2                | 0.4%  | 0               |       |
| <b>LENS</b>     |  |                  |       |                 |       |
| 100.210         | cataract. suspect not inherited/significance unknown | 18               | 3.5%  | 5               | 6.1%  |
| 100.302         | punctate cataract, posterior cortex                  | 2                | 0.4%  | 0               |       |
| 100.303         | punctate cataract, equatorial cortex                 | 1                | 0.2%  | 0               |       |
| 100.304         | punctate cataract, anterior sutures                  | 1                | 0.2%  | 0               |       |
| 100.306         | punctate cataract, nucleus                           | 3                | 0.6%  | 0               |       |
| 100.307         | punctate cataract, capsular                          | 0                |       | 1               | 1.2%  |
| 100.311         | incipient cataract, anterior cortex                  | 2                | 0.4%  | 1               | 1.2%  |
| 100.312         | incipient cataract, posterior cortex                 | 7                | 1.4%  | 0               |       |
| 100.314         | incipient cataract, anterior sutures                 | 1                | 0.2%  | 0               |       |
| 100.315         | incipient cataract, posterior sutures                | 1                | 0.2%  | 0               |       |
| 100.316         | incipient cataract, nucleus                          | 12               | 2.4%  | 0               |       |
| 100.322         | incomplete cataract, posterior cortex                | 1                | 0.2%  | 0               |       |
| 100.323         | incomplete cataract, equatorial cortex               | 1                | 0.2%  | 0               |       |
| 100.328         | y-suture tip opacities                               | 0                |       | 1               | 1.2%  |
| 100.330         | generalized/complete cataract                        | 13               | 2.6%  | 0               |       |
| 100.345         | significant cataracts (summary)                      | 45               | 8.8%  | 3               | 3.7%  |
| <b>VITREOUS</b> |  |                  |       |                 |       |
| 110.120         | persistent hyaloid artery/remnant                    | 1                | 0.2%  | 0               |       |
| <b>FUNDUS</b>   |  |                  |       |                 |       |
| 97.110          | choroidal hypoplasia                                 | 1                | 0.2%  | 1               | 1.2%  |
| <b>RETINA</b>   |  |                  |       |                 |       |
| 120.170         | retinal dysplasia, folds                             | 2                | 0.4%  | 1               | 1.2%  |
| 120.310         | generalized progressive retinal atrophy (PRA)        | 9                | 1.8%  | 0               |       |
| <b>OTHER</b>    |  |                  |       |                 |       |
| 900.000         | other, unspecified                                   | 6                | 1.2%  | 0               |       |
| 900.100         | other, not inherited                                 | 18               | 3.5%  | 0               |       |
| 900.110         | other. suspect not inherited/significance unknown    | 4                | 0.8%  | 3               | 3.7%  |
| <b>NORMAL</b>   |  |                  |       |                 |       |
| 0.000           | normal globe   | 415              | 81.5% | 69              | 84.1% |

# CANADIAN ESKIMO DOG

| DISORDER  | INHERITANCE | REFERENCE | BREEDING ADVICE |
|---|-------------|-----------|-----------------|
| A. Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Canadian Eskimo Dog breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.



# OCULAR DISORDERS REPORT CANADIAN ESKIMO DOG

| Diagnostic Name | TOTAL DOGS EXAMINED                               | 1991-2015<br>19 |       | 2016-2020<br>27 |       |
|-----------------|---|-----------------|-------|-----------------|-------|
|                 |   | #               | %     | #               | %     |
| <b>CORNEA</b>   |   |                 |       |                 |       |
| 70.700          | corneal dystrophy                                 | 1               | 5.3%  | 0               |       |
| <b>UVEA</b>     |   |                 |       |                 |       |
| 93.710          | persistent pupillary membranes, iris to iris      | 3               | 15.8% | 6               | 22.2% |
| <b>LENS</b>     |   |                 |       |                 |       |
| 100.302         | punctate cataract, posterior cortex               | 0               |       | 1               | 3.7%  |
| 100.307         | punctate cataract, capsular                       | 1               | 5.3%  | 0               |       |
| 100.345         | <i>significant cataracts (summary)</i>            | 1               | 5.3%  | 1               | 3.7%  |
| <b>VITREOUS</b> |   |                 |       |                 |       |
| 110.120         | persistent hyaloid artery/remnant                 | 1               | 5.3%  | 0               |       |
| <b>RETINA</b>   |   |                 |       |                 |       |
| 120.180         | retinal dysplasia, geographic                     | 1               | 5.3%  | 0               |       |
| <b>OTHER</b>    |   |                 |       |                 |       |
| 900.110         | other. suspect not inherited/significance unknown | 2               | 10.5% | 0               |       |
| <b>NORMAL</b>   |   |                 |       |                 |       |
| 0.000           | normal globe                                      | 15              | 78.9% | 20              | 74.1% |

# CANE CORSO

|    | DISORDER                                | INHERITANCE            | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|---|------------------------|-----------|-----------------|--------------------------------------|
| A. | Distichiasis                            | Not defined            | 1         | Breeder option  |                                      |
| B. | Multifocal retinopathy<br>- <i>cmr1</i> | Autosomal<br>recessive | 2, 3      | Breeder option  | Mutation in the<br><i>BEST1</i> gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (*cmr1*) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breed Report.
2. Zangerl B, Wickstrom K, Slavik J, et al. Assessment of canine BEST1 variations identifies new mutations and establishes an independent bestrophinopathy model (cmr3). *Mol Vis.* 2010;16:2791-2804.
3. Guziewicz KE, Slavik J, Lindauer SP et al. Molecular consequences of BEST1 gene mutations in canine multifocal retinopathy predict functional implications for human bestrophinopathies. *IOVS* 52(7) 2011; 4497-505.

# OCULAR DISORDERS REPORT CANE CORSO

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|------------------|--|-----------|-------|-----------|-------|
|                  |  | #         | %     | #         | %     |
| <b>EYELIDS</b>   |  |           |       |           |       |
| 21.000           | entropion, unspecified                                       | 4         | 3.3%  | 2         | 1.3%  |
| 22.000           | ectropion, unspecified                                       | 10        | 8.1%  | 5         | 3.2%  |
| 25.110           | distichiasis   | 5         | 4.1%  | 10        | 6.5%  |
| <b>NICTITANS</b> |  |           |       |           |       |
| 51.100           | third eyelid cartilage anomaly                               | 1         | 0.8%  | 0         |       |
| 52.110           | prolapsed gland of the third eyelid                          | 2         | 1.6%  | 1         | 0.6%  |
| <b>CORNEA</b>    |  |           |       |           |       |
| 70.700           | corneal dystrophy  | 1         | 0.8%  | 0         |       |
| <b>UVEA</b>      |  |           |       |           |       |
| 93.110           | iris hypoplasia  | 0         |       | 1         | 0.6%  |
| 93.710           | persistent pupillary membranes, iris to iris                 | 3         | 2.4%  | 0         |       |
| 93.730           | persistent pupillary membranes, iris to cornea               | 0         |       | 1         | 0.6%  |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands | 1         | 0.8%  | 2         | 1.3%  |
| 93.999           | uveal cysts  | 2         | 1.6%  | 2         | 1.3%  |
| <b>LENS</b>      |  |           |       |           |       |
| 100.210          | cataract. suspect not inherited/significance unknown         | 4         | 3.3%  | 7         | 4.5%  |
| 100.301          | punctate cataract, anterior cortex                           | 1         | 0.8%  | 0         |       |
| 100.302          | punctate cataract, posterior cortex                          | 1         | 0.8%  | 2         | 1.3%  |
| 100.305          | punctate cataract, posterior sutures                         | 0         |       | 1         | 0.6%  |
| 100.328          | y-suture tip opacities                                       | 0         |       | 2         | 1.3%  |
| 100.330          | generalized/complete cataract                                | 1         | 0.8%  | 0         |       |
| 100.345          | significant cataracts (summary)                              | 3         | 2.4%  | 5         | 3.2%  |
| 100.375          | subluxation/luxation, unspecified                            | 0         |       | 1         | 0.6%  |
| <b>VITREOUS</b>  |  |           |       |           |       |
| 110.135          | PHPV/PTVL  | 1         | 0.8%  | 0         |       |
| 110.320          | vitreal degeneration   | 0         |       | 1         | 0.6%  |
| <b>RETINA</b>    |  |           |       |           |       |
| 120.170          | retinal dysplasia, folds                                     | 0         |       | 2         | 1.3%  |
| 120.180          | retinal dysplasia, geographic                                | 0         |       | 1         | 0.6%  |
| 120.310          | generalized progressive retinal atrophy (PRA)                | 0         |       | 1         | 0.6%  |
| 120.960          | retinopathy  | 1         | 0.8%  | 0         |       |
| <b>OTHER</b>     |  |           |       |           |       |
| 900.000          | other, unspecified   | 1         | 0.8%  | 0         |       |
| 900.100          | other, not inherited   | 0         |       | 1         | 0.6%  |
| 900.110          | other. suspect not inherited/significance unknown            | 0         |       | 4         | 2.6%  |
| <b>NORMAL</b>    |  |           |       |           |       |
| 0.000            | normal globe   | 98        | 79.7% | 120       | 77.9% |

## CARDIGAN WELSH CORGI

|    | DISORDER  | INHERITANCE                        | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|---|------------------------------------|-----------|-----------------|--------------------------------------|
| A. | Distichiasis  | Not defined                        | 1         | Breeder option  |                                      |
| B. | Persistent pupillary membranes<br>- iris to iris                  | Not defined                        | 1         | Breeder option  |                                      |
| C. | Cataract  | Not defined                        | 1         | NO              |                                      |
| D. | Retinal atrophy<br>- rod-cone dysplasia<br>type 3 ( <i>rcd3</i> ) | Presumed<br>autosomal<br>recessive | 1, 2-4    | NO              | Mutation in the<br><i>PDE6A</i> gene |

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### Description and Comments

#### A. Distichiasis

Eyelashes abnormally located on the eyelid margin that may cause ocular irritation. Distichiasis may occur any time in the life of the dog. It is difficult to make a strong recommendation about breeding dogs with this entity. The hereditary basis is not known although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

#### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

#### C. Cataract

Lens opacity which may affect one or both eyes and may involve the lens partially or completely. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membranes, persistent hyaloid, or nutritional deficiencies.

D. Retinal atrophy - rod-cone dysplasia type 3 (*rcd3*)

PRA in the Cardigan Welsh Corgi is an autosomal recessive trait caused by a one base pair deletion in the gene encoding the alpha subunit of cyclic GMP phosphodiesterase (*rcd3*). PRA begins early in life with clinical signs of night blindness and a lack of rod ERG responses is seen at 6-8 weeks of age. Dogs are completely blind by 2-3 years of age when ophthalmoscopic signs are first visible. The mutation is found in the *PDE6A* gene. A DNA test is available.

**Historical Note:**

Central progressive retinal atrophy was previously a condition listed for this breed. However as the condition is no longer identified in the breed, the condition has been removed. Central progressive retinal atrophy was a progressive retinal degeneration in which photoreceptor death occurred secondary to disease of the underlying pigment epithelium. Progression was slow and some animals never lost vision. CPRA occurred in England, but was uncommon elsewhere.

**References**

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Petersen-Jones SM, Entz DD, Sargan DR. cGMP phosphodiesterase-alpha mutation causes progressive retinal atrophy in the Cardigan Welsh Corgi dog. *Invest Ophthalmol Vis Sci.* 1999;40:1637-1644.
3. Petersen-Jones SM, Entz DD. An improved DNA-based test for detection of the codon 616 mutation in the alpha cyclic GMP phosphodiesterase gene that causes progressive retinal atrophy in the Cardigan Welsh Corgi. *Vet Ophthalmol.* 2002;5:103-106.
4. Keep JM. Clinical aspects of progressive retinal atrophy in the Cardigan Welsh Corgi. *Aust Vet J.* 1972;48:197-199.

# OCULAR DISORDERS REPORT CARDIGAN WELSH CORGI

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015<br>3,671 |      | 2016-2020<br>557 |      |
|--|---------------------|--------------------|------|------------------|------|
|  |                     | #                  | %    | #                | %    |
| <b>GLOBE</b>   |                     |                    |      |                  |      |
| 0.110 microphthalmia   |                     | 2                  | 0.1% | 0                |      |
| <b>EYELIDS</b>   |                     |                    |      |                  |      |
| 25.110 distichiasis  |                     | 137                | 3.7% | 19               | 3.4% |
| <b>NASOLACRIMAL</b>  |                     |                    |      |                  |      |
| 32.110 imperforate lower nasolacrimal punctum                |                     | 0                  |      | 1                | 0.2% |
| <b>CORNEA</b>  |                     |                    |      |                  |      |
| 70.700 corneal dystrophy                                     |                     | 15                 | 0.4% | 2                | 0.4% |
| 70.730 corneal endothelial degeneration                      |                     | 2                  | 0.1% | 0                |      |
| <b>UVEA</b>  |                     |                    |      |                  |      |
| 93.150 iris coloboma   |                     | 1                  | 0.0% | 0                |      |
| 93.180 liris sphincter dysplasia                             |                     | 1                  | 0.0% | 0                |      |
| 93.710 persistent pupillary membranes, iris to iris          |                     | 105                | 2.9% | 15               | 2.7% |
| 93.720 persistent pupillary membranes, iris to lens          |                     | 3                  | 0.1% | 1                | 0.2% |
| 93.730 persistent pupillary membranes, iris to cornea        |                     | 9                  | 0.2% | 0                |      |
| 93.740 persistent pupillary membranes, iris sheets           |                     | 1                  | 0.0% | 0                |      |
| 93.810 uveal melanoma  |                     | 1                  | 0.0% | 0                |      |
| <b>LENS</b>  |                     |                    |      |                  |      |
| 100.200 cataract, unspecified                                |                     | 15                 | 0.4% | 0                |      |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 108                | 2.9% | 30               | 5.4% |
| 100.301 punctate cataract, anterior cortex                   |                     | 10                 | 0.3% | 1                | 0.2% |
| 100.302 punctate cataract, posterior cortex                  |                     | 11                 | 0.3% | 0                |      |
| 100.303 punctate cataract, equatorial cortex                 |                     | 13                 | 0.4% | 1                | 0.2% |
| 100.304 punctate cataract, anterior sutures                  |                     | 2                  | 0.1% | 0                |      |
| 100.305 punctate cataract, posterior sutures                 |                     | 4                  | 0.1% | 0                |      |
| 100.306 punctate cataract, nucleus                           |                     | 2                  | 0.1% | 0                |      |
| 100.311 incipient cataract, anterior cortex                  |                     | 33                 | 0.9% | 1                | 0.2% |
| 100.312 incipient cataract, posterior cortex                 |                     | 18                 | 0.5% | 4                | 0.7% |
| 100.313 incipient cataract, equatorial cortex                |                     | 15                 | 0.4% | 1                | 0.2% |
| 100.314 incipient cataract, anterior sutures                 |                     | 3                  | 0.1% | 1                | 0.2% |
| 100.315 incipient cataract, posterior sutures                |                     | 2                  | 0.1% | 1                | 0.2% |
| 100.316 incipient cataract, nucleus                          |                     | 7                  | 0.2% | 1                | 0.2% |
| 100.317 incipient cataract, capsular                         |                     | 2                  | 0.1% | 1                | 0.2% |
| 100.321 incomplete cataract, anterior cortex                 |                     | 1                  | 0.0% | 0                |      |
| 100.322 incomplete cataract, posterior cortex                |                     | 0                  |      | 1                | 0.2% |
| 100.328 y-suture tip opacities                               |                     | 3                  | 0.1% | 0                |      |
| 100.330 generalized/complete cataract                        |                     | 8                  | 0.2% | 0                |      |
| 100.340 resorbing/hypermature cataract                       |                     | 1                  | 0.0% | 0                |      |
| 100.345 <i>significant cataracts (summary)</i>               |                     | 150                | 4.1% | 13               | 2.3% |
| <b>VITREOUS</b>  |                     |                    |      |                  |      |
| 110.120 persistent hyaloid artery/remnant                    |                     | 4                  | 0.1% | 1                | 0.2% |
| 110.200 vitreous degeneration-anterior chamber               |                     | 1                  | 0.0% | 1                | 0.2% |
| 110.320 vitreal degeneration                                 |                     | 7                  | 0.2% | 1                | 0.2% |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>FUNDUS</b>   |             |           |
| 97.110 choroidal hypoplasia                               | 3 0.1%      | 0         |
| 97.120 coloboma   | 2 0.1%      | 0         |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 24 0.7%     | 1 0.2%    |
| 120.180 retinal dysplasia, geographic                     | 6 0.2%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 9 0.2%      | 0         |
| 120.400 retinal hemorrhage                                | 1 0.0%      | 0         |
| 120.910 retinal detachment without dialysis               | 2 0.1%      | 0         |
| 120.960 retinopathy                                       | 0           | 1 0.2%    |
| <b>OPTIC NERVE</b>  |             |           |
| 130.120 optic nerve hypoplasia                            | 3 0.1%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 16 0.4%     | 0         |
| 900.100 other, not inherited                              | 39 1.1%     | 1 0.2%    |
| 900.110 other. suspect not inherited/significance unknown | 19 0.5%     | 14 2.5%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 3,232 88.0% | 466 83.7% |



## CAVALIER KING CHARLES SPANIEL

|    | DISORDER                                      | INHERITANCE         | REFERENCE | BREEDING<br>ADVICE |
|----|---|---------------------|-----------|--------------------|
| A. | Microphthalmia with multiple ocular defects   | Not defined         | 1, 2      | NO                 |
| B. | Keratoconjunctivitis sicca                    | Not defined         | 3         | NO                 |
| C. | Congenital KCS and ichthyosiform dermatosis   | Autosomal recessive | 4, 5      | NO                 |
| D. | Distichiasis                                  | Not defined         | 1         | Breeder option     |
| E. | Corneal dystrophy - epithelial/stromal        | Not defined         | 1, 6      | Breeder option     |
| F. | Exposure/pigmentary keratitis                 | Not defined         | 1         | Breeder option     |
| G. | Persistent pupillary membranes - iris to iris | Not defined         | 1         | Breeder option     |
| H. | Cataract                                      | Not defined         | 1, 7      | NO                 |
| I. | Y-suture tip opacity                          | Not defined         | 1         | Breeder option     |
| J. | Vitreous degeneration                         | Not defined         | 1         | Breeder option     |
| K. | Retinal dysplasia - folds                     | Not defined         | 1         | Breeder option     |
| L. | Retinal dysplasia - geographic                | Not defined         | 1         | NO                 |

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### Description and Comments

#### A. Microphthalmia

Microphthalmia is a congenital defect characterized by a small eye often associated with other ocular malformations, including defects of the cornea, anterior chamber, lens and/or retina.

B. Keratoconjunctivitis sicca (KCS)

An abnormality of the tear film, most commonly a deficiency of the aqueous portion, although the mucin and/or lipid layers may be affected; results in ocular irritation and/or vision impairment.

C. Congenital KCS and ichthyosiform dermatosis

A syndrome in which dogs are born with severe to absolute keratoconjunctivitis sicca (KCS) which is poorly responsive to lacrimostimulant treatment. Co-morbid congenital dermatopathy affecting haircoat, skin and footpads is severe and requires intensive life-long care. Clinical signs are so devastating that affected dogs are often euthanized.

D. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

E. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral. In the Cavalier King Charles Spaniel, lesions are circular or semicircular central crystalline deposits in the anterior corneal stroma that appear between 2 and 5 years of age. It may be associated with exophthalmos and lagophthalmos common in these dogs.

F. Exposure/pigmentary keratitis

A condition characterized by variable degrees of superficial vascularization, fibrosis and/or pigmentation of the cornea. May be associated with excessive exposure/irritation of the globe due to shallow orbits, lower eyelid medial entropion, lagophthalmos and macropalpebral fissure.

G. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

H. Cataract

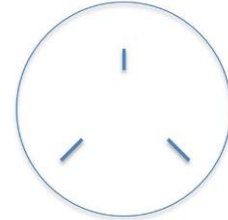
A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely

(diffuse) or in a localized region.

In the Cavalier King Charles Spaniel, onset is at an early age (less than 6 months), affecting the cortex and nucleus with rapid progression to complete cataract, resulting in blindness.

I. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

J. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

K. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

L. Retinal dysplasia – geographic

Abnormal development of the retina present at birth. Any irregularly shaped area of abnormal retinal development containing both areas of thinning and areas of elevation representing folds and retinal disorganization.

## References

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2. Narfstrom K, Dubielzig R. Posterior lenticonus, cataracts and microphthalmia: Congenital defects in the Cavalier King Charles spaniel. *J Small Anim Pract.* 1984;25.
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4. Hartley C, Donaldson D, Smith KC, et al. Congenital keratoconjunctivitis sicca and ichthyosiform dermatosis in 25 Cavalier King Charles spaniel dogs – part I: clinical signs, histopathology, and inheritance. *Vet Ophthalmol.* 2012;15:315-326.
5. Barnett KC. Congenital keratoconjunctivitis sicca and ichthyosiform dermatosis in the Cavalier King Charles Spaniel. *J Small Anim Pract.* 2006;47:524-528.
6. Crispin SM, Barnett KC. Dystrophy, degeneration and infiltration of the canine cornea. *J Small Anim Pract.* 1983;24:63-83.
7. Barnett KC. The diagnosis and differential diagnosis of cataract in the dog. *J Small Anim Pract.* 1985;26:305-316.

# OCULAR DISORDERS REPORT CAVALIER KING CHARLES SPANIEL

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>49,732 |      | 2016-2020<br>15,634 |      |
|---------------------|--|---------------------|------|---------------------|------|
|                     |  | #                   | %    | #                   | %    |
| <b>GLOBE</b>        |  |                     |      |                     |      |
| 0.110               | microphthalmia   | 73                  | 0.1% | 22                  | 0.1% |
| 10.000              | glaucoma   | 3                   | 0.0% | 0                   |      |
| <b>EYELIDS</b>      |  |                     |      |                     |      |
| 20.140              | ectopic cilia  | 3                   | 0.0% | 1                   | 0.0% |
| 20.160              | macropalpebral fissure   | 126                 | 0.3% | 0                   |      |
| 21.000              | entropion, unspecified   | 208                 | 0.4% | 65                  | 0.4% |
| 22.000              | ectropion, unspecified   | 10                  | 0.0% | 1                   | 0.0% |
| 25.110              | distichiasis   | 4,529               | 9.1% | 1,359               | 8.7% |
| <b>NASOLACRIMAL</b> |  |                     |      |                     |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 25                  | 0.1% | 51                  | 0.3% |
| 40.910              | keratoconjunctivitis sicca                                     | 87                  | 0.2% | 37                  | 0.2% |
| <b>NICTITANS</b>    |  |                     |      |                     |      |
| 50.210              | pannus of third eyelid   | 1                   | 0.0% | 1                   | 0.0% |
| 51.100              | third eyelid cartilage anomaly                                 | 6                   | 0.0% | 1                   | 0.0% |
| 52.110              | prolapsed gland of the third eyelid                            | 18                  | 0.0% | 2                   | 0.0% |
| <b>CORNEA</b>       |  |                     |      |                     |      |
| 70.210              | corneal pannus   | 14                  | 0.0% | 4                   | 0.0% |
| 70.220              | pigmentary keratitis   | 245                 | 0.5% | 118                 | 0.8% |
| 70.700              | corneal dystrophy  | 4,415               | 8.9% | 1,245               | 8.0% |
| 70.730              | corneal endothelial degeneration                               | 51                  | 0.1% | 13                  | 0.1% |
| <b>UVEA</b>         |  |                     |      |                     |      |
| 93.110              | iris hypoplasia  | 4                   | 0.0% | 0                   |      |
| 93.140              | corneal endothelial pigment without PPM                        | 7                   | 0.0% | 0                   |      |
| 93.150              | iris coloboma  | 4                   | 0.0% | 1                   | 0.0% |
| 93.180              | iris sphincter dysplasia                                       | 0                   |      | 3                   | 0.0% |
| 93.710              | persistent pupillary membranes, iris to iris                   | 519                 | 1.0% | 191                 | 1.2% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 36                  | 0.1% | 5                   | 0.0% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 33                  | 0.1% | 3                   | 0.0% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 44                  | 0.1% | 1                   | 0.0% |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 28                  | 0.1% | 42                  | 0.3% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 11                  | 0.0% | 2                   | 0.0% |
| 93.999              | uveal cysts  | 22                  | 0.0% | 6                   | 0.0% |
| 97.150              | chorioretinal coloboma, congenital                             | 0                   |      | 10                  | 0.1% |
| <b>LENS</b>         |  |                     |      |                     |      |
| 100.200             | cataract, unspecified  | 57                  | 0.1% | 0                   |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 1,837               | 3.7% | 470                 | 3.0% |
| 100.301             | punctate cataract, anterior cortex                             | 254                 | 0.5% | 100                 | 0.6% |
| 100.302             | punctate cataract, posterior cortex                            | 111                 | 0.2% | 34                  | 0.2% |
| 100.303             | punctate cataract, equatorial cortex                           | 85                  | 0.2% | 26                  | 0.2% |
| 100.304             | punctate cataract, anterior sutures                            | 44                  | 0.1% | 12                  | 0.1% |
| 100.305             | punctate cataract, posterior sutures                           | 104                 | 0.2% | 42                  | 0.3% |
| 100.306             | punctate cataract, nucleus                                     | 115                 | 0.2% | 35                  | 0.2% |
| 100.307             | punctate cataract, capsular                                    | 41                  | 0.1% | 28                  | 0.2% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.311 incipient cataract, anterior cortex               | 361              | 0.7%  | 101              | 0.6%  |
| 100.312 incipient cataract, posterior cortex              | 261              | 0.5%  | 70               | 0.4%  |
| 100.313 incipient cataract, equatorial cortex             | 149              | 0.3%  | 34               | 0.2%  |
| 100.314 incipient cataract, anterior sutures              | 26               | 0.1%  | 5                | 0.0%  |
| 100.315 incipient cataract, posterior sutures             | 76               | 0.2%  | 24               | 0.2%  |
| 100.316 incipient cataract, nucleus                       | 220              | 0.4%  | 48               | 0.3%  |
| 100.317 incipient cataract, capsular                      | 61               | 0.1%  | 14               | 0.1%  |
| 100.321 incomplete cataract, anterior cortex              | 23               | 0.0%  | 20               | 0.1%  |
| 100.322 incomplete cataract, posterior cortex             | 28               | 0.1%  | 40               | 0.3%  |
| 100.323 incomplete cataract, equatorial cortex            | 7                | 0.0%  | 6                | 0.0%  |
| 100.325 incomplete cataract, posterior sutures            | 4                | 0.0%  | 1                | 0.0%  |
| 100.326 incomplete cataract, nucleus                      | 16               | 0.0%  | 22               | 0.1%  |
| 100.327 incomplete cataract, capsular                     | 4                | 0.0%  | 10               | 0.1%  |
| 100.328 y-suture tip opacities                            | 26               | 0.1%  | 66               | 0.4%  |
| 100.330 generalized/complete cataract                     | 217              | 0.4%  | 12               | 0.1%  |
| 100.340 resorbing/hypermature cataract                    | 9                | 0.0%  | 3                | 0.0%  |
| 100.345 <i>significant cataracts (summary)</i>            | 2,299            | 4.6%  | 753              | 4.8%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 16               | 0.0%  | 2                | 0.0%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 82               | 0.2%  | 32               | 0.2%  |
| 110.135 PHPV/PTVL   | 31               | 0.1%  | 3                | 0.0%  |
| 110.200 vitreous degeneration-anterior chamber            | 4                | 0.0%  | 10               | 0.1%  |
| 110.320 vitreal degeneration                              | 221              | 0.4%  | 69               | 0.4%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 8                | 0.0%  | 1                | 0.0%  |
| 97.120 coloboma   | 4                | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 3,617            | 7.3%  | 589              | 3.8%  |
| 120.180 retinal dysplasia, geographic                     | 1,463            | 2.9%  | 266              | 1.7%  |
| 120.190 retinal dysplasia, detached                       | 163              | 0.3%  | 23               | 0.1%  |
| 120.310 generalized progressive retinal atrophy (PRA)     | 151              | 0.3%  | 15               | 0.1%  |
| 120.400 retinal hemorrhage                                | 6                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 20               | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 1                | 0.0%  | 3                | 0.0%  |
| 120.960 retinopathy                                       | 24               | 0.0%  | 35               | 0.2%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 24               | 0.0%  | 7                | 0.0%  |
| 130.120 optic nerve hypoplasia                            | 12               | 0.0%  | 6                | 0.0%  |
| 130.150 optic disc coloboma                               | 22               | 0.0%  | 29               | 0.2%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 596              | 1.2%  | 0                |       |
| 900.100 other, not inherited                              | 1,138            | 2.3%  | 39               | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown | 574              | 1.2%  | 739              | 4.7%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 36,151           | 72.7% | 10,582           | 67.7% |

# **OCULAR DISORDERS REPORT CESKY TERRIER**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the CESKY TERRIER breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT CESKY TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|---------------------|--|-----------|-------|-----------|-------|
|                     |  | #         | %     | #         | %     |
| <b>EYELIDS</b>      |  |           |       |           |       |
| 25.110              | distichiasis   | 19        | 16.2% | 0         |       |
| <b>NASOLACRIMAL</b> |  |           |       |           |       |
| 32.110              | imperforate lower nasolacrimal punctum                       | 1         | 0.9%  | 0         |       |
| <b>CORNEA</b>       |  |           |       |           |       |
| 70.700              | corneal dystrophy  | 8         | 6.8%  | 0         |       |
| <b>UVEA</b>         |  |           |       |           |       |
| 93.710              | persistent pupillary membranes, iris to iris                 | 3         | 2.6%  | 3         | 15.8% |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands | 1         | 0.9%  | 0         |       |
| 97.150              | chorioretinal coloboma, congenital                           | 1         | 0.9%  | 0         |       |
| <b>LENS</b>         |  |           |       |           |       |
| 100.200             | cataract, unspecified  | 1         | 0.9%  | 0         |       |
| 100.210             | cataract. suspect not inherited/significance unknown         | 1         | 0.9%  | 0         |       |
| 100.301             | punctate cataract, anterior cortex                           | 1         | 0.9%  | 0         |       |
| 100.307             | punctate cataract, capsular                                  | 2         | 1.7%  | 0         |       |
| 100.311             | incipient cataract, anterior cortex                          | 1         | 0.9%  | 0         |       |
| 100.312             | incipient cataract, posterior cortex                         | 1         | 0.9%  | 1         | 5.3%  |
| 100.345             | <i>significant cataracts (summary)</i>                       | 6         | 5.1%  | 1         | 5.3%  |
| <b>FUNDUS</b>       |  |           |       |           |       |
| 97.110              | choroidal hypoplasia   | 1         | 0.9%  | 0         |       |
| <b>RETINA</b>       |  |           |       |           |       |
| 120.170             | retinal dysplasia, folds                                     | 8         | 6.8%  | 0         |       |
| 120.910             | retinal detachment without dialysis                          | 1         | 0.9%  | 0         |       |
| <b>OPTIC NERVE</b>  |  |           |       |           |       |
| 130.110             | micropapilla   | 1         | 0.9%  | 0         |       |
| <b>OTHER</b>        |  |           |       |           |       |
| 900.000             | other, unspecified   | 1         | 0.9%  | 0         |       |
| 900.100             | other, not inherited   | 4         | 3.4%  | 0         |       |
| 900.110             | other. suspect not inherited/significance unknown            | 1         | 0.9%  | 0         |       |
| <b>NORMAL</b>       |  |           |       |           |       |
| 0.000               | normal globe   | 79        | 67.5% | 15        | 78.9% |



# CHESAPEAKE BAY RETRIEVER

|    | DISORDER                        | INHERITANCE                  | REFERENCE | BREEDING ADVICE         | GENETIC TESTS AVAILABLE          |
|----|---------------------------------|------------------------------|-----------|-------------------------|----------------------------------|
| A. | Distichiasis                    | Not defined                  | 1         | Breeder option          |                                  |
| B. | Persistent pupillary membranes  |                              |           |                         |                                  |
|    | - iris to iris                  | Not defined                  | 1         | Breeder option          |                                  |
|    | - lens pigment foci/no strands  | Not defined                  | 1         | Passes with no notation |                                  |
| C. | Cataract                        | Presumed incomplete dominant | 1, 2      | NO                      |                                  |
| D. | Retinal atrophy ( <i>prcd</i> ) | Autosomal recessive          | 1, 3      | NO                      | Mutation in the <i>prcd</i> gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located in the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

Hereditary cataracts have been described in the Chesapeake Bay Retriever and affect the young adult dog. They appear as posterior cortical, axial, triangular opacities and the Y suture tips can be affected in both the anterior and posterior cortices. Extension of the cataract into the posterior cortex and progression to impair vision can occur. An autosomal dominant inheritance with incomplete penetrance has been proposed; however, the genetics have not been completely defined and additional studies will be required.

D. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Chesapeake Bay Retriever is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available. It is important to note that in all breeds in which a molecular diagnostic test for the disease is available, it is possible to have dogs diagnosed clinically as affected, yet the DNA test results are normal. This suggests that other genetic causes of PRA exist or that the diagnosed affected dog has an acquired disease that mimics the inherited disorder.

A second, less common form of PRA is also present in the Chesapeake Bay Retriever with ophthalmoscopic abnormalities characteristic of mid-stage disease found in dogs between 8-12 months of age. The lesions are progressive and end-stage lesions are evident by 2-3 years of age. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gelatt KN. Cataracts in Chesapeake Bay retrievers. *J Am Vet Med Assoc.* 1979;175:1176-1178.
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# OCULAR DISORDERS REPORT CHESAPEAKE BAY RETRIEVER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>12,682 |      | 2016-2020<br>1,866 |      |
|---------------------|--|---------------------|------|--------------------|------|
|                     |  | #                   | %    | #                  | %    |
| <b>GLOBE</b>        |  |                     |      |                    |      |
| 0.110               | microphthalmia   | 7                   | 0.1% | 1                  | 0.1% |
| 10.000              | glaucoma   | 4                   | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                     |      |                    |      |
| 20.140              | ectopic cilia  | 2                   | 0.0% | 0                  |      |
| 20.160              | macropalpebral fissure   | 3                   | 0.0% | 0                  |      |
| 21.000              | entropion, unspecified   | 54                  | 0.4% | 4                  | 0.2% |
| 22.000              | ectropion, unspecified   | 7                   | 0.1% | 0                  |      |
| 25.110              | distichiasis   | 913                 | 7.2% | 171                | 9.2% |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 0                   |      | 1                  | 0.1% |
| <b>NICTITANS</b>    |  |                     |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 2                   | 0.0% | 1                  | 0.1% |
| 52.110              | prolapsed gland of the third eyelid                            | 2                   | 0.0% | 0                  |      |
| <b>CORNEA</b>       |  |                     |      |                    |      |
| 70.210              | corneal pannus   | 1                   | 0.0% | 0                  |      |
| 70.700              | corneal dystrophy  | 76                  | 0.6% | 14                 | 0.8% |
| 70.730              | corneal endothelial degeneration                               | 1                   | 0.0% | 0                  |      |
| <b>UVEA</b>         |  |                     |      |                    |      |
| 93.110              | iris hypoplasia  | 0                   |      | 1                  | 0.1% |
| 93.150              | iris coloboma  | 1                   | 0.0% | 0                  |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 221                 | 1.7% | 57                 | 3.1% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 11                  | 0.1% | 0                  |      |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 3                   | 0.0% | 0                  |      |
| 93.740              | persistent pupillary membranes, iris sheets                    | 14                  | 0.1% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 41                  | 0.3% | 57                 | 3.1% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 4                   | 0.0% | 0                  |      |
| 93.810              | uveal melanoma   | 1                   | 0.0% | 0                  |      |
| 93.999              | uveal cysts  | 25                  | 0.2% | 11                 | 0.6% |
| <b>LENS</b>         |  |                     |      |                    |      |
| 100.200             | cataract, unspecified  | 74                  | 0.6% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 537                 | 4.2% | 101                | 5.4% |
| 100.301             | punctate cataract, anterior cortex                             | 44                  | 0.3% | 13                 | 0.7% |
| 100.302             | punctate cataract, posterior cortex                            | 109                 | 0.9% | 20                 | 1.1% |
| 100.303             | punctate cataract, equatorial cortex                           | 34                  | 0.3% | 5                  | 0.3% |
| 100.304             | punctate cataract, anterior sutures                            | 9                   | 0.1% | 5                  | 0.3% |
| 100.305             | punctate cataract, posterior sutures                           | 40                  | 0.3% | 3                  | 0.2% |
| 100.306             | punctate cataract, nucleus                                     | 7                   | 0.1% | 3                  | 0.2% |
| 100.307             | punctate cataract, capsular                                    | 16                  | 0.1% | 15                 | 0.8% |
| 100.311             | incipient cataract, anterior cortex                            | 52                  | 0.4% | 12                 | 0.6% |
| 100.312             | incipient cataract, posterior cortex                           | 218                 | 1.7% | 33                 | 1.8% |
| 100.313             | incipient cataract, equatorial cortex                          | 53                  | 0.4% | 7                  | 0.4% |
| 100.314             | incipient cataract, anterior sutures                           | 7                   | 0.1% | 0                  |      |
| 100.315             | incipient cataract, posterior sutures                          | 44                  | 0.3% | 7                  | 0.4% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.316 incipient cataract, nucleus                       | 18 0.1%          | 2 0.1%           |
| 100.317 incipient cataract, capsular                      | 22 0.2%          | 4 0.2%           |
| 100.321 incomplete cataract, anterior cortex              | 1 0.0%           | 0                |
| 100.322 incomplete cataract, posterior cortex             | 3 0.0%           | 5 0.3%           |
| 100.323 incomplete cataract, equatorial cortex            | 0                | 2 0.1%           |
| 100.325 incomplete cataract, posterior sutures            | 2 0.0%           | 2 0.1%           |
| 100.326 incomplete cataract, nucleus                      | 0                | 1 0.1%           |
| 100.328 y-suture tip opacities                            | 6 0.0%           | 10 0.5%          |
| 100.330 generalized/complete cataract                     | 43 0.3%          | 1 0.1%           |
| 100.345 <i>significant cataracts (summary)</i>            | 802 6.3%         | 150 8.0%         |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 7 0.1%           | 1 0.1%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 20 0.2%          | 2 0.1%           |
| 110.135 PHPV/PTVL   | 10 0.1%          | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 17 0.1%          | 15 0.8%          |
| 110.320 vitreal degeneration                              | 72 0.6%          | 12 0.6%          |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 3 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 80 0.6%          | 7 0.4%           |
| 120.180 retinal dysplasia, geographic                     | 49 0.4%          | 5 0.3%           |
| 120.190 retinal dysplasia, detached                       | 2 0.0%           | 1 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 91 0.7%          | 5 0.3%           |
| 120.400 retinal hemorrhage                                | 1 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| 120.960 retinopathy                                       | 1 0.0%           | 10 0.5%          |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 1 0.0%           | 0                |
| 130.120 optic nerve hypoplasia                            | 2 0.0%           | 0                |
| 130.150 optic disc coloboma                               | 2 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 127 1.0%         | 0                |
| 900.100 other, not inherited                              | 331 2.6%         | 7 0.4%           |
| 900.110 other. suspect not inherited/significance unknown | 142 1.1%         | 112 6.0%         |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 10,406 82.1%     | 1,293 69.3%      |

# CHIHUAHUA

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TEST<br>AVAILABLE           |
|----|---|------------------------|-----------|--------------------|-------------------------------------|
| A. | Distichiasis  | Not defined            | 1         | Breeder option     |                                     |
| B. | Corneal dystrophy<br>- endothelial                  | Not defined            | 1, 2      | NO                 |                                     |
| C. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined            | 1         | Breeder option     |                                     |
| D. | Cataract  | Not defined            | 1         | NO                 |                                     |
| E. | Vitreous<br>degeneration                            | Not defined            | 1         | Breeder option     |                                     |
| F. | Retinal atrophy<br>( <i>prcd</i> )                  | Autosomal<br>recessive | 3, 4      | NO                 | Mutation in the <i>prcd</i><br>gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Corneal dystrophy - endothelial

An abnormal loss of the inner lining of the cornea that causes progressive fluid retention (edema). With time the edema results in keratitis and decreased vision. This usually does not occur until the animal is older.

In the Chihuahua, this is a primary degenerative endothelial disease leading to progressive and permanent corneal edema. It is suspected to be a heritable disorder. There is no sex predilection. The condition is observed in older dogs, 6 to 13 years of age with a mean of 9.5 years. The corneal edema starts asymptotically in the dorsal temporal corneal quadrant of one eye and slowly progresses medially, eventually involving the entire cornea. Typically, it becomes bilateral. In the later stages, discomfort, intracorneal bullae with subsequent ulceration and keratoconus may develop. Histologically, the primary endothelial disease appears slightly different from the

clinically similar disorder of the Boston Terrier.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

F. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as Progressive Retinal Atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Chihuahua is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Martin CL and Dice PF. Corneal endothelial dystrophy in the dog. *J Am Anim Hosp Assoc.* 1982;18:327.
3. Hyama M, Tada N, Mitsui H, et al. Real-time PCR genotyping in assay for canine progressive rod-cone degeneration and mutant allele frequency in Toy Poodles, Chihuahuas, and Miniature Dachshunds in Japan. *J Vet Med Sci* 2016; 78(3): 481.

4. Downs LM, Hitti R, Pregolato S, et al. Genetic screening for PRA-associated mutations in multiple dog breeds shows that PRA is heterogeneous within and between breeds. *Vet Ophthalmol.* 2014;17:126-130.

# OCULAR DISORDERS REPORT CHIHUAHUA

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 0.110 microphthalmia  |                     | 0         |      | 1         | 0.1% |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 20.140 ectopic cilia  |                     | 1         | 0.1% | 0         |      |
| 21.000 entropion, unspecified   |                     | 3         | 0.2% | 3         | 0.3% |
| 25.110 distichiasis   |                     | 80        | 5.4% | 36        | 3.4% |
| <b>NASOLACRIMAL</b>   |                     |           |      |           |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 3         | 0.2% | 3         | 0.3% |
| 40.910 keratoconjunctivitis sicca                                     |                     | 1         | 0.1% | 2         | 0.2% |
| <b>NICTITANS</b>  |                     |           |      |           |      |
| 52.110 prolapsed gland of the third eyelid                            |                     | 4         | 0.3% | 2         | 0.2% |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.220 pigmentary keratitis   |                     | 2         | 0.1% | 3         | 0.3% |
| 70.700 corneal dystrophy  |                     | 3         | 0.2% | 3         | 0.3% |
| 70.730 corneal endothelial degeneration                               |                     | 5         | 0.3% | 3         | 0.3% |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 112       | 7.6% | 55        | 5.2% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 4         | 0.3% | 0         |      |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 2         | 0.1% | 0         |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 7         | 0.5% | 4         | 0.4% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 2         | 0.1% | 0         |      |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.200 cataract, unspecified   |                     | 3         | 0.2% | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 44        | 3.0% | 21        | 2.0% |
| 100.301 punctate cataract, anterior cortex                            |                     | 7         | 0.5% | 6         | 0.6% |
| 100.302 punctate cataract, posterior cortex                           |                     | 0         |      | 1         | 0.1% |
| 100.303 punctate cataract, equatorial cortex                          |                     | 2         | 0.1% | 0         |      |
| 100.304 punctate cataract, anterior sutures                           |                     | 1         | 0.1% | 0         |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 3         | 0.2% | 0         |      |
| 100.306 punctate cataract, nucleus                                    |                     | 1         | 0.1% | 0         |      |
| 100.307 punctate cataract, capsular                                   |                     | 1         | 0.1% | 3         | 0.3% |
| 100.311 incipient cataract, anterior cortex                           |                     | 25        | 1.7% | 7         | 0.7% |
| 100.312 incipient cataract, posterior cortex                          |                     | 16        | 1.1% | 5         | 0.5% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 6         | 0.4% | 3         | 0.3% |
| 100.314 incipient cataract, anterior sutures                          |                     | 1         | 0.1% | 0         |      |
| 100.315 incipient cataract, posterior sutures                         |                     | 1         | 0.1% | 1         | 0.1% |
| 100.316 incipient cataract, nucleus                                   |                     | 6         | 0.4% | 3         | 0.3% |
| 100.317 incipient cataract, capsular                                  |                     | 2         | 0.1% | 3         | 0.3% |
| 100.321 incomplete cataract, anterior cortex                          |                     | 2         | 0.1% | 3         | 0.3% |
| 100.325 incomplete cataract, posterior sutures                        |                     | 1         | 0.1% | 0         |      |
| 100.326 incomplete cataract, nucleus                                  |                     | 1         | 0.1% | 2         | 0.2% |
| 100.328 y-suture tip opacities  |                     | 1         | 0.1% | 0         |      |
| 100.330 generalized/complete cataract                                 |                     | 12        | 0.8% | 0         |      |
| 100.345 significant cataracts (summary)                               |                     | 92        | 6.2% | 37        | 3.5% |



| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.375 subluxation/luxation, unspecified                 | 1 0.1%           | 2 0.2%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 2 0.1%           | 0                |
| 110.135 PHPV/PTVL   | 2 0.1%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 5 0.3%           | 15 1.4%          |
| 110.320 vitreal degeneration                              | 58 3.9%          | 15 1.4%          |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 1 0.1%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 7 0.5%           | 2 0.2%           |
| 120.180 retinal dysplasia, geographic                     | 3 0.2%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 11 0.7%          | 1 0.1%           |
| 120.960 retinopathy                                       | 1 0.1%           | 1 0.1%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 1 0.1%           | 1 0.1%           |
| 130.120 optic nerve hypoplasia                            | 0                | 1 0.1%           |
| 130.150 optic disc coloboma                               | 1 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 21 1.4%          | 0                |
| 900.100 other, not inherited                              | 23 1.6%          | 1 0.1%           |
| 900.110 other. suspect not inherited/significance unknown | 25 1.7%          | 41 3.9%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,179 79.9%      | 854 81.0%        |

## CHINESE CRESTED

|    | DISORDER   | INHERITANCE                  | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|--|------------------------------|-----------|-----------------|--------------------------------------|
| A. | Distichiasis   | Not defined                  | 1         | Breeder option  |                                      |
| B. | Persistent pupillary membranes<br>- iris to iris               | Not defined                  | 1         | Breeder option  |                                      |
| C. | Cataract   | Not defined                  | 4         | NO              |                                      |
| D. | Lens luxation  | Autosomal recessive          | 2, 3      | NO              | Mutation in the <i>ADAMTS17</i> gene |
| E. | Vitreous degeneration  | Not defined                  | 1, 3-5    | Breeder option  |                                      |
| F. | Retinal atrophy<br>( <i>prcd</i> )                             | Autosomal recessive          | 1, 5      | NO              | Mutation in the <i>prcd</i> gene     |
| G. | Retinal atrophy<br>- rod-cone dysplasia type 3 ( <i>rcd3</i> ) | Presumed autosomal recessive | 5         | NO              | Mutation in the PDE6A gene           |

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### Description and Comments

#### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

#### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Lens luxation

Partial (subluxation) or complete displacement of the lens from its normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

E. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

F. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as Progressive Retinal Atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Chinese Crested is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

In the Chinese Crested, a second, but very infrequency type of PRA has been identified that is caused by the mutation in the *PDE6A* gene that causes PRA in Cardigan Welsh Corgis. However, most cases of PRA that test normal for the *prcd* gene defect likely results from a gene defect that is still to be identified.

G. Retinal atrophy - rod-cone dysplasia type 3 (*rcd3*)

PRA in the Chinese Crested is an autosomal recessive trait caused by a one base pair deletion in the gene encoding the alpha subunit of cyclic GMP phosphodiesterase (*rcd3*). PRA begins early in life with clinical signs of night blindness and a lack of rod ERG responses is seen at 6-8 weeks of age. Dogs are completely blind by 2-3 years of age when ophthalmoscopic signs are first visible. The mutation is found in the *PDE6A* gene. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Farias FH, Johnson GS, Taylor JF, et al. An ADAMTS17 splice donor site mutation in dogs with primary lens luxation. *Invest Ophthalmol Vis Sci.* 2010;51:4716-4721.
3. Gould D, Pettitt L, McLaughlin B, et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011;14:378-384.
4. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics.* 2006;88:551-563. PMID: 16938425
5. Downs LM, Hitti R, Pregnotato S, et al. Genetic screening for PRA-associated mutations in multiple dog breeds shows that PRA is heterogeneous within and between breeds. *Vet Ophthalmol.* 2014; 17:126-130.

# OCULAR DISORDERS REPORT CHINESE CRESTED

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>6,493 |      | 2016-2020<br>504 |      |
|---------------------|--|--------------------|------|------------------|------|
|                     |  | #                  | %    | #                | %    |
| <b>GLOBE</b>        |  |                    |      |                  |      |
| 0.110               | microphthalmia   | 4                  | 0.1% | 0                |      |
| 10.000              | glaucoma   | 2                  | 0.0% | 0                |      |
| <b>EYELIDS</b>      |  |                    |      |                  |      |
| 20.140              | ectopic cilia  | 2                  | 0.0% | 0                |      |
| 21.000              | entropion, unspecified   | 4                  | 0.1% | 0                |      |
| 25.110              | distichiasis   | 39                 | 0.6% | 5                | 1.0% |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 3                  | 0.0% | 2                | 0.4% |
| 40.910              | keratoconjunctivitis sicca                                     | 18                 | 0.3% | 0                |      |
| <b>NICTITANS</b>    |  |                    |      |                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 3                  | 0.0% | 0                |      |
| <b>CORNEA</b>       |  |                    |      |                  |      |
| 70.210              | corneal pannus   | 5                  | 0.1% | 0                |      |
| 70.220              | pigmentary keratitis   | 7                  | 0.1% | 1                | 0.2% |
| 70.700              | corneal dystrophy  | 34                 | 0.5% | 3                | 0.6% |
| 70.730              | corneal endothelial degeneration                               | 2                  | 0.0% | 1                | 0.2% |
| <b>UVEA</b>         |  |                    |      |                  |      |
| 93.110              | iris hypoplasia  | 5                  | 0.1% | 0                |      |
| 93.150              | iris coloboma  | 2                  | 0.0% | 0                |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 164                | 2.5% | 17               | 3.4% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 11                 | 0.2% | 0                |      |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 10                 | 0.2% | 0                |      |
| 93.740              | persistent pupillary membranes, iris sheets                    | 5                  | 0.1% | 0                |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 3                  | 0.0% | 0                |      |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 2                  | 0.0% | 1                | 0.2% |
| 93.999              | uveal cysts  | 4                  | 0.1% | 0                |      |
| <b>LENS</b>         |  |                    |      |                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 152                | 2.3% | 19               | 3.8% |
| 100.301             | punctate cataract, anterior cortex                             | 29                 | 0.4% | 8                | 1.6% |
| 100.302             | punctate cataract, posterior cortex                            | 17                 | 0.3% | 1                | 0.2% |
| 100.303             | punctate cataract, equatorial cortex                           | 12                 | 0.2% | 2                | 0.4% |
| 100.304             | punctate cataract, anterior sutures                            | 2                  | 0.0% | 1                | 0.2% |
| 100.305             | punctate cataract, posterior sutures                           | 6                  | 0.1% | 1                | 0.2% |
| 100.306             | punctate cataract, nucleus                                     | 8                  | 0.1% | 1                | 0.2% |
| 100.307             | punctate cataract, capsular                                    | 5                  | 0.1% | 3                | 0.6% |
| 100.311             | incipient cataract, anterior cortex                            | 41                 | 0.6% | 3                | 0.6% |
| 100.312             | incipient cataract, posterior cortex                           | 30                 | 0.5% | 1                | 0.2% |
| 100.313             | incipient cataract, equatorial cortex                          | 29                 | 0.4% | 1                | 0.2% |
| 100.314             | incipient cataract, anterior sutures                           | 2                  | 0.0% | 0                |      |
| 100.315             | incipient cataract, posterior sutures                          | 5                  | 0.1% | 1                | 0.2% |
| 100.316             | incipient cataract, nucleus                                    | 5                  | 0.1% | 0                |      |
| 100.317             | incipient cataract, capsular                                   | 1                  | 0.0% | 1                | 0.2% |
| 100.321             | incomplete cataract, anterior cortex                           | 2                  | 0.0% | 2                | 0.4% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.322 incomplete cataract, posterior cortex             | 2 0.0%           | 2 0.4%           |
| 100.323 incomplete cataract, equatorial cortex            | 1 0.0%           | 0                |
| 100.326 incomplete cataract, nucleus                      | 0                | 1 0.2%           |
| 100.327 incomplete cataract, capsular                     | 0                | 1 0.2%           |
| 100.328 y-suture tip opacities                            | 2 0.0%           | 1 0.2%           |
| 100.330 generalized/complete cataract                     | 26 0.4%          | 1 0.2%           |
| 100.340 resorbing/hypermature cataract                    | 0                | 2 0.4%           |
| 100.345 <i>significant cataracts (summary)</i>            | 225 3.5%         | 34 6.7%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 25 0.4%          | 5 1.0%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 6 0.1%           | 2 0.4%           |
| 110.135 PHPV/PTVL   | 2 0.0%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 20 0.3%          | 25 5.0%          |
| 110.320 vitreal degeneration                              | 736 11.3%        | 30 6.0%          |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 3 0.0%           | 0                |
| 97.120 coloboma   | 2 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 31 0.5%          | 1 0.2%           |
| 120.180 retinal dysplasia, geographic                     | 6 0.1%           | 0                |
| 120.190 retinal dysplasia, detached                       | 2 0.0%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 96 1.5%          | 3 0.6%           |
| 120.400 retinal hemorrhage                                | 4 0.1%           | 0                |
| 120.910 retinal detachment without dialysis               | 8 0.1%           | 0                |
| 120.960 retinopathy                                       | 2 0.0%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 4 0.1%           | 0                |
| 130.120 optic nerve hypoplasia                            | 13 0.2%          | 0                |
| 130.150 optic disc coloboma                               | 8 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 68 1.0%          | 0                |
| 900.100 other, not inherited                              | 152 2.3%         | 2 0.4%           |
| 900.110 other. suspect not inherited/significance unknown | 44 0.7%          | 20 4.0%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 5,513 84.9%      | 391 77.6%        |

# CHINESE FOO DOG

|    | DISORDER      | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|---------------|------------------------|-----------|--------------------|---|
| A. | Lens luxation | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>ADAMTS17</i> gene |

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## Description and Comments

### A. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Chinese Foo Dog. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011; 14: 378-384.

## CHINESE SHAR-PEI

|    | DISORDER                      | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|-------------------------------|---------------------|-----------|-----------------|--------------------------------------|
| A. | Glaucoma – POAG               | Autosomal recessive | 2         | NO              | Mutation in the <i>ADAMTS17</i> gene |
| B. | Entropion                     | Not defined         | 1, 3-6    | NO              |                                      |
| C. | Secondary keratitis - chronic | Not defined         | 1         | Breeder option  |                                      |
| D. | Lens luxation                 | Autosomal recessive | 1, 7      | NO              | Mutation in the <i>ADAMTS17</i> gene |

### Description and Comments

#### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of the intraocular pressure (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening exam.

A 6 base pair deletion in exon 22 of *ADAMTS17* has been found in some affected Chinese Shar-Pei. Results supported phenotype is an autosomal recessive trait. A genetic test is available.

#### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

The condition is a particularly severe problem in the Chinese Shar-Pei and is compounded by breeder selection for facial conformation with heavy skin folds which encourages formation of entropion.

#### C. Secondary keratitis - chronic

A specific designation does not exist on the CAER form for this condition. We ask examiners to mark other – unlisted conditions suspected as inherited. Then in the comments box please write secondary keratitis – chronic.



A condition characterized by variable degrees of superficial vascularization, fibrosis and/or pigmentation of the cornea. Often associated with entropion or a combination of entropion and ectropion.

D. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness.

A 6 base pair deletion in exon 22 of *ADAMTS17* has been found in some affected Chinese Shar-Pei. Results supported phenotype is an autosomal recessive trait. A genetic test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Oliver, JAC, Rustidge S, Pettit L, et al. Evaluation of *ADAMTS17* in Chinese Shar-Pei with primary open-angle glaucoma, primary lens luxation, or both. *Am J Vet Res.* 2018 Jan; 79(1): 98-106.
3. Lenarduzzi R. Management of eyelid problems in Chinese Shar-Pei puppies. *Vet Med Small Anim Clin.* 1983;78:548-550.
4. Bedford PGC. Entropion in Shar-Peis (Correspondence). *Vet Rec.* 1984;115:666.
5. Startup FG. Entropion in the Shar-Pei (Correspondence). *Vet Rec.* 1985;116:57.
6. Barnett KC. Inherited eye disease in the dog and cat. *J Small Anim Pract.* 1988;29:462-475.
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# OCULAR DISORDERS REPORT CHINESE SHAR-PEI

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015<br>603 |       | 2016-2020<br>94 |       |
|------------------|--|------------------|-------|-----------------|-------|
|                  |  | #                | %     | #               | %     |
| <b>GLOBE</b>     |  |                  |       |                 |       |
| 0.110            | microphthalmia   | 1                | 0.2%  | 1               | 1.1%  |
| 10.000           | glaucoma   | 0                |       | 1               | 1.1%  |
| <b>EYELIDS</b>   |  |                  |       |                 |       |
| 21.000           | entropion, unspecified   | 306              | 50.7% | 27              | 28.7% |
| 22.000           | ectropion, unspecified   | 12               | 2.0%  | 0               |       |
| 25.110           | distichiasis   | 3                | 0.5%  | 0               |       |
| <b>NICTITANS</b> |  |                  |       |                 |       |
| 51.100           | third eyelid cartilage anomaly                                 | 1                | 0.2%  | 1               | 1.1%  |
| 52.110           | prolapsed gland of the third eyelid                            | 2                | 0.3%  | 1               | 1.1%  |
| <b>CORNEA</b>    |  |                  |       |                 |       |
| 70.210           | corneal pannus   | 29               | 4.8%  | 0               |       |
| 70.220           | pigmentary keratitis   | 11               | 1.8%  | 10              | 10.6% |
| 70.700           | corneal dystrophy  | 4                | 0.7%  | 0               |       |
| 70.730           | corneal endothelial degeneration                               | 6                | 1.0%  | 1               | 1.1%  |
| <b>UVEA</b>      |  |                  |       |                 |       |
| 93.710           | persistent pupillary membranes, iris to iris                   | 15               | 2.5%  | 1               | 1.1%  |
| 93.720           | persistent pupillary membranes, iris to lens                   | 5                | 0.8%  | 0               |       |
| 93.730           | persistent pupillary membranes, iris to cornea                 | 5                | 0.8%  | 3               | 3.2%  |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands   | 2                | 0.3%  | 2               | 2.1%  |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands | 1                | 0.2%  | 0               |       |
| 93.810           | uveal melanoma   | 1                | 0.2%  | 0               |       |
| <b>LENS</b>      |  |                  |       |                 |       |
| 100.200          | cataract, unspecified  | 4                | 0.7%  | 0               |       |
| 100.210          | cataract. suspect not inherited/significance unknown           | 13               | 2.2%  | 3               | 3.2%  |
| 100.301          | punctate cataract, anterior cortex                             | 2                | 0.3%  | 0               |       |
| 100.302          | punctate cataract, posterior cortex                            | 1                | 0.2%  | 0               |       |
| 100.305          | punctate cataract, posterior sutures                           | 2                | 0.3%  | 0               |       |
| 100.306          | punctate cataract, nucleus                                     | 1                | 0.2%  | 0               |       |
| 100.307          | punctate cataract, capsular                                    | 1                | 0.2%  | 0               |       |
| 100.311          | incipient cataract, anterior cortex                            | 2                | 0.3%  | 0               |       |
| 100.312          | incipient cataract, posterior cortex                           | 5                | 0.8%  | 1               | 1.1%  |
| 100.313          | incipient cataract, equatorial cortex                          | 0                |       | 1               | 1.1%  |
| 100.314          | incipient cataract, anterior sutures                           | 1                | 0.2%  | 0               |       |
| 100.315          | incipient cataract, posterior sutures                          | 2                | 0.3%  | 0               |       |
| 100.316          | incipient cataract, nucleus                                    | 1                | 0.2%  | 0               |       |
| 100.330          | generalized/complete cataract                                  | 2                | 0.3%  | 0               |       |
| 100.345          | significant cataracts (summary)                                | 24               | 4.0%  | 2               | 2.1%  |
| 100.375          | subluxation/luxation, unspecified                              | 9                | 1.5%  | 0               |       |
| <b>VITREOUS</b>  |  |                  |       |                 |       |
| 110.120          | persistent hyaloid artery/remnant                              | 1                | 0.2%  | 0               |       |
| 110.320          | vitreal degeneration   | 1                | 0.2%  | 0               |       |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 4 0.7%    | 0         |
| 120.180 retinal dysplasia, geographic                     | 1 0.2%    | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 2 0.3%    | 0         |
| 120.910 retinal detachment without dialysis               | 1 0.2%    | 0         |
| <b>OPTIC NERVE</b>  |           |           |
| 130.120 optic nerve hypoplasia                            | 1 0.2%    | 0         |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 9 1.5%    | 0         |
| 900.100 other, not inherited                              | 17 2.8%   | 1 1.1%    |
| 900.110 other. suspect not inherited/significance unknown | 21 3.5%   | 7 7.4%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 286 47.4% | 48 51.1%  |

# CHINOOK

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--------------------------------|-------------|-----------|-----------------|
| A. | Persistent pupillary membranes | Not defined | 1         | Breeder option  |
|    | - iris to iris                 | Not defined | 1         | Breeder option  |
| B. | Cataract                       | Not defined | 1         | NO              |
| C. | Retinal dysplasia              | Not defined | 1         | Breeder option  |
|    | - folds                        |             |           |                 |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT CHINOOK

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>1,396 |      | 2016-2020<br>262 |      |
|---------------------|--|--------------------|------|------------------|------|
|                     |  | #                  | %    | #                | %    |
| <b>EYELIDS</b>      |  |                    |      |                  |      |
| 20.140              | ectopic cilia  | 1                  | 0.1% | 0                |      |
| 25.110              | distichiasis   | 5                  | 0.4% | 0                |      |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |      |
| 40.910              | keratoconjunctivitis sicca                                     | 1                  | 0.1% | 0                |      |
| <b>NICTITANS</b>    |  |                    |      |                  |      |
| 51.100              | third eyelid cartilage anomaly                                 | 3                  | 0.2% | 3                | 1.1% |
| <b>CORNEA</b>       |  |                    |      |                  |      |
| 70.700              | corneal dystrophy  | 2                  | 0.1% | 1                | 0.4% |
| 70.730              | corneal endothelial degeneration                               | 1                  | 0.1% | 0                |      |
| <b>UVEA</b>         |  |                    |      |                  |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 86                 | 6.2% | 9                | 3.4% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 2                  | 0.1% | 0                |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 1                  | 0.1% | 1                | 0.4% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 1                  | 0.1% | 0                |      |
| 93.810              | uveal melanoma   | 1                  | 0.1% | 0                |      |
| <b>LENS</b>         |  |                    |      |                  |      |
| 100.200             | cataract, unspecified  | 2                  | 0.1% | 0                |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 75                 | 5.4% | 17               | 6.5% |
| 100.301             | punctate cataract, anterior cortex                             | 6                  | 0.4% | 1                | 0.4% |
| 100.302             | punctate cataract, posterior cortex                            | 1                  | 0.1% | 1                | 0.4% |
| 100.303             | punctate cataract, equatorial cortex                           | 0                  |      | 1                | 0.4% |
| 100.305             | punctate cataract, posterior sutures                           | 2                  | 0.1% | 0                |      |
| 100.306             | punctate cataract, nucleus                                     | 6                  | 0.4% | 1                | 0.4% |
| 100.307             | punctate cataract, capsular                                    | 0                  |      | 1                | 0.4% |
| 100.311             | incipient cataract, anterior cortex                            | 8                  | 0.6% | 2                | 0.8% |
| 100.312             | incipient cataract, posterior cortex                           | 16                 | 1.1% | 2                | 0.8% |
| 100.313             | incipient cataract, equatorial cortex                          | 7                  | 0.5% | 1                | 0.4% |
| 100.314             | incipient cataract, anterior sutures                           | 1                  | 0.1% | 0                |      |
| 100.315             | incipient cataract, posterior sutures                          | 9                  | 0.6% | 0                |      |
| 100.316             | incipient cataract, nucleus                                    | 7                  | 0.5% | 1                | 0.4% |
| 100.317             | incipient cataract, capsular                                   | 5                  | 0.4% | 0                |      |
| 100.321             | incomplete cataract, anterior cortex                           | 1                  | 0.1% | 0                |      |
| 100.322             | incomplete cataract, posterior cortex                          | 2                  | 0.1% | 1                | 0.4% |
| 100.328             | y-suture tip opacities   | 1                  | 0.1% | 1                | 0.4% |
| 100.330             | generalized/complete cataract                                  | 9                  | 0.6% | 1                | 0.4% |
| 100.345             | <i>significant cataracts (summary)</i>                         | 83                 | 5.9% | 14               | 5.3% |
| 100.375             | <i>subluxation/luxation, unspecified</i>                       | 1                  | 0.1% | 0                |      |
| <b>VITREOUS</b>     |  |                    |      |                  |      |
| 110.120             | persistent hyaloid artery/remnant                              | 2                  | 0.1% | 0                |      |
| 110.320             | vitreal degeneration   | 16                 | 1.1% | 3                | 1.1% |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>FUNDUS</b>   |             |           |
| 97.110 choroidal hypoplasia                               | 0           | 1 0.4%    |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 63 4.5%     | 2 0.8%    |
| 120.180 retinal dysplasia, geographic                     | 1 0.1%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 1 0.1%      | 0         |
| 120.920 retinal detachment with dialysis                  | 0           | 1 0.4%    |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 19 1.4%     | 0         |
| 900.100 other, not inherited                              | 41 2.9%     | 0         |
| 900.110 other. suspect not inherited/significance unknown | 11 0.8%     | 9 3.4%    |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,177 84.3% | 210 80.2% |

# CHOW CHOW

|    | DISORDER                         | INHERITANCE         | REFERENCE | BREEDING ADVICE         |
|----|----------------------------------|---------------------|-----------|-------------------------|
| A. | Glaucoma                         | Autosomal recessive | 1-3       | NO                      |
| B. | Entropion                        | Not defined         | 1         | NO                      |
| C. | Persistent pupillary membranes   |                     |           |                         |
|    | - iris to iris                   | Not defined         | 1         | Breeder option          |
|    | - iris to cornea                 | Not defined         | 1         | NO                      |
|    | - lens pigment foci/no strands   | Not defined         | 1         | Passes with no notation |
|    | - endothelial opacity/no strands | Not defined         |           | NO                      |
| D. | Cataract                         | Not defined         | 1, 4      | NO                      |

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## DESCRIPTION AND COMMENTS

### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of the IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine screening exam for certification.

Age of onset in the Chow Chow appears to be anywhere between 3-6 years of age and has been observed as a bilateral condition. Gonioscopy has shown extremely narrow iridocorneal angles and in many regions no evidence of trabecular meshwork.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

Entropion in the Chow Chow has been observed for decades and is definitely related to the amount of skin covering the head and face. Because of the conformation admired by both fanciers and the judges, it is doubtful that we will see a significant change in the incidence of

entropion as folds are, in many cases, desired by these individuals. Entropion requires surgical correction in the Chow Chow to return comfort and decrease chances for vision loss.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Major PPM's have been observed in the Chow Chow. Many ophthalmologists have observed puppies so severely affected that they are temporarily or permanently blind. The blindness is due to adherence of the membranes to the cornea and/or lens.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Chow Chow, the only reported cataract is congenital. The clinical appearance is variable, ranging from small nuclear or capsular opacities to generalized opacity. The central lens (nucleus) is most consistently affected with variable involvement of the peripheral lens (cortex). Concurrent ocular anomalies may include entropion, microphthalmia, persistent pupillary membranes, and retinal folds, although any direct relationship of these latter conditions to the cataract is unclear.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gelatt KN, MacKay EO. Prevalence of the breed-related glaucomas in pure-bred dogs in North America. *Vet Ophthalmol.* 2004;7:97-111.
3. Corcaran KA, Koch SA. Primary glaucoma in the Chow chows. *Prog Vet Comp Ophthalmol.* 1994;4:193-197.
4. Collins BK, Collier LL, Johnson GS, et al. Familial cataracts and concurrent ocular anomalies in chow chows. *J Am Vet Med Assoc.* 1992;200:1485-1491.



# OCULAR DISORDERS REPORT CHOW CHOW

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 0.110 microphthalmia  |                     | 4         | 0.3%  | 0         |       |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 20.160 macropalpebral fissure   |                     | 3         | 0.2%  | 0         |       |
| 21.000 entropion, unspecified   |                     | 365       | 27.5% | 35        | 17.9% |
| 22.000 ectropion, unspecified   |                     | 22        | 1.7%  | 4         | 2.0%  |
| 25.110 distichiasis   |                     | 8         | 0.6%  | 2         | 1.0%  |
| <b>NASOLACRIMAL</b>   |                     |           |       |           |       |
| 40.910 keratoconjunctivitis sicca                                     |                     | 2         | 0.2%  | 0         |       |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.210 corneal pannus   |                     | 9         | 0.7%  | 0         |       |
| 70.220 pigmentary keratitis   |                     | 25        | 1.9%  | 2         | 1.0%  |
| 70.700 corneal dystrophy  |                     | 8         | 0.6%  | 1         | 0.5%  |
| 70.730 corneal endothelial degeneration                               |                     | 17        | 1.3%  | 0         |       |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.140 corneal endothelial pigment without PPM                        |                     | 5         | 0.4%  | 0         |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 458       | 34.5% | 62        | 31.6% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 17        | 1.3%  | 1         | 0.5%  |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 54        | 4.1%  | 8         | 4.1%  |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 8         | 0.6%  | 1         | 0.5%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 12        | 0.9%  | 7         | 3.6%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 2         | 0.2%  | 7         | 3.6%  |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 29        | 2.2%  | 3         | 1.5%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 2         | 0.2%  | 0         |       |
| 100.302 punctate cataract, posterior cortex                           |                     | 5         | 0.4%  | 0         |       |
| 100.303 punctate cataract, equatorial cortex                          |                     | 2         | 0.2%  | 0         |       |
| 100.305 punctate cataract, posterior sutures                          |                     | 1         | 0.1%  | 0         |       |
| 100.306 punctate cataract, nucleus                                    |                     | 2         | 0.2%  | 0         |       |
| 100.307 punctate cataract, capsular                                   |                     | 1         | 0.1%  | 0         |       |
| 100.311 incipient cataract, anterior cortex                           |                     | 5         | 0.4%  | 0         |       |
| 100.312 incipient cataract, posterior cortex                          |                     | 9         | 0.7%  | 1         | 0.5%  |
| 100.315 incipient cataract, posterior sutures                         |                     | 1         | 0.1%  | 0         |       |
| 100.316 incipient cataract, nucleus                                   |                     | 3         | 0.2%  | 0         |       |
| 100.326 incomplete cataract, nucleus                                  |                     | 0         |       | 1         | 0.5%  |
| 100.328 y-suture tip opacities  |                     | 1         | 0.1%  | 0         |       |
| 100.330 generalized/complete cataract                                 |                     | 1         | 0.1%  | 0         |       |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 33        | 2.5%  | 2         | 1.0%  |
| <b>VITREOUS</b>   |                     |           |       |           |       |
| 110.120 persistent hyaloid artery/remnant                             |                     | 5         | 0.4%  | 0         |       |
| 110.320 vitreal degeneration  |                     | 2         | 0.2%  | 1         | 0.5%  |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 2 0.2%    | 0         |
| 120.180 retinal dysplasia, geographic                     | 1 0.1%    | 0         |
| 120.190 retinal dysplasia, detached                       | 1 0.1%    | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 8 0.6%    | 0         |
| <b>OPTIC NERVE</b>  |           |           |
| 130.120 optic nerve hypoplasia                            | 1 0.1%    | 0         |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 17 1.3%   | 0         |
| 900.100 other, not inherited                              | 22 1.7%   | 0         |
| 900.110 other. suspect not inherited/significance unknown | 21 1.6%   | 7 3.6%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 597 45.0% | 86 43.9%  |

# CLUMBER SPANIEL

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Entropion  | Not defined | 1, 2      | Breeder option  |
| B. | Ectropion  | Not defined | 1         | Breeder option  |
| C. | Distichiasis                                     | Not defined | 1         | Breeder option  |
| D. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| E. | Cataract   | Not defined | 1         | NO              |
| F. | Retinal dysplasia<br>- folds                     | Not defined | 1         | Breeder option  |

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## Description and Comments

### A. Entropion

A conformational defect resulting in "in-rolling" of one or both of the eyelids, which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Ectropion

A conformational defect resulting in eversion of the eyelids, which may cause ocular irritation. It is likely that ectropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### C. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

F. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Hodgman SFJ. Abnormalities and defects in pedigree dogs: I. An investigation into the existence of abnormalities in pedigree dogs in British Isles. *J Small Anim Pract.* 1963;4:447-456.

# OCULAR DISORDERS REPORT CLUMBER SPANIEL

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>2,686 |       | 2016-2020<br>266 |       |
|---|---------------------|--------------------|-------|------------------|-------|
|   |                     | #                  | %     | #                | %     |
| <b>GLOBE</b>  |                     |                    |       |                  |       |
| 0.110 microphthalmia  |                     | 6                  | 0.2%  | 0                |       |
| <b>EYELIDS</b>  |                     |                    |       |                  |       |
| 20.140 ectopic cilia  |                     | 1                  | 0.0%  | 0                |       |
| 20.160 macropalpebral fissure   |                     | 167                | 6.2%  | 0                |       |
| 21.000 entropion, unspecified   |                     | 583                | 21.7% | 56               | 21.1% |
| 22.000 ectropion, unspecified   |                     | 435                | 16.2% | 30               | 11.3% |
| 25.110 distichiasis   |                     | 192                | 7.1%  | 29               | 10.9% |
| <b>NASOLACRIMAL</b>   |                     |                    |       |                  |       |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 5                  | 0.2%  | 0                |       |
| 40.910 keratoconjunctivitis sicca                                     |                     | 18                 | 0.7%  | 3                | 1.1%  |
| <b>NICTITANS</b>  |                     |                    |       |                  |       |
| 52.110 prolapsed gland of the third eyelid                            |                     | 1                  | 0.0%  | 0                |       |
| <b>CORNEA</b>   |                     |                    |       |                  |       |
| 70.210 corneal pannus   |                     | 13                 | 0.5%  | 0                |       |
| 70.220 pigmentary keratitis   |                     | 11                 | 0.4%  | 1                | 0.4%  |
| 70.700 corneal dystrophy  |                     | 5                  | 0.2%  | 0                |       |
| 70.730 corneal endothelial degeneration                               |                     | 0                  |       | 1                | 0.4%  |
| <b>UVEA</b>   |                     |                    |       |                  |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 62                 | 2.3%  | 7                | 2.6%  |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 2                  | 0.1%  | 0                |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 6                  | 0.2%  | 0                |       |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 1                  | 0.0%  | 0                |       |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 2                  | 0.1%  | 0                |       |
| <b>LENS</b>   |                     |                    |       |                  |       |
| 100.200 cataract, unspecified   |                     | 15                 | 0.6%  | 0                |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 86                 | 3.2%  | 14               | 5.3%  |
| 100.300 punctate cataract, unspecified                                |                     | 0                  |       | 1                | 0.4%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 20                 | 0.7%  | 3                | 1.1%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 28                 | 1.0%  | 1                | 0.4%  |
| 100.303 punctate cataract, equatorial cortex                          |                     | 5                  | 0.2%  | 0                |       |
| 100.304 punctate cataract, anterior sutures                           |                     | 1                  | 0.0%  | 0                |       |
| 100.305 punctate cataract, posterior sutures                          |                     | 15                 | 0.6%  | 3                | 1.1%  |
| 100.306 punctate cataract, nucleus                                    |                     | 5                  | 0.2%  | 2                | 0.8%  |
| 100.307 punctate cataract, capsular                                   |                     | 1                  | 0.0%  | 0                |       |
| 100.311 incipient cataract, anterior cortex                           |                     | 15                 | 0.6%  | 1                | 0.4%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 41                 | 1.5%  | 4                | 1.5%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 7                  | 0.3%  | 0                |       |
| 100.314 incipient cataract, anterior sutures                          |                     | 2                  | 0.1%  | 0                |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 15                 | 0.6%  | 1                | 0.4%  |
| 100.316 incipient cataract, nucleus                                   |                     | 7                  | 0.3%  | 0                |       |
| 100.317 incipient cataract, capsular                                  |                     | 5                  | 0.2%  | 0                |       |
| 100.322 incomplete cataract, posterior cortex                         |                     | 1                  | 0.0%  | 1                | 0.4%  |
| 100.323 incomplete cataract, equatorial cortex                        |                     | 0                  |       | 1                | 0.4%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.325 incomplete cataract, posterior sutures            | 0                | 2 0.8%           |
| 100.326 incomplete cataract, nucleus                      | 0                | 1 0.4%           |
| 100.328 y-suture tip opacities                            | 2 0.1%           | 3 1.1%           |
| 100.330 generalized/complete cataract                     | 5 0.2%           | 0                |
| 100.345 <i>significant cataracts (summary)</i>            | 190 7.1%         | 24 9.0%          |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 6 0.2%           | 0                |
| 110.135 PHPV/PTVL   | 3 0.1%           | 0                |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 2 0.1%           | 0                |
| 97.120 coloboma   | 3 0.1%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 181 6.7%         | 3 1.1%           |
| 120.180 retinal dysplasia, geographic                     | 8 0.3%           | 2 0.8%           |
| 120.190 retinal dysplasia, detached                       | 0                | 1 0.4%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 15 0.6%          | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| 120.960 retinopathy                                       | 1 0.0%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.150 optic disc coloboma                               | 2 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 25 0.9%          | 0                |
| 900.100 other, not inherited                              | 64 2.4%          | 2 0.8%           |
| 900.110 other. suspect not inherited/significance unknown | 29 1.1%          | 5 1.9%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,426 53.1%      | 128 48.1%        |

# COCKER SPANIEL

(\*American)

\*The official breed name is Cocker Spaniel. The designation "American" has been used to avoid confusion and emphasize the distinction from the English Cocker Spaniel breed.

|    | DISORDER                               | INHERITANCE                  | REFERENCE  | BREEDING ADVICE         | GENETIC TESTS AVAILABLE          |
|----|--|------------------------------|------------|-------------------------|----------------------------------|
| A. | Keratoconjunctivitis sicca             | Not defined                  | 1, 2       | NO                      |                                  |
| B. | Glaucoma                               | Not defined                  | 1, 3, 4    | NO                      |                                  |
| C. | Ectropion                              | Not defined                  | 1          | Breeder option          |                                  |
| D. | Distichiasis                           | Not defined                  | 1, 2, 5, 6 | Breeder option          |                                  |
| E. | Imperforate lacrimal punctum           | Not defined                  | 1          | Breeder option          |                                  |
| F. | Prolapsed gland of the third eyelid    | Not defined                  | 1, 7       | Breeder option          |                                  |
| G. | Corneal dystrophy - epithelial/stromal | Not defined                  | 1          | Breeder option          |                                  |
| H. | Secondary keratitis – chronic          | Not defined                  | 1          | Passes with no notation |                                  |
| I. | Cataract                               | Presumed autosomal recessive | 1, 2, 8-11 | NO                      |                                  |
| J. | Retinal atrophy ( <i>prcd</i> )        | Autosomal recessive          | 1, 12-14   | NO                      | Mutation in the <i>prcd</i> gene |
| K. | Retinal dysplasia - folds              | Not defined                  | 1, 15      | Breeder option          |                                  |

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## Description and Comments

### A. Keratoconjunctivitis sicca

An abnormality of the tear film, most commonly a deficiency of the aqueous portion, although the mucin and/or lipid layers may be affected; results in ocular irritation and/or vision impairment.

B. Glaucoma

An elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening exam.

C. Ectropion

A conformational defect resulting in eversion of the eyelids, which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

D. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

E. Imperforate lacrimal punctum

A developmental anomaly resulting in failure of opening of the lacrimal duct adjacent to the eye. The lower punctum is more frequently affected. This defect usually results in epiphora, an overflow of tears onto the face.

F. Prolapsed gland of the third eyelid

Protrusion of the tear gland associated with the third eyelid. The mode of inheritance of this disorder is unknown. The exposed gland may become irritated. Commonly referred to as "cherry eye."

G. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

H. Secondary keratitis - chronic

A specific designation does not exist on the CAER form for this condition. We ask examiners to mark other – unlisted conditions suspected as inherited. Then in the comments box please write secondary keratitis – chronic.

A condition characterized by variable degrees of superficial vascularization, fibrosis and/or pigmentation of the cornea. Often associated with entropion or a combination of entropion and ectropion.



I. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In this breed, the onset of cataract may occur at an early age (less than 2 years) with rapid progression to maturity and associated with significant lens-induced inflammation.

J. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Cocker Spaniel is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

K. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

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# OCULAR DISORDERS REPORT COCKER SPANIEL

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>56,961 |       | 2016-2020<br>6,373 |       |
|---------------------|--|---------------------|-------|--------------------|-------|
|                     |  | #                   | %     | #                  | %     |
| <b>GLOBE</b>        |  |                     |       |                    |       |
| 0.110               | microphthalmia   | 35                  | 0.1%  | 1                  | 0.0%  |
| 10.000              | glaucoma   | 34                  | 0.1%  | 6                  | 0.1%  |
| <b>EYELIDS</b>      |  |                     |       |                    |       |
| 20.110              | eyelid dermoid   | 2                   | 0.0%  | 0                  |       |
| 20.140              | ectopic cilia  | 55                  | 0.1%  | 2                  | 0.0%  |
| 20.160              | macropalpebral fissure   | 179                 | 0.3%  | 0                  |       |
| 21.000              | entropion, unspecified   | 158                 | 0.3%  | 7                  | 0.1%  |
| 22.000              | ectropion, unspecified   | 981                 | 1.7%  | 35                 | 0.5%  |
| 25.110              | distichiasis   | 28,697              | 50.4% | 3,104              | 48.7% |
| <b>NASOLACRIMAL</b> |  |                     |       |                    |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 473                 | 0.8%  | 241                | 3.8%  |
| 40.910              | keratoconjunctivitis sicca                                     | 341                 | 0.6%  | 93                 | 1.5%  |
| <b>NICTITANS</b>    |  |                     |       |                    |       |
| 50.210              | pannus of third eyelid   | 0                   |       | 1                  | 0.0%  |
| 51.100              | third eyelid cartilage anomaly                                 | 8                   | 0.0%  | 1                  | 0.0%  |
| 52.110              | prolapsed gland of the third eyelid                            | 218                 | 0.4%  | 21                 | 0.3%  |
| <b>CORNEA</b>       |  |                     |       |                    |       |
| 70.210              | corneal pannus   | 497                 | 0.9%  | 1                  | 0.0%  |
| 70.220              | pigmentary keratitis   | 487                 | 0.9%  | 123                | 1.9%  |
| 70.700              | corneal dystrophy  | 1,581               | 2.8%  | 137                | 2.1%  |
| 70.730              | corneal endothelial degeneration                               | 37                  | 0.1%  | 6                  | 0.1%  |
| <b>UVEA</b>         |  |                     |       |                    |       |
| 90.250              | pigmentary uveitis   | 1                   | 0.0%  | 0                  |       |
| 93.110              | iris hypoplasia  | 2                   | 0.0%  | 2                  | 0.0%  |
| 93.140              | corneal endothelial pigment without PPM                        | 2                   | 0.0%  | 0                  |       |
| 93.150              | iris coloboma  | 8                   | 0.0%  | 2                  | 0.0%  |
| 93.180              | liris sphincter dysplasia                                      | 1                   | 0.0%  | 0                  |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 160                 | 0.3%  | 29                 | 0.5%  |
| 93.720              | persistent pupillary membranes, iris to lens                   | 30                  | 0.1%  | 2                  | 0.0%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 35                  | 0.1%  | 1                  | 0.0%  |
| 93.740              | persistent pupillary membranes, iris sheets                    | 28                  | 0.0%  | 0                  |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 36                  | 0.1%  | 35                 | 0.5%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 4                   | 0.0%  | 3                  | 0.0%  |
| 93.810              | uveal melanoma   | 1                   | 0.0%  | 0                  |       |
| 93.999              | uveal cysts  | 20                  | 0.0%  | 4                  | 0.1%  |
| 97.150              | chorioretinal coloboma, congenital                             | 5                   | 0.0%  | 1                  | 0.0%  |
| <b>LENS</b>         |  |                     |       |                    |       |
| 100.200             | cataract, unspecified  | 1,023               | 1.8%  | 0                  |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 3,354               | 5.9%  | 418                | 6.6%  |
| 100.301             | punctate cataract, anterior cortex                             | 926                 | 1.6%  | 143                | 2.2%  |
| 100.302             | punctate cataract, posterior cortex                            | 526                 | 0.9%  | 66                 | 1.0%  |
| 100.303             | punctate cataract, equatorial cortex                           | 139                 | 0.2%  | 24                 | 0.4%  |
| 100.304             | punctate cataract, anterior sutures                            | 134                 | 0.2%  | 11                 | 0.2%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.305 punctate cataract, posterior sutures              | 188              | 0.3%  | 40               | 0.6%  |
| 100.306 punctate cataract, nucleus                        | 76               | 0.1%  | 7                | 0.1%  |
| 100.307 punctate cataract, capsular                       | 58               | 0.1%  | 24               | 0.4%  |
| 100.311 incipient cataract, anterior cortex               | 1,032            | 1.8%  | 130              | 2.0%  |
| 100.312 incipient cataract, posterior cortex              | 1,201            | 2.1%  | 130              | 2.0%  |
| 100.313 incipient cataract, equatorial cortex             | 310              | 0.5%  | 42               | 0.7%  |
| 100.314 incipient cataract, anterior sutures              | 105              | 0.2%  | 6                | 0.1%  |
| 100.315 incipient cataract, posterior sutures             | 182              | 0.3%  | 19               | 0.3%  |
| 100.316 incipient cataract, nucleus                       | 190              | 0.3%  | 18               | 0.3%  |
| 100.317 incipient cataract, capsular                      | 80               | 0.1%  | 23               | 0.4%  |
| 100.321 incomplete cataract, anterior cortex              | 45               | 0.1%  | 62               | 1.0%  |
| 100.322 incomplete cataract, posterior cortex             | 43               | 0.1%  | 71               | 1.1%  |
| 100.323 incomplete cataract, equatorial cortex            | 6                | 0.0%  | 17               | 0.3%  |
| 100.324 incomplete cataract, anterior sutures             | 1                | 0.0%  | 2                | 0.0%  |
| 100.325 incomplete cataract, posterior sutures            | 4                | 0.0%  | 2                | 0.0%  |
| 100.326 incomplete cataract, nucleus                      | 9                | 0.0%  | 21               | 0.3%  |
| 100.327 incomplete cataract, capsular                     | 1                | 0.0%  | 2                | 0.0%  |
| 100.328 y-suture tip opacities                            | 19               | 0.0%  | 30               | 0.5%  |
| 100.330 generalized/complete cataract                     | 1,015            | 1.8%  | 50               | 0.8%  |
| 100.340 resorbing/hypermature cataract                    | 16               | 0.0%  | 21               | 0.3%  |
| 100.345 <i>significant cataracts (summary)</i>            | 7,329            | 12.9% | 961              | 15.1% |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 70               | 0.1%  | 20               | 0.3%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 42               | 0.1%  | 9                | 0.1%  |
| 110.135 PHPV/PTVL   | 9                | 0.0%  | 1                | 0.0%  |
| 110.200 vitreous degeneration-anterior chamber            | 4                | 0.0%  | 5                | 0.1%  |
| 110.320 vitreal degeneration                              | 151              | 0.3%  | 16               | 0.3%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 33               | 0.1%  | 0                |       |
| 97.120 coloboma   | 14               | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 6,764            | 11.9% | 338              | 5.3%  |
| 120.180 retinal dysplasia, geographic                     | 166              | 0.3%  | 9                | 0.1%  |
| 120.190 retinal dysplasia, detached                       | 9                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 460              | 0.8%  | 14               | 0.2%  |
| 120.400 retinal hemorrhage                                | 7                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 14               | 0.0%  | 0                |       |
| 120.960 retinopathy                                       | 19               | 0.0%  | 23               | 0.4%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 4                | 0.0%  | 0                |       |
| 130.120 optic nerve hypoplasia                            | 10               | 0.0%  | 0                |       |
| 130.150 optic disc coloboma                               | 111              | 0.2%  | 3                | 0.0%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 451              | 0.8%  | 0                |       |
| 900.100 other, not inherited                              | 1,059            | 1.9%  | 17               | 0.3%  |
| 900.110 other. suspect not inherited/significance unknown | 872              | 1.5%  | 310              | 4.9%  |

|                                     | 1991-2015    | 2016-2020   |
|-------------------------------------|--------------|-------------|
| <b>NORMAL</b><br>0.000 normal globe | 23,461 41.2% | 2,211 34.7% |

# COLLIE

(Rough and Smooth varieties)

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE           |
|----|---|------------------------|-----------|--------------------|--------------------------------------|
| A. | Microphthalmia  | Not defined            | 1, 2      | NO                 |                                      |
| B. | Distichiasis  | Not defined            | 1         | Breeder option     |                                      |
| C. | Corneal dystrophy<br>- epithelial/stromal   | Not defined            | 1         | Breeder option     |                                      |
| D. | Persistent pupillary<br>membranes   |                        |           |                    |                                      |
|    | - iris to iris  | Not defined            | 1         | Breeder option     |                                      |
|    | - iris to lens  | Not defined            | 1         | NO                 |                                      |
| E. | Cataract  | Not defined            | 1         | NO                 |                                      |
| F. | Persistent hyaloid artery   | Not defined            | 1         | Breeder option     |                                      |
| G. | Retinal atrophy<br>- generalized  | Not defined            | 1         | NO                 |                                      |
| H. | Retinal atrophy-<br>Rod/cone dysplasia type<br>2- ( <i>rcd2</i> )   | Autosomal<br>recessive | 3-66      | NO                 | Mutation in the<br><i>RD3</i> gene   |
| I. | Retinal dysplasia<br>- folds  | Not defined            | 1         | Breeder option     |                                      |
| J. | Choroidal hypoplasia<br>(Collie Eye Anomaly)<br>- staphyloma/coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- optic nerve coloboma | Autosomal<br>recessive | 1, 7-31   | NO                 | Mutation in the<br><i>NHEJ1</i> gene |

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## Description and Comments

### A. Microphthalmia

Microphthalmia is a congenital defect characterized by a small eye often associated with defects of the cornea, iris (coloboma), anterior chamber, lens (cataract) and/or retina.

An association has been made between partial albinism, multiple ocular defects (especially

microphthalmia) and deafness in a number of canine breeds including the Collie. From these reports it appears that a predominantly white hair coat is associated with a higher incidence of ocular defects.

B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. In the Collie, because there is significant clinical disease associated with the abnormal hairs, breeding of affected animals should be discouraged.

C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

In the Collie, this is a particularly serious problem noted frequently on routine screening examination. The majority of persistent pupillary membranes identified on routine screening examinations include iris sheets, and bridging from the iris to cornea and the iris to lens. These may result in vision impairment.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

F. Persistent hyaloid artery (PHA)

Congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small patent vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

G. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. PRA is inherited as an autosomal recessive trait in most breeds. In the Collie, the

rod/cone degeneration occurs very rarely and in those cases has not been caused by any of the known genetic mutations.

H. Retinal atrophy - Rod-cone dysplasia type 2- (*rcd2*)

An inherited retinal disease characterized by abortive or abnormal development of rods and cones. The disease can be detected histologically by 6 weeks. Clinical night blindness is observed as early as 6 weeks with total blindness by 1 year of age. It may be diagnosed as early as 24 days with an ERG. Histologically the disease can be detected by 6 weeks. This form of retinal dysplasia is clinically similar to, but genetically distinct from that seen in the Irish Setter. This condition is caused by an insertion in *RD3*. A DNA test is available.

I. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

J. Choroidal hypoplasia (Collie Eye Anomaly)

- staphyloma/coloboma
- retinal detachment
- retinal hemorrhage
- optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

**Historical Note:**

Central progressive retinal atrophy was previously a condition listed for this breed. However as the condition is no longer identified in the breed, the condition has been removed. Central progressive retinal atrophy was a progressive retinal degeneration in which photoreceptor death occurred secondary to disease of the underlying pigment epithelium. Progression was slow and some animals never lost vision. CPRA occurred in England, but was uncommon elsewhere.



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# OCULAR DISORDERS REPORT COLLIE

| Diagnostic Name     | TOTAL DOGS EXAMINED  |  | 1991-2015<br>55,225 |       | 2016-2020<br>8,566 |       |
|---------------------|--|--|---------------------|-------|--------------------|-------|
|                     |  |  | #                   | %     | #                  | %     |
| <b>GLOBE</b>        |  |  |                     |       |                    |       |
| 0.110               | microphthalmia   |  | 835                 | 1.5%  | 310                | 3.6%  |
| 10.000              | glaucoma   |  | 7                   | 0.0%  | 0                  |       |
| <b>EYELIDS</b>      |  |  |                     |       |                    |       |
| 20.110              | eyelid dermoid   |  | 1                   | 0.0%  | 0                  |       |
| 20.140              | ectopic cilia  |  | 5                   | 0.0%  | 0                  |       |
| 20.160              | macropalpebral fissure   |  | 1                   | 0.0%  | 0                  |       |
| 21.000              | entropion, unspecified   |  | 55                  | 0.1%  | 3                  | 0.0%  |
| 22.000              | ectropion, unspecified   |  | 8                   | 0.0%  | 0                  |       |
| 25.110              | distichiasis   |  | 1,043               | 1.9%  | 128                | 1.5%  |
| <b>NASOLACRIMAL</b> |  |  |                     |       |                    |       |
| 32.110              | imperforate lower nasolacrimal punctum                         |  | 8                   | 0.0%  | 1                  | 0.0%  |
| 40.910              | keratoconjunctivitis sicca                                     |  | 5                   | 0.0%  | 0                  |       |
| <b>NICTITANS</b>    |  |  |                     |       |                    |       |
| 51.100              | third eyelid cartilage anomaly                                 |  | 8                   | 0.0%  | 5                  | 0.1%  |
| 52.110              | prolapsed gland of the third eyelid                            |  | 2                   | 0.0%  | 0                  |       |
| <b>CORNEA</b>       |  |  |                     |       |                    |       |
| 70.210              | corneal pannus   |  | 2                   | 0.0%  | 1                  | 0.0%  |
| 70.220              | pigmentary keratitis   |  | 7                   | 0.0%  | 1                  | 0.0%  |
| 70.700              | corneal dystrophy  |  | 386                 | 0.7%  | 35                 | 0.4%  |
| 70.730              | corneal endothelial degeneration                               |  | 12                  | 0.0%  | 0                  |       |
| <b>UVEA</b>         |  |  |                     |       |                    |       |
| 90.250              | pigmentary uveitis   |  | 1                   | 0.0%  | 0                  |       |
| 93.110              | iris hypoplasia  |  | 3                   | 0.0%  | 5                  | 0.1%  |
| 93.140              | corneal endothelial pigment without PPM                        |  | 1                   | 0.0%  | 0                  |       |
| 93.150              | iris coloboma  |  | 23                  | 0.0%  | 1                  | 0.0%  |
| 93.180              | iris sphincter dysplasia                                       |  | 1                   | 0.0%  | 2                  | 0.0%  |
| 93.710              | persistent pupillary membranes, iris to iris                   |  | 8,796               | 15.9% | 2,233              | 26.1% |
| 93.720              | persistent pupillary membranes, iris to lens                   |  | 432                 | 0.8%  | 133                | 1.6%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 |  | 123                 | 0.2%  | 14                 | 0.2%  |
| 93.740              | persistent pupillary membranes, iris sheets                    |  | 65                  | 0.1%  | 4                  | 0.0%  |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   |  | 29                  | 0.1%  | 21                 | 0.2%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands |  | 12                  | 0.0%  | 2                  | 0.0%  |
| 93.810              | uveal melanoma   |  | 2                   | 0.0%  | 3                  | 0.0%  |
| 93.999              | uveal cysts  |  | 21                  | 0.0%  | 10                 | 0.1%  |
| 97.150              | chorioretinal coloboma, congenital                             |  | 159                 | 0.3%  | 290                | 3.4%  |
| <b>LENS</b>         |  |  |                     |       |                    |       |
| 100.200             | cataract, unspecified  |  | 114                 | 0.2%  | 0                  |       |
| 100.210             | cataract. suspect not inherited/significance unknown           |  | 525                 | 1.0%  | 100                | 1.2%  |
| 100.301             | punctate cataract, anterior cortex                             |  | 78                  | 0.1%  | 10                 | 0.1%  |
| 100.302             | punctate cataract, posterior cortex                            |  | 22                  | 0.0%  | 3                  | 0.0%  |
| 100.303             | punctate cataract, equatorial cortex                           |  | 5                   | 0.0%  | 0                  |       |
| 100.304             | punctate cataract, anterior sutures                            |  | 25                  | 0.0%  | 4                  | 0.0%  |
| 100.305             | punctate cataract, posterior sutures                           |  | 20                  | 0.0%  | 5                  | 0.1%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.306 punctate cataract, nucleus                        | 129              | 0.2%  | 27               | 0.3%  |
| 100.307 punctate cataract, capsular                       | 29               | 0.1%  | 8                | 0.1%  |
| 100.311 incipient cataract, anterior cortex               | 90               | 0.2%  | 13               | 0.2%  |
| 100.312 incipient cataract, posterior cortex              | 107              | 0.2%  | 4                | 0.0%  |
| 100.313 incipient cataract, equatorial cortex             | 35               | 0.1%  | 7                | 0.1%  |
| 100.314 incipient cataract, anterior sutures              | 34               | 0.1%  | 7                | 0.1%  |
| 100.315 incipient cataract, posterior sutures             | 23               | 0.0%  | 3                | 0.0%  |
| 100.316 incipient cataract, nucleus                       | 134              | 0.2%  | 27               | 0.3%  |
| 100.317 incipient cataract, capsular                      | 27               | 0.0%  | 5                | 0.1%  |
| 100.321 incomplete cataract, anterior cortex              | 1                | 0.0%  | 1                | 0.0%  |
| 100.322 incomplete cataract, posterior cortex             | 1                | 0.0%  | 2                | 0.0%  |
| 100.325 incomplete cataract, posterior sutures            | 0                |       | 1                | 0.0%  |
| 100.326 incomplete cataract, nucleus                      | 2                | 0.0%  | 4                | 0.0%  |
| 100.327 incomplete cataract, capsular                     | 0                |       | 1                | 0.0%  |
| 100.328 y-suture tip opacities                            | 4                | 0.0%  | 2                | 0.0%  |
| 100.330 generalized/complete cataract                     | 49               | 0.1%  | 0                |       |
| 100.345 <i>significant cataracts (summary)</i>            | 929              | 1.7%  | 134              | 1.6%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 8                | 0.0%  | 1                | 0.0%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 372              | 0.7%  | 35               | 0.4%  |
| 110.135 PHPV/PTVL   | 48               | 0.1%  | 4                | 0.0%  |
| 110.320 vitreal degeneration                              | 46               | 0.1%  | 2                | 0.0%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 37,676           | 68.2% | 6,424            | 75.0% |
| 97.120 coloboma   | 2,298            | 4.2%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 3,704            | 6.7%  | 699              | 8.2%  |
| 120.180 retinal dysplasia, geographic                     | 55               | 0.1%  | 6                | 0.1%  |
| 120.190 retinal dysplasia, detached                       | 86               | 0.2%  | 29               | 0.3%  |
| 120.310 generalized progressive retinal atrophy (PRA)     | 813              | 1.5%  | 2                | 0.0%  |
| 120.400 retinal hemorrhage                                | 105              | 0.2%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 823              | 1.5%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 59               | 0.1%  | 111              | 1.3%  |
| 120.960 retinopathy                                       | 1                | 0.0%  | 1                | 0.0%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 132              | 0.2%  | 47               | 0.5%  |
| 130.120 optic nerve hypoplasia                            | 231              | 0.4%  | 36               | 0.4%  |
| 130.150 optic disc coloboma                               | 4,319            | 7.8%  | 789              | 9.2%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 132              | 0.2%  | 0                |       |
| 900.100 other, not inherited                              | 286              | 0.5%  | 17               | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown | 608              | 1.1%  | 36               | 0.4%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 14,182           | 25.7% | 1,378            | 16.1% |

## COTON DE TULEAR

|    | DISORDER                                      | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|---|---------------------|-----------|-----------------|-----------------------------------|
| A. | Prolapsed gland of third eyelid               | Not defined         | 1         | Breeder option  |                                   |
| B. | Corneal dystrophy - epithelial/stromal        | Not defined         | 1         | Breeder option  |                                   |
| C. | Persistent pupillary membranes - iris to iris | Not defined         | 1         | Breeder option  |                                   |
| D. | Cataract                                      | Not defined         | 1         | NO              |                                   |
| E. | Y-suture tip opacity                          | Not defined         | 1         | Breeder option  |                                   |
| F. | Vitreous degeneration                         | Not defined         | 1         | Breeder option  |                                   |
| G. | Retinal atrophy ( <i>prcd</i> )               | Not defined         | 1         | NO              | Mutation in the <i>prcd</i> gene  |
| H. | Multifocal retinopathy - <i>cmr2</i>          | Autosomal recessive | 2, 3      | Breeder Option  | Mutation in the <i>BEST1</i> gene |

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### Description and Comments

A. Prolapse of the gland of the third eyelid

Protrusion of the tear gland associated with the third eyelid. The mode of inheritance of this disorder is unknown. The exposed gland may become irritated and severe chronic inflammation or keratoconjunctivitis sicca/dry eye syndrome may ensue. Commonly referred to as "cherry eye."

B. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to

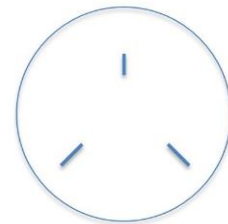
lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

F. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

G. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as Progressive Retinal Atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Coton de Tulear is *prcd* which is a

late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

#### H. Multifocal retinopathy – *cmr2*

Canine Multifocal Retinopathy type 2 (*cmr2*) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There is typically a serous sub-retinal fluid in the Coton de Tulear, although there may be accumulation of sub-retinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 15 weeks to 1 year of age. The lesions typically remain static in size and color beyond 1 year of age. The bullae appear to gradually lose the serous sub-retinal fluid after 4-5 years of age. Discrete areas of tapetal hyper-reflectivity might also be seen. Most dogs exhibit no noticeable problem with vision despite their abnormal appearing retinas. Electroretinograms reveal significant differences in photopic flickers in affected dogs.

Canine Multifocal Retinopathy type 2 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Coton du Tulear. A DNA test is available.

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# OCULAR DISORDERS REPORT COTON DE TULEAR

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>5,075 |      | 2016-2020<br>736 |      |
|---|---------------------|--------------------|------|------------------|------|
|   |                     | #                  | %    | #                | %    |
| <b>GLOBE</b>  |                     |                    |      |                  |      |
| 0.110 microphthalmia  |                     | 1                  | 0.0% | 0                |      |
| <b>EYELIDS</b>  |                     |                    |      |                  |      |
| 20.140 ectopic cilia  |                     | 1                  | 0.0% | 0                |      |
| 21.000 entropion, unspecified   |                     | 4                  | 0.1% | 2                | 0.3% |
| 25.110 distichiasis   |                     | 43                 | 0.8% | 4                | 0.5% |
| <b>NASOLACRIMAL</b>   |                     |                    |      |                  |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 1                  | 0.0% | 10               | 1.4% |
| 40.910 keratoconjunctivitis sicca                                     |                     | 1                  | 0.0% | 1                | 0.1% |
| <b>NICTITANS</b>  |                     |                    |      |                  |      |
| 52.110 prolapsed gland of the third eyelid                            |                     | 13                 | 0.3% | 10               | 1.4% |
| <b>CORNEA</b>   |                     |                    |      |                  |      |
| 70.220 pigmentary keratitis   |                     | 1                  | 0.0% | 0                |      |
| 70.700 corneal dystrophy  |                     | 47                 | 0.9% | 12               | 1.6% |
| 70.730 corneal endothelial degeneration                               |                     | 1                  | 0.0% | 0                |      |
| <b>UVEA</b>   |                     |                    |      |                  |      |
| 93.110 iris hypoplasia  |                     | 2                  | 0.0% | 0                |      |
| 93.150 iris coloboma  |                     | 2                  | 0.0% | 0                |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 438                | 8.6% | 63               | 8.6% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 8                  | 0.2% | 1                | 0.1% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 6                  | 0.1% | 0                |      |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 1                  | 0.0% | 0                |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 2                  | 0.0% | 2                | 0.3% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 8                  | 0.2% | 0                |      |
| 93.999 uveal cysts  |                     | 4                  | 0.1% | 0                |      |
| 97.150 chorioretinal coloboma, congenital                             |                     | 1                  | 0.0% | 0                |      |
| <b>LENS</b>   |                     |                    |      |                  |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 173                | 3.4% | 35               | 4.8% |
| 100.301 punctate cataract, anterior cortex                            |                     | 8                  | 0.2% | 1                | 0.1% |
| 100.302 punctate cataract, posterior cortex                           |                     | 4                  | 0.1% | 0                |      |
| 100.303 punctate cataract, equatorial cortex                          |                     | 3                  | 0.1% | 1                | 0.1% |
| 100.305 punctate cataract, posterior sutures                          |                     | 11                 | 0.2% | 1                | 0.1% |
| 100.306 punctate cataract, nucleus                                    |                     | 2                  | 0.0% | 0                |      |
| 100.307 punctate cataract, capsular                                   |                     | 3                  | 0.1% | 5                | 0.7% |
| 100.311 incipient cataract, anterior cortex                           |                     | 13                 | 0.3% | 2                | 0.3% |
| 100.312 incipient cataract, posterior cortex                          |                     | 15                 | 0.3% | 2                | 0.3% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 10                 | 0.2% | 1                | 0.1% |
| 100.314 incipient cataract, anterior sutures                          |                     | 2                  | 0.0% | 1                | 0.1% |
| 100.315 incipient cataract, posterior sutures                         |                     | 2                  | 0.0% | 4                | 0.5% |
| 100.316 incipient cataract, nucleus                                   |                     | 4                  | 0.1% | 1                | 0.1% |
| 100.317 incipient cataract, capsular                                  |                     | 6                  | 0.1% | 0                |      |
| 100.321 incomplete cataract, anterior cortex                          |                     | 1                  | 0.0% | 0                |      |
| 100.322 incomplete cataract, posterior cortex                         |                     | 0                  |      | 1                | 0.1% |
| 100.328 y-suture tip opacities  |                     | 5                  | 0.1% | 9                | 1.2% |



| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.330 generalized/complete cataract                     | 7 0.1%           | 1 0.1%           |
| 100.340 resorbing/hypermature cataract                    | 0                | 1 0.1%           |
| 100.345 significant cataracts (summary)                   | 96 1.9%          | 31 4.2%          |
| 100.375 subluxation/luxation, unspecified                 | 1 0.0%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 6 0.1%           | 3 0.4%           |
| 110.135 PHPV/PTVL   | 1 0.0%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 2 0.0%           | 0                |
| 110.320 vitreal degeneration                              | 46 0.9%          | 9 1.2%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 1 0.0%           | 1 0.1%           |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 21 0.4%          | 0                |
| 120.180 retinal dysplasia, geographic                     | 11 0.2%          | 0                |
| 120.190 retinal dysplasia, detached                       | 3 0.1%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 31 0.6%          | 5 0.7%           |
| 120.370 multifocal retinopathy                            | 2 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| 120.960 retinopathy                                       | 1 0.0%           | 3 0.4%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 3 0.1%           | 0                |
| 130.120 optic nerve hypoplasia                            | 2 0.0%           | 0                |
| 130.150 optic disc coloboma                               | 1 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 44 0.9%          | 0                |
| 900.100 other, not inherited                              | 151 3.0%         | 2 0.3%           |
| 900.110 other. suspect not inherited/significance unknown | 51 1.0%          | 36 4.9%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 4,345 85.6%      | 558 75.8%        |

# CURLY-COATED RETRIEVER

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--------------------------------|-------------|-----------|-------------------------|
| A. | Distichiasis                   | Not defined | 1         | Breeder option          |
| B. | Persistent pupillary membranes |             |           |                         |
|    | - iris to iris                 | Not defined | 1         | Breeder option          |
|    | - lens pigment foci/no strands | Not defined | 1         | Passes with no notation |
| C. | Cataract                       | Not defined | 2         | NO                      |
| D. | Y-suture tip opacity           | Not defined | 1         | Breeder option          |
| E. | Retinal dysplasia - folds      | Not defined | 1         | Breeder option          |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membrane (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### C. Cataract

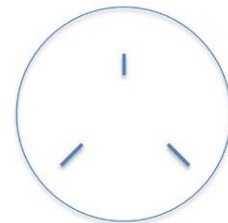
A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Curly-Coated Retriever the following cataracts have been reported:

1. **Anterior cortical subcapsular cataract:** Anterior subcapsular striate cortical cataracts usually occur bilaterally, slowly progress and usually occur between 5-8 years of age.
2. **Posterior subcapsular cataract:** Posterior polar subcapsular opacities occur at 2-4 years of age and progress slowly.

D. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

E. Retinal dysplasia – folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

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# OCULAR DISORDERS REPORT CURLY-COATED RETRIEVER

| Diagnostic Name   | TOTAL DOGS EXAMINED |  | 1991-2015<br>1,900 |      | 2016-2020<br>182 |       |
|---|---------------------|--|--------------------|------|------------------|-------|
|   |                     |  | #                  | %    | #                | %     |
| <b>GLOBE</b>  |                     |  |                    |      |                  |       |
| 0.110 microphthalmia  |                     |  | 1                  | 0.1% | 0                |       |
| <b>EYELIDS</b>  |                     |  |                    |      |                  |       |
| 20.140 ectopic cilia  |                     |  | 4                  | 0.2% | 0                |       |
| 21.000 entropion, unspecified   |                     |  | 11                 | 0.6% | 0                |       |
| 22.000 ectropion, unspecified   |                     |  | 3                  | 0.2% | 0                |       |
| 25.110 distichiasis   |                     |  | 149                | 7.8% | 13               | 7.1%  |
| <b>NICTITANS</b>  |                     |  |                    |      |                  |       |
| 51.100 third eyelid cartilage anomaly                                 |                     |  | 2                  | 0.1% | 1                | 0.5%  |
| 52.110 prolapsed gland of the third eyelid                            |                     |  | 1                  | 0.1% | 0                |       |
| <b>CORNEA</b>   |                     |  |                    |      |                  |       |
| 70.700 corneal dystrophy  |                     |  | 14                 | 0.7% | 0                |       |
| 70.730 corneal endothelial degeneration                               |                     |  | 1                  | 0.1% | 0                |       |
| <b>UVEA</b>   |                     |  |                    |      |                  |       |
| 90.250 pigmentary uveitis   |                     |  | 1                  | 0.1% | 0                |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     |  | 70                 | 3.7% | 11               | 6.0%  |
| 93.720 persistent pupillary membranes, iris to lens                   |                     |  | 4                  | 0.2% | 0                |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     |  | 5                  | 0.3% | 0                |       |
| 93.740 persistent pupillary membranes, iris sheets                    |                     |  | 2                  | 0.1% | 0                |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     |  | 12                 | 0.6% | 11               | 6.0%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     |  | 1                  | 0.1% | 0                |       |
| 93.999 uveal cysts  |                     |  | 1                  | 0.1% | 0                |       |
| <b>LENS</b>   |                     |  |                    |      |                  |       |
| 100.200 cataract, unspecified   |                     |  | 19                 | 1.0% | 0                |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     |  | 108                | 5.7% | 29               | 15.9% |
| 100.301 punctate cataract, anterior cortex                            |                     |  | 11                 | 0.6% | 2                | 1.1%  |
| 100.302 punctate cataract, posterior cortex                           |                     |  | 12                 | 0.6% | 1                | 0.5%  |
| 100.303 punctate cataract, equatorial cortex                          |                     |  | 2                  | 0.1% | 1                | 0.5%  |
| 100.304 punctate cataract, anterior sutures                           |                     |  | 1                  | 0.1% | 0                |       |
| 100.305 punctate cataract, posterior sutures                          |                     |  | 12                 | 0.6% | 8                | 4.4%  |
| 100.307 punctate cataract, capsular                                   |                     |  | 8                  | 0.4% | 0                |       |
| 100.311 incipient cataract, anterior cortex                           |                     |  | 11                 | 0.6% | 0                |       |
| 100.312 incipient cataract, posterior cortex                          |                     |  | 13                 | 0.7% | 0                |       |
| 100.313 incipient cataract, equatorial cortex                         |                     |  | 11                 | 0.6% | 0                |       |
| 100.314 incipient cataract, anterior sutures                          |                     |  | 1                  | 0.1% | 0                |       |
| 100.315 incipient cataract, posterior sutures                         |                     |  | 5                  | 0.3% | 1                | 0.5%  |
| 100.316 incipient cataract, nucleus                                   |                     |  | 3                  | 0.2% | 0                |       |
| 100.317 incipient cataract, capsular                                  |                     |  | 3                  | 0.2% | 0                |       |
| 100.328 y-suture tip opacities  |                     |  | 3                  | 0.2% | 8                | 4.4%  |
| 100.345 significant cataracts (summary)                               |                     |  | 115                | 6.1% | 21               | 11.5% |
| 100.375 subluxation/luxation, unspecified                             |                     |  | 3                  | 0.2% | 0                |       |
| <b>VITREOUS</b>   |                     |  |                    |      |                  |       |
| 110.120 persistent hyaloid artery/remnant                             |                     |  | 1                  | 0.1% | 2                | 1.1%  |
| 110.320 vitreal degeneration  |                     |  | 20                 | 1.1% | 0                |       |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>FUNDUS</b>   |             |           |
| 97.110 choroidal hypoplasia                               | 13 0.7%     | 0         |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 17 0.9%     | 5 2.7%    |
| 120.180 retinal dysplasia, geographic                     | 3 0.2%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 11 0.6%     | 1 0.5%    |
| 120.960 retinopathy                                       | 1 0.1%      | 0         |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 0           | 1 0.5%    |
| 130.120 optic nerve hypoplasia                            | 3 0.2%      | 0         |
| 130.150 optic disc coloboma                               | 13 0.7%     | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 16 0.8%     | 0         |
| 900.100 other, not inherited                              | 34 1.8%     | 1 0.5%    |
| 900.110 other. suspect not inherited/significance unknown | 27 1.4%     | 9 4.9%    |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,523 80.2% | 105 57.7% |

# DACHSHUND

|    | DISORDER   | INHERITANCE                | REFERENCE | BREEDING<br>ADVICE                        | GENETIC TESTS<br>AVAILABLE        |
|----|--|----------------------------|-----------|---|-----------------------------------|
| A. | Microphthalmia and multiple ocular defects   | Not defined                | 1-3       | NO  |                                   |
| B. | Distichiasis   | Not defined                | 1         | Breeder option                            |                                   |
| C. | Pigmentary keratitis   | Not defined                | 4         | NO  |                                   |
| D. | Corneal dystrophy - endothelial  | Not defined                | 1, 5, 6   | NO  |                                   |
| E. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined | 1<br>1    | Breeder option<br>Passes with no notation |                                   |
| F. | Cataract   | Not defined                | 1         | NO  |                                   |
| G. | Retinal atrophy ( <i>crd1</i> )  | Autosomal recessive        | 1, 7-18   | NO  | Mutation in the <i>NPHP4</i> gene |
| H. | Retinopathy - associated with ceroid lipofuscinosis                                | Autosomal recessive        | 2, 19-20  | NO  | Mutation in the <i>TPP1</i> gene  |

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## Description and Comments

### A. Microphthalmia and multiple ocular anomalies

Microphthalmia is a congenital defect characterized by a small eye often with associated defects of the cornea, anterior chamber, lens and/or retina. An association has been made between partial albinism, multiple ocular defects (especially microphthalmia) and deafness in a number of canine breeds including the Dachshund. From these reports it appears that a predominantly white hair coat is associated with a higher incidence of ocular defects.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

C. Punctate keratitis

Focal circular rings usually affecting the central sub-epithelial and/or anterior portion of the cornea. There often is an associated dry eye with corneal erosions. The mode of inheritance is unknown.

D. Corneal dystrophy - endothelial

An abnormal loss of the inner lining of the cornea that causes progressive fluid retention (edema). With time the edema results in keratitis and decreased vision.

E. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

F. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

G. Retinal atrophy – *crd1*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram before it is apparent clinically.

In Miniature Dachshunds there is a recessively inherited disorder caused by a 44 base pair insertion in the *RPGRIP1* gene. The insertion presumably truncates the protein and its major C-terminal RPGR binding domain. The resulting disease is called cone-rod dystrophy 1 (*crd1*) as the salient clinical abnormalities are a cone ERG dysfunction which does not correlate with photopic vision defects. The onset of the disease is variable, and is influenced by a second modifier locus which also is located on canine chromosome 15. Dogs homozygous for both defects have retinal abnormalities on ophthalmoscopy before 1-2 years of age. Dogs homozygous only for the *RPGRIP* insertion may have a late onset (>6 years) retinal degeneration diagnosed by ophthalmoscopy. Although the *RPGRIP1* molecular defect can be identified by means of a DNA test, questions have been raised about its validity given the poor genotype-phenotype correlation. A DNA test is available.

In a previous study using an inbred research colony, a 44-nucleotide insertion (*ins44*) in exon 2 of *RPGRIP1* was associated with retinal degeneration. Despite concordance of *ins44* with retinal degeneration, evidence indicate that there was phenotype-genotype discordance within the



miniature long-haired dachshunds that were not directly related to the experimental colony as not all dogs that were homozygous for ins44 were developing early onset retinal degeneration, but were developing retinal degeneration at a much later stage or not at all. In this investigation MAP9 deletion associated with early retinal degeneration onset was identified. Given the new genome assembly, the nominal title is CanFam3.1MAP9 corrected. Deletion was confirmed in early onset retinal degeneration cases and not late onset retinal degeneration cases, there is a variable age of onset and demonstrate the interaction of two independent loci that contribute to the phenotype. This study has shown that RPGRIP1 ins44/ins44 dogs with early onset retinal degeneration has several polymorphisms in MAP9, some of them potentially harmful, when compared with MAP9 in late onset retinal degeneration dogs. Detection of the presence or absence of MAP9 early onset retinal degeneration by qPCR can be used to specify early onset or late onset status for ins44 homozygotes. The story, however, is not as straightforward as suggested by the Forman et al. 2016 paper. Unpublished work by K. Miyadera and G. Aguirre in a research colony in which one of the founders originated from a MLHD at the Animal Health Trust finds that dogs that are homozygous for the RPGRIP1 ins 44 and the newly identified MAP9 deletion still do not show early-onset retinal degeneration. This suggests that there probably is a third genetic locus that interacts with MAP9 and RPGRIP1 in determining the age of disease onset and severity of the phenotype. Regardless, the identification of the MAP9 deletion is a major finding that will help unravel the complex genetics of this retinal disorder.

#### H. Retinopathy associated with ceroid lipofuscinosis

Progressive, multifocal serous retinal detachments first appear in Longhaired Dachshunds with late infantile neuronal ceroid lipofuscinosis at age 5-10 months. Late infantile ceroid neuronal lipofuscinosis in Miniature Dachshunds is a fatal, autosomal recessive, inherited lysosomal storage disease characterized by progressive neurodegeneration. The disease results from a defect in the *TPP1* (Tripeptidyl peptidase) gene. Inheritance of the retinopathy is linked to the gene causing late infantile neuronal ceroid lipofuscinosis.

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# OCULAR DISORDERS REPORT DACHSHUND

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>6,154 |      | 2016-2020<br>1,368 |      |
|---------------------|--|--------------------|------|--------------------|------|
|                     |  | #                  | %    | #                  | %    |
| <b>GLOBE</b>        |  |                    |      |                    |      |
| 0.110               | microphthalmia   | 22                 | 0.4% | 2                  | 0.1% |
| 10.000              | glaucoma   | 2                  | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                    |      |                    |      |
| 21.000              | entropion, unspecified   | 7                  | 0.1% | 1                  | 0.1% |
| 25.110              | distichiasis   | 374                | 6.1% | 118                | 8.6% |
| <b>NASOLACRIMAL</b> |  |                    |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 1                  | 0.0% | 1                  | 0.1% |
| 40.910              | keratoconjunctivitis sicca                                     | 2                  | 0.0% | 3                  | 0.2% |
| <b>NICTITANS</b>    |  |                    |      |                    |      |
| 50.210              | pannus of third eyelid   | 1                  | 0.0% | 0                  |      |
| 51.100              | third eyelid cartilage anomaly                                 | 2                  | 0.0% | 0                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 8                  | 0.1% | 2                  | 0.1% |
| <b>CORNEA</b>       |  |                    |      |                    |      |
| 70.210              | corneal pannus   | 3                  | 0.0% | 0                  |      |
| 70.700              | corneal dystrophy  | 33                 | 0.5% | 2                  | 0.1% |
| 70.730              | corneal endothelial degeneration                               | 9                  | 0.1% | 0                  |      |
| <b>UVEA</b>         |  |                    |      |                    |      |
| 93.110              | iris hypoplasia  | 7                  | 0.1% | 6                  | 0.4% |
| 93.150              | iris coloboma  | 25                 | 0.4% | 0                  |      |
| 93.180              | iris sphincter dysplasia                                       | 0                  |      | 2                  | 0.1% |
| 93.710              | persistent pupillary membranes, iris to iris                   | 250                | 4.1% | 72                 | 5.3% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 25                 | 0.4% | 5                  | 0.4% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 31                 | 0.5% | 3                  | 0.2% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 4                  | 0.1% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 75                 | 1.2% | 78                 | 5.7% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 11                 | 0.2% | 5                  | 0.4% |
| 93.999              | uveal cysts  | 4                  | 0.1% | 0                  |      |
| 97.150              | chorioretinal coloboma, congenital                             | 1                  | 0.0% | 2                  | 0.1% |
| <b>LENS</b>         |  |                    |      |                    |      |
| 100.200             | cataract, unspecified  | 43                 | 0.7% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 255                | 4.1% | 40                 | 2.9% |
| 100.301             | punctate cataract, anterior cortex                             | 29                 | 0.5% | 3                  | 0.2% |
| 100.302             | punctate cataract, posterior cortex                            | 15                 | 0.2% | 3                  | 0.2% |
| 100.303             | punctate cataract, equatorial cortex                           | 10                 | 0.2% | 1                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 4                  | 0.1% | 0                  |      |
| 100.305             | punctate cataract, posterior sutures                           | 9                  | 0.1% | 5                  | 0.4% |
| 100.306             | punctate cataract, nucleus                                     | 8                  | 0.1% | 2                  | 0.1% |
| 100.307             | punctate cataract, capsular                                    | 10                 | 0.2% | 5                  | 0.4% |
| 100.311             | incipient cataract, anterior cortex                            | 49                 | 0.8% | 2                  | 0.1% |
| 100.312             | incipient cataract, posterior cortex                           | 22                 | 0.4% | 2                  | 0.1% |
| 100.313             | incipient cataract, equatorial cortex                          | 14                 | 0.2% | 1                  | 0.1% |
| 100.314             | incipient cataract, anterior sutures                           | 2                  | 0.0% | 0                  |      |
| 100.315             | incipient cataract, posterior sutures                          | 18                 | 0.3% | 0                  |      |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.316 incipient cataract, nucleus                       | 9 0.1%           | 1 0.1%           |
| 100.317 incipient cataract, capsular                      | 7 0.1%           | 3 0.2%           |
| 100.321 incomplete cataract, anterior cortex              | 1 0.0%           | 2 0.1%           |
| 100.322 incomplete cataract, posterior cortex             | 0                | 1 0.1%           |
| 100.324 incomplete cataract, anterior sutures             | 1 0.0%           | 0                |
| 100.328 y-suture tip opacities                            | 0                | 6 0.4%           |
| 100.330 generalized/complete cataract                     | 39 0.6%          | 2 0.1%           |
| 100.340 resorbing/hypermature cataract                    | 2 0.0%           | 1 0.1%           |
| 100.345 <i>significant cataracts (summary)</i>            | 292 4.7%         | 40 2.9%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 7 0.1%           | 2 0.1%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 39 0.6%          | 4 0.3%           |
| 110.135 PHPV/PTVL   | 15 0.2%          | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 1 0.0%           | 1 0.1%           |
| 110.320 vitreal degeneration                              | 35 0.6%          | 5 0.4%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 5 0.1%           | 2 0.1%           |
| 97.120 coloboma   | 14 0.2%          | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 56 0.9%          | 16 1.2%          |
| 120.180 retinal dysplasia, geographic                     | 7 0.1%           | 0                |
| 120.190 retinal dysplasia, detached                       | 1 0.0%           | 1 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 119 1.9%         | 8 0.6%           |
| 120.400 retinal hemorrhage                                | 1 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 5 0.1%           | 0                |
| 120.920 retinal detachment with dialysis                  | 1 0.0%           | 2 0.1%           |
| 120.960 retinopathy                                       | 2 0.0%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 19 0.3%          | 5 0.4%           |
| 130.120 optic nerve hypoplasia                            | 40 0.6%          | 0                |
| 130.150 optic disc coloboma                               | 26 0.4%          | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 89 1.4%          | 0                |
| 900.100 other, not inherited                              | 198 3.2%         | 5 0.4%           |
| 900.110 other. suspect not inherited/significance unknown | 96 1.6%          | 72 5.3%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 4,807 78.1%      | 971 71.0%        |

## DALMATIAN

It is recommended that this breed be examined prior to pharmacological dilation to best facilitate identification of iris hypoplasia/sphincter dysplasia.

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--|-------------|-----------|-------------------------|
| A. | Glaucoma   | Not defined | 1-3       | NO                      |
| B. | Distichiasis                                       | Not defined | 1         | Breeder option          |
| C. | Corneal dystrophy<br>- epithelial/stromal          | Not defined | 1         | Breeder option          |
| D. | Iris hypoplasia                                    | Not defined | 1         | Breeder option          |
| E. | Iris sphincter dysplasia                           | Not defined | 1         | Passes with no notation |
| F. | Persistent pupillary<br>membrane<br>- iris to iris | Not defined | 1         | Breeder option          |
| G. | Cataract   | Not defined | 1, 2      | NO                      |
| H. | Vitreous degeneration                              | Not defined | 1         | Breeder option          |

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### Description and Comments

#### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure which, when sustained, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of the intraocular pressure (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening exam.

#### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

D. Iris Hypoplasia

A congenital abnormality in iris development usually characterized by a reduced quantity of tissue identified as partial-thickness defect in iris tissue. Full-thickness iris hypoplasia is rare and should be recorded as an iris coloboma on the OFA form.

E. Iris sphincter dysplasia

A congenital abnormality in iris development usually characterized by a full-thickness defect in iris tissue at the level of the iris sphincter, causing pupillary dilation. This abnormality has been noted in the Dalmatian breed.

F. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

G. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

H. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

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# OCULAR DISORDERS REPORT DALMATIAN

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>2,765 |      | 2016-2020<br>1,067 |      |
|---|---------------------|--------------------|------|--------------------|------|
|   |                     | #                  | %    | #                  | %    |
| <b>GLOBE</b>  |                     |                    |      |                    |      |
| 0.110 microphthalmia  |                     | 1                  | 0.0% | 0                  |      |
| <b>EYELIDS</b>  |                     |                    |      |                    |      |
| 20.140 ectopic cilia  |                     | 1                  | 0.0% | 0                  |      |
| 21.000 entropion, unspecified                                       |                     | 5                  | 0.2% | 0                  |      |
| 22.000 ectropion, unspecified                                       |                     | 1                  | 0.0% | 0                  |      |
| 25.110 distichiasis   |                     | 125                | 4.5% | 61                 | 5.7% |
| <b>NASOLACRIMAL</b>   |                     |                    |      |                    |      |
| 32.110 imperforate lower nasolacrimal punctum                       |                     | 1                  | 0.0% | 1                  | 0.1% |
| <b>NICTITANS</b>  |                     |                    |      |                    |      |
| 52.110 prolapsed gland of the third eyelid                          |                     | 1                  | 0.0% | 0                  |      |
| <b>CORNEA</b>   |                     |                    |      |                    |      |
| 70.210 corneal pannus   |                     | 1                  | 0.0% | 0                  |      |
| 70.700 corneal dystrophy  |                     | 77                 | 2.8% | 23                 | 2.2% |
| 70.730 corneal endothelial degeneration                             |                     | 2                  | 0.1% | 0                  |      |
| <b>UVEA</b>   |                     |                    |      |                    |      |
| 93.110 iris hypoplasia  |                     | 50                 | 1.8% | 10                 | 0.9% |
| 93.150 iris coloboma  |                     | 15                 | 0.5% | 1                  | 0.1% |
| 93.180 iris sphincter dysplasia                                     |                     | 14                 | 0.5% | 10                 | 0.9% |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 24                 | 0.9% | 9                  | 0.8% |
| 93.720 persistent pupillary membranes, iris to lens                 |                     | 3                  | 0.1% | 0                  |      |
| 93.730 persistent pupillary membranes, iris to cornea               |                     | 6                  | 0.2% | 0                  |      |
| 93.740 persistent pupillary membranes, iris sheets                  |                     | 1                  | 0.0% | 0                  |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 2                  | 0.1% | 1                  | 0.1% |
| 93.999 uveal cysts  |                     | 3                  | 0.1% | 1                  | 0.1% |
| 97.150 chorioretinal coloboma, congenital                           |                     | 1                  | 0.0% | 0                  |      |
| <b>LENS</b>   |                     |                    |      |                    |      |
| 100.110 microphakia, congenital                                     |                     | 0                  |      | 1                  | 0.1% |
| 100.200 cataract, unspecified                                       |                     | 1                  | 0.0% | 0                  |      |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 55                 | 2.0% | 22                 | 2.1% |
| 100.301 punctate cataract, anterior cortex                          |                     | 6                  | 0.2% | 4                  | 0.4% |
| 100.302 punctate cataract, posterior cortex                         |                     | 5                  | 0.2% | 1                  | 0.1% |
| 100.303 punctate cataract, equatorial cortex                        |                     | 7                  | 0.3% | 3                  | 0.3% |
| 100.305 punctate cataract, posterior sutures                        |                     | 0                  |      | 2                  | 0.2% |
| 100.306 punctate cataract, nucleus                                  |                     | 3                  | 0.1% | 0                  |      |
| 100.307 punctate cataract, capsular                                 |                     | 1                  | 0.0% | 1                  | 0.1% |
| 100.311 incipient cataract, anterior cortex                         |                     | 17                 | 0.6% | 7                  | 0.7% |
| 100.312 incipient cataract, posterior cortex                        |                     | 11                 | 0.4% | 1                  | 0.1% |
| 100.313 incipient cataract, equatorial cortex                       |                     | 12                 | 0.4% | 1                  | 0.1% |
| 100.314 incipient cataract, anterior sutures                        |                     | 3                  | 0.1% | 0                  |      |
| 100.315 incipient cataract, posterior sutures                       |                     | 1                  | 0.0% | 0                  |      |
| 100.316 incipient cataract, nucleus                                 |                     | 5                  | 0.2% | 1                  | 0.1% |
| 100.317 incipient cataract, capsular                                |                     | 2                  | 0.1% | 2                  | 0.2% |
| 100.321 incomplete cataract, anterior cortex                        |                     | 3                  | 0.1% | 2                  | 0.2% |
| 100.322 incomplete cataract, posterior cortex                       |                     | 2                  | 0.1% | 2                  | 0.2% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.323 incomplete cataract, equatorial cortex            | 1 0.0%           | 0                |
| 100.327 incomplete cataract, capsular                     | 1 0.0%           | 0                |
| 100.328 y-suture tip opacities                            | 0                | 1 0.1%           |
| 100.330 generalized/complete cataract                     | 6 0.2%           | 0                |
| 100.340 resorbing/hypermature cataract                    | 1 0.0%           | 0                |
| 100.345 <i>significant cataracts (summary)</i>            | 88 3.2%          | 28 2.6%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 4 0.1%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 0                | 4 0.4%           |
| 110.135 PHPV/PTVL   | 2 0.1%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 2 0.1%           | 1 0.1%           |
| 110.320 vitreal degeneration                              | 28 1.0%          | 2 0.2%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 12 0.4%          | 7 0.7%           |
| 120.180 retinal dysplasia, geographic                     | 0                | 4 0.4%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 5 0.2%           | 3 0.3%           |
| 120.400 retinal hemorrhage                                | 1 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| 120.960 retinopathy                                       | 1 0.0%           | 4 0.4%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 1 0.0%           | 1 0.1%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 43 1.6%          | 0                |
| 900.100 other, not inherited                              | 88 3.2%          | 3 0.3%           |
| 900.110 other. suspect not inherited/significance unknown | 112 4.1%         | 33 3.1%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 2,282 82.5%      | 881 82.6%        |



# DANDIE DINMONT TERRIER

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Glaucoma   | Not defined | 2, 3      | NO              |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| C. | Cataract   | Not defined | 1         | NO              |

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## Description and Comments

### A. Glaucoma

Glaucoma is an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening exam.

In the Dandie Dinmont terrier a 9.5 Mb susceptibility locus has been identified on canine chromosome 8. The definitive mutation has not been determined. A genetic test is not yet available.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

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# OCULAR DISORDERS REPORT DANDIE DINMONT TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>259 |       | 2016-2020<br>29 |       |
|---|---------------------|------------------|-------|-----------------|-------|
|   |                     | #                | %     | #               | %     |
| <b>GLOBE</b>  |                     |                  |       |                 |       |
| 0.110 microphthalmia  |                     | 1                | 0.4%  | 0               |       |
| 10.000 glaucoma   |                     | 1                | 0.4%  | 0               |       |
| <b>EYELIDS</b>  |                     |                  |       |                 |       |
| 25.110 distichiasis   |                     | 20               | 7.7%  | 1               | 3.4%  |
| <b>CORNEA</b>   |                     |                  |       |                 |       |
| 70.700 corneal dystrophy  |                     | 6                | 2.3%  | 1               | 3.4%  |
| <b>UVEA</b>   |                     |                  |       |                 |       |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 26               | 10.0% | 3               | 10.3% |
| 93.720 persistent pupillary membranes, iris to lens                 |                     | 1                | 0.4%  | 0               |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 2                | 0.8%  | 3               | 10.3% |
| 93.999 uveal cysts  |                     | 2                | 0.8%  | 0               |       |
| <b>LENS</b>   |                     |                  |       |                 |       |
| 100.200 cataract, unspecified                                       |                     | 4                | 1.5%  | 0               |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 28               | 10.8% | 1               | 3.4%  |
| 100.301 punctate cataract, anterior cortex                          |                     | 2                | 0.8%  | 1               | 3.4%  |
| 100.302 punctate cataract, posterior cortex                         |                     | 3                | 1.2%  | 0               |       |
| 100.305 punctate cataract, posterior sutures                        |                     | 1                | 0.4%  | 0               |       |
| 100.307 punctate cataract, capsular                                 |                     | 3                | 1.2%  | 0               |       |
| 100.311 incipient cataract, anterior cortex                         |                     | 2                | 0.8%  | 3               | 10.3% |
| 100.312 incipient cataract, posterior cortex                        |                     | 1                | 0.4%  | 0               |       |
| 100.330 generalized/complete cataract                               |                     | 5                | 1.9%  | 0               |       |
| 100.345 significant cataracts (summary)                             |                     | 21               | 8.1%  | 4               | 13.8% |
| 100.375 subluxation/luxation, unspecified                           |                     | 1                | 0.4%  | 0               |       |
| <b>VITREOUS</b>   |                     |                  |       |                 |       |
| 110.120 persistent hyaloid artery/remnant                           |                     | 3                | 1.2%  | 0               |       |
| <b>OTHER</b>  |                     |                  |       |                 |       |
| 900.000 other, unspecified  |                     | 6                | 2.3%  | 0               |       |
| 900.100 other, not inherited  |                     | 7                | 2.7%  | 0               |       |
| 900.110 other. suspect not inherited/significance unknown           |                     | 5                | 1.9%  | 2               | 6.9%  |
| <b>NORMAL</b>   |                     |                  |       |                 |       |
| 0.000 normal globe  |                     | 176              | 68.0% | 18              | 62.1% |

# DOBERMAN PINSCHER

|    | DISORDER  | INHERITANCE                                 | REFERENCE | BREEDING ADVICE                           |
|----|---|---|-----------|---|
| A. | Microphthalmia with multiple ocular defects   | Not defined                                 | 1-5       | NO  |
| B. | Distichiasis  | Not defined                                 | 1         | Breeder option                            |
| C. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands                        | Not defined<br>Not defined                  | 1-5<br>1  | Breeder option<br>Passes with no notation |
| D. | Cataract  | Not defined                                 | 1         | NO  |
| E. | Persistent hyperplastic primary vitreous/<br>Persistent hyperplastic tunica vasculosa lentis (PHPV/PHTVL) | Presumed dominant/<br>incomplete penetrance | 1, 6-14   | NO  |
| F. | Retinal dysplasia<br>- folds  | Not defined                                 | 1         | Breeder option                            |
| G. | Ligneous conjunctivitis   | Not defined                                 | 15        | NO  |

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## Description and Comments

### A. Microphthalmia with multiple ocular defects

Microphthalmia is a congenital defect characterized by a small eye often associated with defects of the cornea, iris (coloboma), anterior chamber, lens (cataract) and/or retina (retinal dysplasia). Note that this syndrome is distinct from "E," PHPV/PHTVL, which may also be associated with microphthalmia.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time. It is difficult to make a strong recommendation with regards to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded and breeding discretion is advised.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

Lens opacity which may affect one or both eyes and may involve the lens partially or completely. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membranes, persistent hyaloid, or nutritional deficiencies.

Cataracts have been infrequently observed in the Doberman Pinscher and there is no specific location attributed to cataracts within the Doberman lens. Most cataracts are bilateral, usually observed within the first two years of life, and may cause significant vision loss.

E. Persistent hyperplastic primary vitreous (PHPV)/Persistent hyperplastic tunica vasculosa lentis (PHTVL)

Persistent hyperplastic primary vitreous is a congenital defect resulting from abnormalities in the development and regression of the hyaloid artery (the primary vitreous) and the interaction of this blood vessel with the posterior lens capsule/cortex during embryogenesis. This condition is often associated with persistent hyperplastic tunica vasculosa lentis which results from failure of regression of the embryologic vascular network which surrounds the developing lens.

The condition in the Doberman includes a spectrum of malformations ranging from spots of pigment on the posterior surface of the lens to posterior lenticonus, cataract and a dense fibrous plaque on the posterior surface of the lens. In the more severe forms, partial or complete vision impairment occurs. PHPV has been extensively studied in the Doberman in Europe. This disorder has been observed occasionally in the Doberman in the United States.

F. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## G. Ligneous conjunctivitis

A rare type of conjunctivitis characterized by the formation of thick membranes covering conjunctiva of the nictitans and eyelids of affected dogs. This condition has been diagnosed in four unrelated Doberman Pinschers, three of which had life-threatening systemic disease. Ligneous conjunctivitis has also been reported in one Yorkshire Terrier.

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# OCULAR DISORDERS REPORT DOBERMAN PINSCHER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>5,267 |      | 2016-2020<br>1,222 |       |
|---------------------|--|--------------------|------|--------------------|-------|
|                     |  | #                  | %    | #                  | %     |
| <b>GLOBE</b>        |  |                    |      |                    |       |
| 0.110               | microphthalmia   | 7                  | 0.1% | 0                  |       |
| 10.000              | glaucoma   | 0                  |      | 1                  | 0.1%  |
| <b>EYELIDS</b>      |  |                    |      |                    |       |
| 20.140              | ectopic cilia  | 1                  | 0.0% | 0                  |       |
| 21.000              | entropion, unspecified   | 6                  | 0.1% | 1                  | 0.1%  |
| 22.000              | ectropion, unspecified   | 1                  | 0.0% | 0                  |       |
| 25.110              | distichiasis   | 87                 | 1.7% | 19                 | 1.6%  |
| <b>NASOLACRIMAL</b> |  |                    |      |                    |       |
| 40.910              | keratoconjunctivitis sicca                                     | 1                  | 0.0% | 1                  | 0.1%  |
| <b>NICTITANS</b>    |  |                    |      |                    |       |
| 51.100              | third eyelid cartilage anomaly                                 | 7                  | 0.1% | 2                  | 0.2%  |
| 52.110              | prolapsed gland of the third eyelid                            | 7                  | 0.1% | 0                  |       |
| <b>CORNEA</b>       |  |                    |      |                    |       |
| 70.700              | corneal dystrophy  | 10                 | 0.2% | 1                  | 0.1%  |
| 70.730              | corneal endothelial degeneration                               | 4                  | 0.1% | 0                  |       |
| <b>UVEA</b>         |  |                    |      |                    |       |
| 93.110              | iris hypoplasia  | 1                  | 0.0% | 0                  |       |
| 93.140              | corneal endothelial pigment without PPM                        | 2                  | 0.0% | 0                  |       |
| 93.150              | iris coloboma  | 1                  | 0.0% | 0                  |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 116                | 2.2% | 19                 | 1.6%  |
| 93.720              | persistent pupillary membranes, iris to lens                   | 33                 | 0.6% | 2                  | 0.2%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 8                  | 0.2% | 2                  | 0.2%  |
| 93.740              | persistent pupillary membranes, iris sheets                    | 4                  | 0.1% | 0                  |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 79                 | 1.5% | 137                | 11.2% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 3                  | 0.1% | 4                  | 0.3%  |
| 93.810              | uveal melanoma   | 3                  | 0.1% | 1                  | 0.1%  |
| 93.999              | uveal cysts  | 7                  | 0.1% | 7                  | 0.6%  |
| 97.150              | chorioretinal coloboma, congenital                             | 0                  |      | 1                  | 0.1%  |
| <b>LENS</b>         |  |                    |      |                    |       |
| 100.200             | cataract, unspecified  | 32                 | 0.6% | 0                  |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 288                | 5.5% | 56                 | 4.6%  |
| 100.301             | punctate cataract, anterior cortex                             | 15                 | 0.3% | 1                  | 0.1%  |
| 100.302             | punctate cataract, posterior cortex                            | 4                  | 0.1% | 3                  | 0.2%  |
| 100.303             | punctate cataract, equatorial cortex                           | 1                  | 0.0% | 0                  |       |
| 100.304             | punctate cataract, anterior sutures                            | 4                  | 0.1% | 0                  |       |
| 100.305             | punctate cataract, posterior sutures                           | 10                 | 0.2% | 1                  | 0.1%  |
| 100.306             | punctate cataract, nucleus                                     | 6                  | 0.1% | 4                  | 0.3%  |
| 100.307             | punctate cataract, capsular                                    | 16                 | 0.3% | 3                  | 0.2%  |
| 100.311             | incipient cataract, anterior cortex                            | 9                  | 0.2% | 3                  | 0.2%  |
| 100.312             | incipient cataract, posterior cortex                           | 18                 | 0.3% | 2                  | 0.2%  |
| 100.313             | incipient cataract, equatorial cortex                          | 7                  | 0.1% | 3                  | 0.2%  |
| 100.315             | incipient cataract, posterior sutures                          | 8                  | 0.2% | 0                  |       |
| 100.316             | incipient cataract, nucleus                                    | 16                 | 0.3% | 4                  | 0.3%  |



| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.317 incipient cataract, capsular                      | 10 0.2%          | 4 0.3%           |
| 100.321 incomplete cataract, anterior cortex              | 0                | 2 0.2%           |
| 100.322 incomplete cataract, posterior cortex             | 0                | 3 0.2%           |
| 100.323 incomplete cataract, equatorial cortex            | 0                | 1 0.1%           |
| 100.326 incomplete cataract, nucleus                      | 0                | 2 0.2%           |
| 100.328 y-suture tip opacities                            | 1 0.0%           | 1 0.1%           |
| 100.330 generalized/complete cataract                     | 14 0.3%          | 1 0.1%           |
| 100.345 <i>significant cataracts (summary)</i>            | 171 3.2%         | 38 3.1%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 2 0.0%           | 3 0.2%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 19 0.4%          | 8 0.7%           |
| 110.135 PHPV/PTVL   | 43 0.8%          | 9 0.7%           |
| 110.200 vitreous degeneration-anterior chamber            | 0                | 1 0.1%           |
| 110.320 vitreal degeneration                              | 10 0.2%          | 1 0.1%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 2 0.0%           | 0                |
| 97.120 coloboma   | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 97 1.8%          | 5 0.4%           |
| 120.180 retinal dysplasia, geographic                     | 12 0.2%          | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 12 0.2%          | 3 0.2%           |
| 120.910 retinal detachment without dialysis               | 2 0.0%           | 0                |
| 120.920 retinal detachment with dialysis                  | 0                | 2 0.2%           |
| 120.960 retinopathy                                       | 1 0.0%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.120 optic nerve hypoplasia                            | 3 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 57 1.1%          | 0                |
| 900.100 other, not inherited                              | 167 3.2%         | 7 0.6%           |
| 900.110 other. suspect not inherited/significance unknown | 86 1.6%          | 90 7.4%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 4,428 84.1%      | 878 71.8%        |
| <b>SCLERA</b>   |                  |                  |
| 80.810 limbal melanoma                                    | 0                | 1 0.1%           |

# DOGUE DE BORDEAUX

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|--|---------------------|-----------|-----------------|-----------------------------------|
| A. | Distichiasis                                     | Not defined         | 1         | Breeder option  |                                   |
| B. | Entropion  | Not defined         | 1         | Breeder option  |                                   |
| C. | Ectropion  | Not defined         | 1         | Breeder option  |                                   |
| D. | Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option  |                                   |
| E. | Cataract   | Not defined         | 1         | NO              |                                   |
| F. | Multifocal retinopathy<br>- <i>cmr1</i>          | Autosomal recessive | 2         | Breeder option  | Mutation in the <i>BEST1</i> gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. Entropion in the Mastiff is severe and may require multiple surgical corrections.

### C. Ectropion

A conformational defect resulting in eversion of the eyelids, which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

F. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (cmr1) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
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# OCULAR DISORDERS REPORT DOGUE DE BORDEAUX

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|------------------|--|-----------|-------|-----------|-------|
|                  |  | #         | %     | #         | %     |
| <b>EYELIDS</b>   |  |           |       |           |       |
| 20.160           | macropalpebral fissure   | 9         | 3.0%  | 0         |       |
| 21.000           | entropion, unspecified   | 16        | 5.3%  | 21        | 21.0% |
| 22.000           | ectropion, unspecified   | 33        | 10.9% | 13        | 13.0% |
| 25.110           | distichiasis   | 30        | 9.9%  | 7         | 7.0%  |
| <b>NICTITANS</b> |  |           |       |           |       |
| 52.110           | prolapsed gland of the third eyelid                            | 1         | 0.3%  | 0         |       |
| <b>CORNEA</b>    |  |           |       |           |       |
| 70.700           | corneal dystrophy  | 6         | 2.0%  | 6         | 6.0%  |
| 70.730           | corneal endothelial degeneration                               | 1         | 0.3%  | 0         |       |
| <b>UVEA</b>      |  |           |       |           |       |
| 93.710           | persistent pupillary membranes, iris to iris                   | 13        | 4.3%  | 4         | 4.0%  |
| 93.720           | persistent pupillary membranes, iris to lens                   | 1         | 0.3%  | 1         | 1.0%  |
| 93.730           | persistent pupillary membranes, iris to cornea                 | 4         | 1.3%  | 1         | 1.0%  |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands   | 5         | 1.7%  | 0         |       |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands | 1         | 0.3%  | 1         | 1.0%  |
| 93.999           | uveal cysts  | 3         | 1.0%  | 3         | 3.0%  |
| <b>LENS</b>      |  |           |       |           |       |
| 100.210          | cataract. suspect not inherited/significance unknown           | 9         | 3.0%  | 0         |       |
| 100.301          | punctate cataract, anterior cortex                             | 1         | 0.3%  | 1         | 1.0%  |
| 100.302          | punctate cataract, posterior cortex                            | 0         |       | 1         | 1.0%  |
| 100.306          | punctate cataract, nucleus                                     | 3         | 1.0%  | 0         |       |
| 100.311          | incipient cataract, anterior cortex                            | 1         | 0.3%  | 0         |       |
| 100.313          | incipient cataract, equatorial cortex                          | 0         |       | 2         | 2.0%  |
| 100.316          | incipient cataract, nucleus                                    | 1         | 0.3%  | 1         | 1.0%  |
| 100.321          | incomplete cataract, anterior cortex                           | 0         |       | 1         | 1.0%  |
| 100.345          | <i>significant cataracts (summary)</i>                         | 6         | 2.0%  | 6         | 6.0%  |
| <b>VITREOUS</b>  |  |           |       |           |       |
| 110.120          | persistent hyaloid artery/remnant                              | 1         | 0.3%  | 0         |       |
| <b>RETINA</b>    |  |           |       |           |       |
| 120.170          | retinal dysplasia, folds                                       | 6         | 2.0%  | 1         | 1.0%  |
| 120.960          | retinopathy  | 1         | 0.3%  | 0         |       |
| <b>OTHER</b>     |  |           |       |           |       |
| 900.000          | other, unspecified   | 6         | 2.0%  | 0         |       |
| 900.100          | other, not inherited   | 10        | 3.3%  | 0         |       |
| 900.110          | other. suspect not inherited/significance unknown              | 4         | 1.3%  | 6         | 6.0%  |
| <b>NORMAL</b>    |  |           |       |           |       |
| 0.000            | normal globe   | 206       | 68.2% | 54        | 54.0% |

# DUTCH SHEPHERD

|    | <b>DISORDER</b> | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|-----------------|--------------------|------------------|------------------------|
| A. | Cataract        | Not defined        | 1                | NO                     |

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## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Dutch Shepherd breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT DUTCH SHEPHERD

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 3         | 6.2%  | 0         |       |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.210          | corneal pannus   | 0         |       | 1         | 1.3%  |
| 70.700          | corneal dystrophy  | 1         | 2.1%  | 2         | 2.6%  |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 0         |       | 3         | 3.9%  |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 2         | 4.2%  | 0         |       |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 7         | 14.6% | 9         | 11.8% |
| 100.301         | punctate cataract, anterior cortex                           | 1         | 2.1%  | 1         | 1.3%  |
| 100.302         | punctate cataract, posterior cortex                          | 0         |       | 1         | 1.3%  |
| 100.303         | punctate cataract, equatorial cortex                         | 0         |       | 2         | 2.6%  |
| 100.304         | punctate cataract, anterior sutures                          | 1         | 2.1%  | 0         |       |
| 100.306         | punctate cataract, nucleus                                   | 0         |       | 2         | 2.6%  |
| 100.307         | punctate cataract, capsular                                  | 1         | 2.1%  | 1         | 1.3%  |
| 100.311         | incipient cataract, anterior cortex                          | 1         | 2.1%  | 0         |       |
| 100.312         | incipient cataract, posterior cortex                         | 1         | 2.1%  | 0         |       |
| 100.313         | incipient cataract, equatorial cortex                        | 1         | 2.1%  | 1         | 1.3%  |
| 100.345         | <i>significant cataracts (summary)</i>                       | 6         | 12.5% | 8         | 10.5% |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.310         | generalized progressive retinal atrophy (PRA)                | 1         | 2.1%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.000         | other, unspecified   | 3         | 6.2%  | 0         |       |
| 900.100         | other, not inherited   | 0         |       | 1         | 1.3%  |
| 900.110         | other. suspect not inherited/significance unknown            | 3         | 6.2%  | 3         | 3.9%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 37        | 77.1% | 56        | 73.7% |

# ENGLISH COCKER SPANIEL

|    | DISORDER                            | INHERITANCE                  | REFERENCE | BREEDING ADVICE         | GENETIC TESTS AVAILABLE          |
|----|-------------------------------------|------------------------------|-----------|-------------------------|----------------------------------|
| A. | Keratoconjunctivitis sicca          | Not defined                  | 1         | NO                      |                                  |
| B. | Glaucoma                            | Not defined                  | 1, 3-4    | NO                      |                                  |
| C. | Distichiasis                        | Not defined                  | 1, 4, 5   | Breeder option          |                                  |
| D. | Persistent pupillary membranes      |                              |           |                         |                                  |
|    | - iris to iris                      | Not defined                  | 1, 6      | Breeder option          |                                  |
|    | - iris to cornea                    | Not defined                  | 1, 6      | NO                      |                                  |
|    | - lens pigment foci/no strands      | Not defined                  | 1         | Passes with no notation |                                  |
| E. | Cataract                            | Not defined                  | 1, 6-9    | NO                      |                                  |
| F. | Retinal atrophy ( <i>prcd</i> )     | Autosomal recessive          | 1, 10-13  | NO                      | Mutation in the <i>prcd</i> gene |
| G. | Central progressive retinal atrophy | Not defined                  | 14-16     | NO                      |                                  |
| H. | Retinal dysplasia - folds           | Presumed autosomal recessive | 1         | Breeder option          |                                  |

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## Description and Comments

### A. Keratoconjunctivitis sicca (KCS)

An abnormality of the tear film, most commonly a deficiency of the aqueous portion, although the mucin and/or lipid layers may be affected; results in ocular irritation and/or vision impairment.

### B. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of the IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine screening exam for certification.

Glaucoma in the English Cocker Spaniel is recognized in England. The frequency and significance of this disease in the breed in the United States is not known, but is probably low.

C. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

In the English Cocker Spaniel, this is a particularly serious problem as the majority of PPMs identified on routine screening examination bridge from the iris to the cornea and are associated with corneal opacities which may result in vision impairment.

Lens pigment foci/no strands is considered an insignificant finding and therefore is not noted on the certificate.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

Congenital cataracts have been reported in Red Cocker Spaniels, presumably English Cocker Spaniels, in Denmark. The cataracts affected the anterior capsule; in some cases the cortex and/or nucleus were opaque. Associated findings in some dogs were persistent pupillary membrane (PPM) and/or microphthalmia. It is likely that these cataracts are part of a syndrome characterized by multiple congenital ocular anomalies. The condition is familial, but a specific mode of inheritance has not been defined.

F. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the English Cocker Spaniel is *prcd*



which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. However, in the English Cocker Spaniel, the phenotype can be very variable in the age of onset. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

Other forms of retinal degeneration that are not *prcd* are recognized in the breed. The currently available genetic test will not detect these other forms of PRA.

#### G. Central progressive retinal atrophy (CPRA)

A progressive retinal degeneration in which photoreceptor degeneration occurs secondary to disease of the underlying pigment epithelium. Progression is slow and some animals may never lose vision. CPRA is a frequent occurrence in England, but is uncommon elsewhere.

CPRA is characterized by the appearance of brown spots and patches primarily in the tapetal fundus and retinal degeneration. These areas are created by an accumulation of autofluorescent lipopigment within the retinal pigment epithelium cells. These changes are consistent with retinal changes observed in Vitamin E deficiency. Neurologic signs including ataxia and proprioceptive deficits have also been identified in affected dogs.

In the English Cocker Spaniel, retinal lesions of CPRA have been related to an underlying abnormal metabolism of Vitamin E resulting in a systemic deficiency.

#### H. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
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# OCULAR DISORDERS REPORT ENGLISH COCKER SPANIEL

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>10,945 |       | 2016-2020<br>1,065 |       |
|---------------------|--|---------------------|-------|--------------------|-------|
|                     |  | #                   | %     | #                  | %     |
| <b>GLOBE</b>        |  |                     |       |                    |       |
| 0.110               | microphthalmia   | 14                  | 0.1%  | 1                  | 0.1%  |
| 10.000              | glaucoma   | 1                   | 0.0%  | 0                  |       |
| <b>EYELIDS</b>      |  |                     |       |                    |       |
| 20.110              | eyelid dermoid   | 1                   | 0.0%  | 0                  |       |
| 20.140              | ectopic cilia  | 6                   | 0.1%  | 0                  |       |
| 20.160              | macropalpebral fissure   | 3                   | 0.0%  | 0                  |       |
| 21.000              | entropion, unspecified   | 46                  | 0.4%  | 5                  | 0.5%  |
| 22.000              | ectropion, unspecified   | 97                  | 0.9%  | 1                  | 0.1%  |
| 25.110              | distichiasis   | 1,966               | 18.0% | 157                | 14.7% |
| <b>NASOLACRIMAL</b> |  |                     |       |                    |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 18                  | 0.2%  | 8                  | 0.8%  |
| 40.910              | keratoconjunctivitis sicca                                     | 12                  | 0.1%  | 0                  |       |
| <b>NICTITANS</b>    |  |                     |       |                    |       |
| 52.110              | prolapsed gland of the third eyelid                            | 6                   | 0.1%  | 0                  |       |
| <b>CORNEA</b>       |  |                     |       |                    |       |
| 70.210              | corneal pannus   | 10                  | 0.1%  | 1                  | 0.1%  |
| 70.220              | pigmentary keratitis   | 11                  | 0.1%  | 0                  |       |
| 70.700              | corneal dystrophy  | 95                  | 0.9%  | 10                 | 0.9%  |
| 70.730              | corneal endothelial degeneration                               | 37                  | 0.3%  | 0                  |       |
| <b>UVEA</b>         |  |                     |       |                    |       |
| 90.250              | pigmentary uveitis   | 1                   | 0.0%  | 0                  |       |
| 93.140              | corneal endothelial pigment without PPM                        | 6                   | 0.1%  | 0                  |       |
| 93.150              | iris coloboma  | 2                   | 0.0%  | 0                  |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 135                 | 1.2%  | 25                 | 2.3%  |
| 93.720              | persistent pupillary membranes, iris to lens                   | 41                  | 0.4%  | 3                  | 0.3%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 184                 | 1.7%  | 7                  | 0.7%  |
| 93.740              | persistent pupillary membranes, iris sheets                    | 10                  | 0.1%  | 0                  |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 36                  | 0.3%  | 37                 | 3.5%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 17                  | 0.2%  | 10                 | 0.9%  |
| 93.999              | uveal cysts  | 5                   | 0.0%  | 0                  |       |
| <b>LENS</b>         |  |                     |       |                    |       |
| 100.200             | cataract, unspecified  | 172                 | 1.6%  | 0                  |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 674                 | 6.2%  | 58                 | 5.4%  |
| 100.301             | punctate cataract, anterior cortex                             | 97                  | 0.9%  | 8                  | 0.8%  |
| 100.302             | punctate cataract, posterior cortex                            | 50                  | 0.5%  | 2                  | 0.2%  |
| 100.303             | punctate cataract, equatorial cortex                           | 19                  | 0.2%  | 1                  | 0.1%  |
| 100.304             | punctate cataract, anterior sutures                            | 11                  | 0.1%  | 2                  | 0.2%  |
| 100.305             | punctate cataract, posterior sutures                           | 31                  | 0.3%  | 2                  | 0.2%  |
| 100.306             | punctate cataract, nucleus                                     | 23                  | 0.2%  | 3                  | 0.3%  |
| 100.307             | punctate cataract, capsular                                    | 8                   | 0.1%  | 5                  | 0.5%  |
| 100.311             | incipient cataract, anterior cortex                            | 130                 | 1.2%  | 4                  | 0.4%  |
| 100.312             | incipient cataract, posterior cortex                           | 133                 | 1.2%  | 3                  | 0.3%  |
| 100.313             | incipient cataract, equatorial cortex                          | 85                  | 0.8%  | 2                  | 0.2%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.314 incipient cataract, anterior sutures              | 8                | 0.1%  | 0                |       |
| 100.315 incipient cataract, posterior sutures             | 26               | 0.2%  | 1                | 0.1%  |
| 100.316 incipient cataract, nucleus                       | 60               | 0.5%  | 2                | 0.2%  |
| 100.317 incipient cataract, capsular                      | 16               | 0.1%  | 3                | 0.3%  |
| 100.321 incomplete cataract, anterior cortex              | 0                |       | 5                | 0.5%  |
| 100.322 incomplete cataract, posterior cortex             | 1                | 0.0%  | 6                | 0.6%  |
| 100.323 incomplete cataract, equatorial cortex            | 0                |       | 5                | 0.5%  |
| 100.326 incomplete cataract, nucleus                      | 1                | 0.0%  | 2                | 0.2%  |
| 100.327 incomplete cataract, capsular                     | 1                | 0.0%  | 0                |       |
| 100.328 y-suture tip opacities                            | 1                | 0.0%  | 3                | 0.3%  |
| 100.330 generalized/complete cataract                     | 100              | 0.9%  | 1                | 0.1%  |
| 100.340 resorbing/hypermature cataract                    | 0                |       | 1                | 0.1%  |
| 100.345 <i>significant cataracts (summary)</i>            | 973              | 8.9%  | 61               | 5.7%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 9                | 0.1%  | 0                |       |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 9                | 0.1%  | 3                | 0.3%  |
| 110.135 PHPV/PTVL   | 4                | 0.0%  | 1                | 0.1%  |
| 110.320 vitreal degeneration                              | 25               | 0.2%  | 4                | 0.4%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 0                |       | 1                | 0.1%  |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 161              | 1.5%  | 21               | 2.0%  |
| 120.180 retinal dysplasia, geographic                     | 14               | 0.1%  | 2                | 0.2%  |
| 120.190 retinal dysplasia, detached                       | 2                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 423              | 3.9%  | 1                | 0.1%  |
| 120.400 retinal hemorrhage                                | 3                | 0.0%  | 0                |       |
| 120.960 retinopathy                                       | 2                | 0.0%  | 1                | 0.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 2                | 0.0%  | 0                |       |
| 130.120 optic nerve hypoplasia                            | 2                | 0.0%  | 1                | 0.1%  |
| 130.150 optic disc coloboma                               | 15               | 0.1%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 47               | 0.4%  | 0                |       |
| 900.100 other, not inherited                              | 242              | 2.2%  | 4                | 0.4%  |
| 900.110 other. suspect not inherited/significance unknown | 160              | 1.5%  | 54               | 5.1%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 7,413            | 67.7% | 703              | 66.0% |

## ENGLISH SETTER

|    | DISORDER   | INHERITANCE                        | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE             |
|----|--|------------------------------------|-----------|--------------------|--|
| A. | Distichiasis   | Not defined                        | 1         | Breeder option     |  |
| B. | Persistent pupillary<br>membranes<br>- iris to iris                            | Not defined                        | 1         | Breeder option     |  |
| C. | Cataract   | Not defined                        | 1         | NO                 |  |
| D. | Retinal atrophy<br>- generalized   | Presumed<br>autosomal<br>recessive | 1, 2      | NO                 |  |
| E. | Retinal atrophy<br>- rod-cone<br>dysplasia recessive<br>type 1 ( <i>rcd4</i> ) | Autosomal<br>recessive             | 3         | NO                 | Mutation in the<br><i>C2orf71</i> gene |
| F. | Retinal dysplasia-<br>folds  | Not defined                        | 1, 4      | Breeder option     |  |
| G. | Ceroid lipofuscinosis  | Autosomal<br>recessive             | 5-9       | NO                 | Mutation in the<br><i>CLN8</i> gene    |

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### Description and Comments

#### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of the dog. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal.

#### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Retinal atrophy – generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. Except for X-linked PRA in the Siberian Husky, in all breeds studied to date, PRA is inherited as an autosomal recessive trait.

E. Rod-cone dysplasia, type 4 (*rcd4*)

A form of PRA identified in the Gordon and Irish Setter breeds. Clinical night blindness is observed on average as late as 10 years of age and progresses to total blindness. This form of PRA has been referred to as late-onset PRA (LOPRA). The disorder is caused by a mutation present in the *C2orf71* gene. A DNA test is available that will unequivocally identify genetically normal, affected and carrier dogs. The test is accurate only for this mutation and will not identify other forms of PRA.

F. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

G. Ceroid lipofuscinosis

An inherited disease of humans and animals characterized by the accumulation of lipopigment in various tissues of the body including the eye. It results in progressive neurologic disease including blindness. (Also called Batten's Disease.) A DNA test is available.

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# OCULAR DISORDERS REPORT ENGLISH SETTER

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015<br>1,721 |      | 2016-2020<br>103 |      |
|------------------|--|--------------------|------|------------------|------|
|                  |  | #                  | %    | #                | %    |
| <b>EYELIDS</b>   |  |                    |      |                  |      |
| 21.000           | entropion, unspecified                                       | 8                  | 0.5% | 3                | 2.9% |
| 22.000           | ectropion, unspecified                                       | 3                  | 0.2% | 0                |      |
| 25.110           | distichiasis   | 70                 | 4.1% | 1                | 1.0% |
| <b>NICTITANS</b> |  |                    |      |                  |      |
| 52.110           | prolapsed gland of the third eyelid                          | 2                  | 0.1% | 0                |      |
| <b>CORNEA</b>    |  |                    |      |                  |      |
| 70.700           | corneal dystrophy  | 13                 | 0.8% | 1                | 1.0% |
| 70.730           | corneal endothelial degeneration                             | 3                  | 0.2% | 0                |      |
| <b>UVEA</b>      |  |                    |      |                  |      |
| 93.710           | persistent pupillary membranes, iris to iris                 | 63                 | 3.7% | 5                | 4.9% |
| 93.720           | persistent pupillary membranes, iris to lens                 | 5                  | 0.3% | 0                |      |
| 93.730           | persistent pupillary membranes, iris to cornea               | 7                  | 0.4% | 0                |      |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands | 0                  |      | 2                | 1.9% |
| 93.810           | uveal melanoma   | 0                  |      | 1                | 1.0% |
| 93.999           | uveal cysts  | 1                  | 0.1% | 0                |      |
| <b>LENS</b>      |  |                    |      |                  |      |
| 100.200          | cataract, unspecified  | 5                  | 0.3% | 0                |      |
| 100.210          | cataract. suspect not inherited/significance unknown         | 63                 | 3.7% | 5                | 4.9% |
| 100.301          | punctate cataract, anterior cortex                           | 5                  | 0.3% | 1                | 1.0% |
| 100.302          | punctate cataract, posterior cortex                          | 10                 | 0.6% | 2                | 1.9% |
| 100.305          | punctate cataract, posterior sutures                         | 3                  | 0.2% | 0                |      |
| 100.306          | punctate cataract, nucleus                                   | 2                  | 0.1% | 0                |      |
| 100.307          | punctate cataract, capsular                                  | 2                  | 0.1% | 0                |      |
| 100.311          | incipient cataract, anterior cortex                          | 5                  | 0.3% | 0                |      |
| 100.312          | incipient cataract, posterior cortex                         | 8                  | 0.5% | 1                | 1.0% |
| 100.313          | incipient cataract, equatorial cortex                        | 1                  | 0.1% | 0                |      |
| 100.315          | incipient cataract, posterior sutures                        | 2                  | 0.1% | 0                |      |
| 100.316          | incipient cataract, nucleus                                  | 2                  | 0.1% | 0                |      |
| 100.317          | incipient cataract, capsular                                 | 2                  | 0.1% | 0                |      |
| 100.321          | incomplete cataract, anterior cortex                         | 0                  |      | 1                | 1.0% |
| 100.322          | incomplete cataract, posterior cortex                        | 0                  |      | 2                | 1.9% |
| 100.326          | incomplete cataract, nucleus                                 | 0                  |      | 1                | 1.0% |
| 100.328          | y-suture tip opacities                                       | 3                  | 0.2% | 0                |      |
| 100.330          | generalized/complete cataract                                | 3                  | 0.2% | 1                | 1.0% |
| 100.345          | <i>significant cataracts (summary)</i>                       | 53                 | 3.1% | 9                | 8.7% |
| 100.375          | <i>subluxation/luxation, unspecified</i>                     | 1                  | 0.1% | 0                |      |
| <b>VITREOUS</b>  |  |                    |      |                  |      |
| 110.120          | persistent hyaloid artery/remnant                            | 7                  | 0.4% | 0                |      |
| 110.135          | PHPV/PTVL  | 1                  | 0.1% | 0                |      |
| 110.320          | vitreal degeneration   | 4                  | 0.2% | 1                | 1.0% |
| <b>RETINA</b>    |  |                    |      |                  |      |
| 120.170          | retinal dysplasia, folds                                     | 35                 | 2.0% | 3                | 2.9% |
| 120.180          | retinal dysplasia, geographic                                | 15                 | 0.9% | 0                |      |
| 120.190          | retinal dysplasia, detached                                  | 1                  | 0.1% | 0                |      |



| <b>RETINA CONTINUED</b>                                   | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 120.310 generalized progressive retinal atrophy (PRA)     | 22 1.3%          | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 1 0.1%           | 0                |
| 130.120 optic nerve hypoplasia                            | 1 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 6 0.3%           | 0                |
| 900.100 other, not inherited                              | 53 3.1%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 5 0.3%           | 2 1.9%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,451 84.3%      | 75 72.8%         |

# ENGLISH SHEPHERD

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|---|------------------------|-----------|-----------------|--------------------------------------|
| A. | Retinal atrophy<br>( <i>prcd</i> )  | Autosomal<br>recessive | 2         | NO              | Mutation in the<br><i>prcd</i> gene  |
| B. | Choroidal hypoplasia<br>(Collie Eye Anomaly)<br>- optic nerve coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- staphyloma/coloboma | Autosomal<br>recessive | 1, 3-4    | NO              | Mutation in the<br><i>NHEJ1</i> gene |

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## Description and Comments

### A. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the English Shepherd is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

### B. Choroidal hypoplasia (Collie Eye Anomaly)

- staphyloma/coloboma
- retinal detachment
- retinal hemorrhage
- optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a

recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

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# OCULAR DISORDERS REPORT ENGLISH SHEPHERD

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015<br>120 |       | 2016-2020<br>33 |       |
|--|---------------------|------------------|-------|-----------------|-------|
|  |                     | #                | %     | #               | %     |
| <b>GLOBE</b>   |                     |                  |       |                 |       |
| 0.110 microphthalmia   |                     | 2                | 1.7%  | 0               |       |
| <b>EYELIDS</b>   |                     |                  |       |                 |       |
| 21.000 entropion, unspecified                                |                     | 5                | 4.2%  | 0               |       |
| 25.110 distichiasis  |                     | 0                |       | 3               | 9.1%  |
| <b>CORNEA</b>  |                     |                  |       |                 |       |
| 70.210 corneal pannus  |                     | 1                | 0.8%  | 0               |       |
| 70.700 corneal dystrophy                                     |                     | 1                | 0.8%  | 0               |       |
| <b>UVEA</b>  |                     |                  |       |                 |       |
| 93.710 persistent pupillary membranes, iris to iris          |                     | 5                | 4.2%  | 2               | 6.1%  |
| 93.720 persistent pupillary membranes, iris to lens          |                     | 1                | 0.8%  | 0               |       |
| <b>LENS</b>  |                     |                  |       |                 |       |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 3                | 2.5%  | 1               | 3.0%  |
| 100.301 punctate cataract, anterior cortex                   |                     | 2                | 1.7%  | 0               |       |
| 100.306 punctate cataract, nucleus                           |                     | 0                |       | 1               | 3.0%  |
| 100.315 incipient cataract, posterior sutures                |                     | 1                | 0.8%  | 0               |       |
| 100.317 incipient cataract, capsular                         |                     | 1                | 0.8%  | 0               |       |
| 100.321 incomplete cataract, anterior cortex                 |                     | 2                | 1.7%  | 0               |       |
| 100.322 incomplete cataract, posterior cortex                |                     | 3                | 2.5%  | 0               |       |
| 100.330 generalized/complete cataract                        |                     | 4                | 3.3%  | 0               |       |
| 100.345 <i>significant cataracts (summary)</i>               |                     | 13               | 10.8% | 1               | 3.0%  |
| <b>RETINA</b>  |                     |                  |       |                 |       |
| 120.170 retinal dysplasia, folds                             |                     | 2                | 1.7%  | 0               |       |
| <b>OTHER</b>   |                     |                  |       |                 |       |
| 900.100 other, not inherited                                 |                     | 4                | 3.3%  | 0               |       |
| 900.110 other. suspect not inherited/significance unknown    |                     | 4                | 3.3%  | 6               | 18.2% |
| <b>NORMAL</b>  |                     |                  |       |                 |       |
| 0.000 normal globe   |                     | 97               | 80.8% | 21              | 63.6% |

# ENGLISH SPRINGER SPANIEL

|    | DISORDER   | INHERITANCE                  | REFERENCE     | BREEDING ADVICE                           | GENETIC TESTS AVAILABLE             |
|----|--|------------------------------|---------------|---|-------------------------------------|
| A. | Entropion  | Not defined                  | 1             | Breeder option                            |                                     |
| B. | Distichiasis   | Not defined                  | 1             | Breeder option                            |                                     |
| C. | Corneal dystrophy<br>- epithelial/stromal  | Not defined                  | 1             | Breeder option                            |                                     |
| D. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined   | 1<br>1        | Breeder option<br>Passes with no notation |                                     |
| E. | Cataract   | Not defined                  | 1             | NO  |                                     |
| F. | Persistent hyaloid artery  | Not defined                  | 1             | Breeder option                            |                                     |
| G. | Retinal atrophy<br>- generalized   | Not defined                  | 3             | NO  |                                     |
| H. | Retinal atrophy<br>- <i>cord-1</i>   | Autosomal recessive          | 4             | NO  | Mutation in the <i>RPGRIP1</i> gene |
| I. | Retinal dysplasia<br>- folds   | Presumed autosomal recessive | 1, 5-7, 10-11 | NO  |                                     |
| J. | Retinal dysplasia<br>- geographic  | Not defined                  | 1, 11         | NO  |                                     |
| K. | Refractive error   | Not defined                  | 8, 9          | Breeder option                            |                                     |

## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. In the English Springer Spaniel this usually involves the lower lateral lid margin.

B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted

E. Cataract

Lens opacity which may affect one or both eyes and may involve the lens partially or completely. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membranes, persistent hyaloid, or nutritional deficiencies.

Cataract in the English Springer Spaniel is reported to be a familial trait usually involving the posterior subcapsular region of the lens that progresses slowly.

F. Persistent hyaloid artery (PHA)

Congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small patent vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

G. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

H. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. PRA is inherited as an autosomal recessive trait in most breeds.

I. Retinal atrophy - *cord-1*

*Cord-1* PRA in the English Springer Spaniel has an onset of clinical signs at 2 to 9 years of age leading to blindness in most affected dogs. *Cord1* PRA in the English Springer Spaniel has been described as beginning with increased granularity of the fundus or tiny hyporeflective brown or grey patches in the far peripheral tapetum. Over time, these abnormalities become more diffuse with mottling over much of the tapetum. Vessel attenuation accompanies the more diffuse changes. In advanced cases, there is generalized tapetal hyperreflectivity and vessel attenuation. Pedigree analysis has shown *cord-1* in the English Springer Spaniel to be an autosomal recessive trait. A mutation in the *RPGRIP1* gene in cone-rod dystrophy (*cord1*) was found through genetic testing to be associated with one form of PRA in English Springer Spaniels, but not all clinically affected dogs have the *RPGRIP1* mutation, implying that other mutations have yet to be identified. A DNA test is available. The test is accurate only for this mutation and will not identify other forms of PRA. Not all dogs homozygous for the *RPGRIP1* genotype demonstrate the phenotype clinically.

J. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

The relationship between folds and geographic/detached lesions has been a topic of dispute for many years. It is the consensus of the English Springer Spaniel Field Trial Association Heritable Defects Committee (the parent breed club in the United States) that none of the forms of retinal dysplasia are desirable in a breeding animal.

K. Retinal dysplasia - geographic

Abnormal development of the retina present at birth. Any irregularly shaped area of abnormal retinal development containing both areas of thinning and areas of elevation representing folds and retinal disorganization.

L. Refractive Myopia

A condition of the eye where the light that comes in does not directly focus on the retina but in front of it. In common terminology, "near-sighted." This condition has been shown to have a genetic component in English Springer Spaniels, although the exact mode of inheritance has not been determined.

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# OCULAR DISORDERS REPORT ENGLISH SPRINGER SPANIEL

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>46,296 |      | 2016-2020<br>7,409 |      |
|---------------------|--|---------------------|------|--------------------|------|
|                     |  | #                   | %    | #                  | %    |
| <b>GLOBE</b>        |  |                     |      |                    |      |
| 0.110               | microphthalmia   | 26                  | 0.1% | 3                  | 0.0% |
| 10.000              | glaucoma   | 5                   | 0.0% | 2                  | 0.0% |
| <b>EYELIDS</b>      |  |                     |      |                    |      |
| 20.110              | eyelid dermoid   | 2                   | 0.0% | 0                  |      |
| 20.160              | macropalpebral fissure   | 3                   | 0.0% | 0                  |      |
| 21.000              | entropion, unspecified   | 278                 | 0.6% | 48                 | 0.6% |
| 22.000              | ectropion, unspecified   | 56                  | 0.1% | 7                  | 0.1% |
| 25.110              | distichiasis   | 371                 | 0.8% | 49                 | 0.7% |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 3                   | 0.0% | 8                  | 0.1% |
| 40.910              | keratoconjunctivitis sicca                                     | 11                  | 0.0% | 0                  |      |
| <b>NICTITANS</b>    |  |                     |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 0                   |      | 1                  | 0.0% |
| 52.110              | prolapsed gland of the third eyelid                            | 8                   | 0.0% | 0                  |      |
| <b>CORNEA</b>       |  |                     |      |                    |      |
| 70.210              | corneal pannus   | 6                   | 0.0% | 1                  | 0.0% |
| 70.220              | pigmentary keratitis   | 4                   | 0.0% | 0                  |      |
| 70.700              | corneal dystrophy  | 564                 | 1.2% | 110                | 1.5% |
| 70.730              | corneal endothelial degeneration                               | 12                  | 0.0% | 1                  | 0.0% |
| <b>UVEA</b>         |  |                     |      |                    |      |
| 93.110              | iris hypoplasia  | 11                  | 0.0% | 3                  | 0.0% |
| 93.140              | corneal endothelial pigment without PPM                        | 4                   | 0.0% | 0                  |      |
| 93.150              | iris coloboma  | 28                  | 0.1% | 3                  | 0.0% |
| 93.180              | iris sphincter dysplasia                                       | 0                   |      | 1                  | 0.0% |
| 93.710              | persistent pupillary membranes, iris to iris                   | 3,465               | 7.5% | 615                | 8.3% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 113                 | 0.2% | 11                 | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 90                  | 0.2% | 2                  | 0.0% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 48                  | 0.1% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 68                  | 0.1% | 59                 | 0.8% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 16                  | 0.0% | 1                  | 0.0% |
| 93.810              | uveal melanoma   | 2                   | 0.0% | 0                  |      |
| 93.999              | uveal cysts  | 17                  | 0.0% | 7                  | 0.1% |
| 97.150              | chorioretinal coloboma, congenital                             | 0                   |      | 1                  | 0.0% |
| <b>LENS</b>         |  |                     |      |                    |      |
| 100.200             | cataract, unspecified  | 97                  | 0.2% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 1,164               | 2.5% | 181                | 2.4% |
| 100.301             | punctate cataract, anterior cortex                             | 145                 | 0.3% | 25                 | 0.3% |
| 100.302             | punctate cataract, posterior cortex                            | 96                  | 0.2% | 15                 | 0.2% |
| 100.303             | punctate cataract, equatorial cortex                           | 46                  | 0.1% | 6                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 20                  | 0.0% | 2                  | 0.0% |
| 100.305             | punctate cataract, posterior sutures                           | 82                  | 0.2% | 11                 | 0.1% |
| 100.306             | punctate cataract, nucleus                                     | 33                  | 0.1% | 4                  | 0.1% |
| 100.307             | punctate cataract, capsular                                    | 34                  | 0.1% | 15                 | 0.2% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.311 incipient cataract, anterior cortex               | 185              | 0.4%  | 26               | 0.4%  |
| 100.312 incipient cataract, posterior cortex              | 186              | 0.4%  | 39               | 0.5%  |
| 100.313 incipient cataract, equatorial cortex             | 93               | 0.2%  | 9                | 0.1%  |
| 100.314 incipient cataract, anterior sutures              | 23               | 0.0%  | 2                | 0.0%  |
| 100.315 incipient cataract, posterior sutures             | 41               | 0.1%  | 4                | 0.1%  |
| 100.316 incipient cataract, nucleus                       | 60               | 0.1%  | 14               | 0.2%  |
| 100.317 incipient cataract, capsular                      | 28               | 0.1%  | 10               | 0.1%  |
| 100.321 incomplete cataract, anterior cortex              | 4                | 0.0%  | 4                | 0.1%  |
| 100.322 incomplete cataract, posterior cortex             | 1                | 0.0%  | 11               | 0.1%  |
| 100.323 incomplete cataract, equatorial cortex            | 0                |       | 1                | 0.0%  |
| 100.326 incomplete cataract, nucleus                      | 0                |       | 4                | 0.1%  |
| 100.327 incomplete cataract, capsular                     | 2                | 0.0%  | 3                | 0.0%  |
| 100.328 y-suture tip opacities                            | 8                | 0.0%  | 15               | 0.2%  |
| 100.330 generalized/complete cataract                     | 86               | 0.2%  | 5                | 0.1%  |
| 100.340 resorbing/hypermature cataract                    | 0                |       | 1                | 0.0%  |
| 100.345 <i>significant cataracts (summary)</i>            | 1,270            | 2.7%  | 226              | 3.1%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 27               | 0.1%  | 3                | 0.0%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 230              | 0.5%  | 54               | 0.7%  |
| 110.135 PHPV/PTVL   | 38               | 0.1%  | 4                | 0.1%  |
| 110.200 vitreous degeneration-anterior chamber            | 3                | 0.0%  | 0                |       |
| 110.320 vitreal degeneration                              | 193              | 0.4%  | 41               | 0.6%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 4                | 0.0%  | 0                |       |
| 97.120 coloboma   | 5                | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 1,872            | 4.0%  | 166              | 2.2%  |
| 120.180 retinal dysplasia, geographic                     | 714              | 1.5%  | 44               | 0.6%  |
| 120.190 retinal dysplasia, detached                       | 125              | 0.3%  | 4                | 0.1%  |
| 120.310 generalized progressive retinal atrophy (PRA)     | 479              | 1.0%  | 27               | 0.4%  |
| 120.400 retinal hemorrhage                                | 8                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 57               | 0.1%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 2                | 0.0%  | 1                | 0.0%  |
| 120.960 retinopathy                                       | 17               | 0.0%  | 8                | 0.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 9                | 0.0%  | 5                | 0.1%  |
| 130.120 optic nerve hypoplasia                            | 7                | 0.0%  | 2                | 0.0%  |
| 130.150 optic disc coloboma                               | 13               | 0.0%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 336              | 0.7%  | 0                |       |
| 900.100 other, not inherited                              | 722              | 1.6%  | 11               | 0.1%  |
| 900.110 other. suspect not inherited/significance unknown | 394              | 0.9%  | 207              | 2.8%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 38,123           | 82.3% | 5,797            | 78.2% |

# ENGLISH TOY SPANIEL

(King Charles, Prince Charles, Ruby, Blenheim)

|    | DISORDER   | INHERITANCE                                       | REFERENCE | BREEDING ADVICE |
|----|--|---|-----------|-----------------|
| A. | Entropion  | Not defined                                       | 1         | Breeder option  |
| B. | Distichiasis   | Not defined                                       | 1         | Breeder option  |
| C. | Corneal dystrophy<br>- epithelial/stromal  | Not defined                                       | 1         | Breeder option  |
| D. | Exposure keratopathy<br>syndrome   | Not defined                                       | 1         | Breeder option  |
| E. | Cataract   | Not defined                                       | 1         | NO              |
| F. | Persistent hyperplastic<br>primary vitreous<br>/Persistent hyperplastic<br>tunica vasculosa lentis<br>(PHPV/PHTVL) | Presumed<br>dominant/<br>incomplete<br>penetrance | 1         | NO              |
| G. | Persistent hyaloid artery  | Not defined                                       | 1         | Breeder option  |
| H. | Retinal dysplasia<br>- folds   | Presumed<br>autosomal<br>recessive                | 1         | Breeder option  |

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## Description and Comments

### A. Entropion

A conformational defect resulting in "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures, which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin, which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded;

breeding discretion is advised.

C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

D. Exposure keratopathy syndrome

A condition characterized by variable degrees of superficial vascularization, fibrosis and/or pigmentation of the cornea. May be associated with excessive exposure/irritation of the globe due to shallow orbits, lower eyelid medial entropion, lagophthalmos and macropalpebral fissure.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

Onset of cataract in the English Toy Spaniel is at an early age (less than 6 months), affecting the cortex and nucleus with rapid progression to complete cataract, resulting in blindness.

F. Persistent hyperplastic primary vitreous (PHPV)/Persistent hyperplastic tunica vasculosa lentis (PHTVL)

Persistent hyperplastic primary vitreous is a congenital defect resulting from abnormalities in the development and regression of the hyaloid artery (the primary vitreous) and the interaction of this blood vessel with the posterior lens capsule/cortex during embryogenesis. This condition is often associated with persistent hyperplastic tunica vasculosa lentis which results from failure of regression of the embryologic vascular network which surrounds the developing lens.

G. Persistent hyaloid artery (PHA)

A congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

H. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal

dysplasia is undetermined.

## **References**

There are no references providing detailed descriptions of hereditary ocular conditions of the English Toy Spaniel breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT ENGLISH TOY SPANIEL

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 0.110 microphthalmia  |                     | 4         | 0.4%  | 3         | 0.8%  |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 20.140 ectopic cilia  |                     | 1         | 0.1%  | 0         |       |
| 20.160 macropalpebral fissure   |                     | 10        | 0.9%  | 0         |       |
| 21.000 entropion, unspecified   |                     | 56        | 5.3%  | 6         | 1.6%  |
| 22.000 ectropion, unspecified   |                     | 3         | 0.3%  | 0         |       |
| 25.110 distichiasis   |                     | 124       | 11.7% | 27        | 7.1%  |
| <b>NASOLACRIMAL</b>   |                     |           |       |           |       |
| 40.910 keratoconjunctivitis sicca                                     |                     | 2         | 0.2%  | 0         |       |
| <b>NICTITANS</b>  |                     |           |       |           |       |
| 51.100 third eyelid cartilage anomaly                                 |                     | 0         |       | 1         | 0.3%  |
| 52.110 prolapsed gland of the third eyelid                            |                     | 2         | 0.2%  | 0         |       |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.210 corneal pannus   |                     | 1         | 0.1%  | 0         |       |
| 70.220 pigmentary keratitis   |                     | 20        | 1.9%  | 4         | 1.0%  |
| 70.700 corneal dystrophy  |                     | 134       | 12.6% | 68        | 17.8% |
| 70.730 corneal endothelial degeneration                               |                     | 4         | 0.4%  | 1         | 0.3%  |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 12        | 1.1%  | 4         | 1.0%  |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 2         | 0.2%  | 0         |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 1         | 0.1%  | 0         |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 4         | 0.4%  | 3         | 0.8%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1         | 0.1%  | 0         |       |
| 93.999 uveal cysts  |                     | 1         | 0.1%  | 1         | 0.3%  |
| 97.150 chorioretinal coloboma, congenital                             |                     | 0         |       | 1         | 0.3%  |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.200 cataract, unspecified   |                     | 10        | 0.9%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 62        | 5.8%  | 14        | 3.7%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 7         | 0.7%  | 3         | 0.8%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 16        | 1.5%  | 1         | 0.3%  |
| 100.303 punctate cataract, equatorial cortex                          |                     | 2         | 0.2%  | 0         |       |
| 100.305 punctate cataract, posterior sutures                          |                     | 6         | 0.6%  | 0         |       |
| 100.306 punctate cataract, nucleus                                    |                     | 3         | 0.3%  | 0         |       |
| 100.307 punctate cataract, capsular                                   |                     | 13        | 1.2%  | 9         | 2.4%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 21        | 2.0%  | 7         | 1.8%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 21        | 2.0%  | 5         | 1.3%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 2         | 0.2%  | 3         | 0.8%  |
| 100.315 incipient cataract, posterior sutures                         |                     | 1         | 0.1%  | 0         |       |
| 100.316 incipient cataract, nucleus                                   |                     | 13        | 1.2%  | 0         |       |
| 100.317 incipient cataract, capsular                                  |                     | 13        | 1.2%  | 1         | 0.3%  |
| 100.321 incomplete cataract, anterior cortex                          |                     | 3         | 0.3%  | 5         | 1.3%  |
| 100.322 incomplete cataract, posterior cortex                         |                     | 3         | 0.3%  | 6         | 1.6%  |
| 100.323 incomplete cataract, equatorial cortex                        |                     | 1         | 0.1%  | 2         | 0.5%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.326 incomplete cataract, nucleus                      | 0                | 6 1.6%           |
| 100.327 incomplete cataract, capsular                     | 1 0.1%           | 1 0.3%           |
| 100.328 y-suture tip opacities                            | 3 0.3%           | 0                |
| 100.330 generalized/complete cataract                     | 20 1.9%          | 1 0.3%           |
| 100.340 resorbing/hypermature cataract                    | 3 0.3%           | 0                |
| 100.345 <i>significant cataracts (summary)</i>            | 162 15.3%        | 50 13.1%         |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 74 7.0%          | 50 13.1%         |
| 110.135 PHPV/PTVL   | 14 1.3%          | 5 1.3%           |
| 110.320 vitreal degeneration                              | 21 2.0%          | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 57 5.4%          | 17 4.5%          |
| 120.180 retinal dysplasia, geographic                     | 7 0.7%           | 4 1.0%           |
| 120.190 retinal dysplasia, detached                       | 1 0.1%           | 1 0.3%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 6 0.6%           | 1 0.3%           |
| 120.920 retinal detachment with dialysis                  | 1 0.1%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 1 0.1%           | 0                |
| 130.150 optic disc coloboma                               | 1 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 55 5.2%          | 0                |
| 900.100 other, not inherited                              | 38 3.6%          | 1 0.3%           |
| 900.110 other. suspect not inherited/significance unknown | 48 4.5%          | 22 5.8%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 543 51.2%        | 170 44.6%        |

# ENTLEBUCHER MOUNTAIN DOG

|    | DISORDER   | INHERITANCE                        | REFERENCE | BREEDING<br>ADVICE                           | GENETIC TESTS<br>AVAILABLE          |
|----|--|------------------------------------|-----------|--|-------------------------------------|
| A. | Glaucoma   | Not defined                        | 2         | NO   |                                     |
| B. | Persistent pupillary<br>membranes<br>- iris to iris<br>- lens pigment foci/no<br>strands | Not defined<br>Not defined         | 1<br>1    | Breeder option<br>Passes with no<br>notation |                                     |
| C. | Cataract   | Presumed<br>autosomal<br>recessive | 2, 3      | NO   |                                     |
| D. | Retinal atrophy<br>( <i>prcd</i> )   | Autosomal<br>recessive             | 1, 2, 4   | NO   | Mutation in the<br><i>prcd</i> gene |
| E. | Retinal dysplasia<br>- folds   | Not defined                        | 1         | Breeder option                               |                                     |

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## Description and Comments

### A. Glaucoma

Glaucoma is an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine breed eye screening exam.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore is not noted on the certificate.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume



cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

Cataracts in the Entlebucher Mountain Dog generally become evident in young to middle-aged dogs (5.5 +/- 2.6 years). The opacities typically begin in the posterior subcapsular/capsular polar region along the suture lines as early as 1-2 years of age. Most dogs are affected with bilaterally symmetrical cataracts, which may or may not progress. Pedigree analysis suggests an autosomal recessive mode of inheritance.

D. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

E. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Entlebucher Mountain Dog is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Spiess BM. [Inherited eye diseases in the Entlebucher Mountain Dog]. *Schweizer Archiv fur Tierheilkunde*. 1994;136:105-110. Vererbte Augenkrankheiten beim Entlebucher Sennenhund.
3. Heitmann M, Hamann H, Brahm R, et al. Analysis of prevalence of presumed inherited eye diseases in Entlebucher Mountain Dogs. *Vet Ophthalmol*. 2005;8:145-151.

4. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006;88:551-563. PMID: 16938425

# OCULAR DISORDERS REPORT ENTLEBUCHER MOUNTAIN DOG

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|------------------|--|-----------|-------|-----------|-------|
|                  |  | #         | %     | #         | %     |
| <b>EYELIDS</b>   |  |           |       |           |       |
| 20.140           | ectopic cilia  | 1         | 0.1%  | 0         |       |
| 21.000           | entropion, unspecified                                       | 1         | 0.1%  | 0         |       |
| 25.110           | distichiasis   | 11        | 1.1%  | 1         | 0.3%  |
| <b>NICTITANS</b> |  |           |       |           |       |
| 52.110           | prolapsed gland of the third eyelid                          | 3         | 0.3%  | 0         |       |
| <b>CORNEA</b>    |  |           |       |           |       |
| 70.700           | corneal dystrophy  | 5         | 0.5%  | 0         |       |
| <b>UVEA</b>      |  |           |       |           |       |
| 93.710           | persistent pupillary membranes, iris to iris                 | 48        | 4.9%  | 11        | 3.6%  |
| 93.720           | persistent pupillary membranes, iris to lens                 | 4         | 0.4%  | 0         |       |
| 93.730           | persistent pupillary membranes, iris to cornea               | 2         | 0.2%  | 0         |       |
| 93.740           | persistent pupillary membranes, iris sheets                  | 1         | 0.1%  | 0         |       |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands | 7         | 0.7%  | 10        | 3.3%  |
| 93.999           | uveal cysts  | 2         | 0.2%  | 0         |       |
| <b>LENS</b>      |  |           |       |           |       |
| 100.210          | cataract. suspect not inherited/significance unknown         | 61        | 6.2%  | 24        | 7.9%  |
| 100.301          | punctate cataract, anterior cortex                           | 3         | 0.3%  | 2         | 0.7%  |
| 100.302          | punctate cataract, posterior cortex                          | 32        | 3.3%  | 9         | 3.0%  |
| 100.303          | punctate cataract, equatorial cortex                         | 5         | 0.5%  | 3         | 1.0%  |
| 100.304          | punctate cataract, anterior sutures                          | 1         | 0.1%  | 0         |       |
| 100.305          | punctate cataract, posterior sutures                         | 3         | 0.3%  | 1         | 0.3%  |
| 100.306          | punctate cataract, nucleus                                   | 2         | 0.2%  | 0         |       |
| 100.307          | punctate cataract, capsular                                  | 7         | 0.7%  | 3         | 1.0%  |
| 100.311          | incipient cataract, anterior cortex                          | 13        | 1.3%  | 1         | 0.3%  |
| 100.312          | incipient cataract, posterior cortex                         | 69        | 7.0%  | 19        | 6.2%  |
| 100.313          | incipient cataract, equatorial cortex                        | 9         | 0.9%  | 2         | 0.7%  |
| 100.315          | incipient cataract, posterior sutures                        | 4         | 0.4%  | 1         | 0.3%  |
| 100.316          | incipient cataract, nucleus                                  | 4         | 0.4%  | 0         |       |
| 100.317          | incipient cataract, capsular                                 | 10        | 1.0%  | 1         | 0.3%  |
| 100.322          | incomplete cataract, posterior cortex                        | 2         | 0.2%  | 2         | 0.7%  |
| 100.330          | generalized/complete cataract                                | 9         | 0.9%  | 0         |       |
| 100.345          | <i>significant cataracts (summary)</i>                       | 173       | 17.7% | 44        | 14.4% |
| 100.375          | <i>subluxation/luxation, unspecified</i>                     | 1         | 0.1%  | 0         |       |
| <b>VITREOUS</b>  |  |           |       |           |       |
| 110.120          | persistent hyaloid artery/remnant                            | 1         | 0.1%  | 0         |       |
| 110.200          | vitreal degeneration-anterior chamber                        | 0         |       | 2         | 0.7%  |
| 110.320          | vitreal degeneration   | 5         | 0.5%  | 9         | 3.0%  |
| <b>RETINA</b>    |  |           |       |           |       |
| 120.170          | retinal dysplasia, folds                                     | 26        | 2.7%  | 5         | 1.6%  |
| 120.180          | retinal dysplasia, geographic                                | 7         | 0.7%  | 1         | 0.3%  |
| 120.190          | retinal dysplasia, detached                                  | 1         | 0.1%  | 0         |       |
| 120.310          | generalized progressive retinal atrophy (PRA)                | 30        | 3.1%  | 0         |       |
| 120.960          | retinopathy  | 2         | 0.2%  | 0         |       |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>OPTIC NERVE</b>  |           |           |
| 130.110 micropapilla                                      | 2 0.2%    | 1 0.3%    |
| 130.120 optic nerve hypoplasia                            | 1 0.1%    | 0         |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 20 2.0%   | 0         |
| 900.100 other, not inherited                              | 39 4.0%   | 3 1.0%    |
| 900.110 other. suspect not inherited/significance unknown | 23 2.3%   | 25 8.2%   |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 725 74.0% | 209 68.5% |

# **OCULAR DISORDERS REPORT EPAGNEUL BRETON**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the EPAGNEUL BRETON breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT EPAGNEUL BRETON

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>UVEA</b>   |                     | 3         |       | 29        |       |
| 93.710 persistent pupillary membranes, iris to iris       |                     | 1         | 33.3% | 1         | 3.4%  |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.311 incipient cataract, anterior cortex               |                     | 0         |       | 1         | 3.4%  |
| 100.313 incipient cataract, equatorial cortex             |                     | 0         |       | 1         | 3.4%  |
| 100.328 y-suture tip opacities                            |                     | 0         |       | 1         | 3.4%  |
| 100.345 significant cataracts (summary)                   |                     | 0         |       | 3         | 10.3% |
| <b>VITREOUS</b>   |                     |           |       |           |       |
| 110.320 vitreal degeneration                              |                     | 0         |       | 1         | 3.4%  |
| <b>OTHER</b>  |                     |           |       |           |       |
| 900.110 other. suspect not inherited/significance unknown |                     | 0         |       | 2         | 6.9%  |
| <b>NORMAL</b>   |                     |           |       |           |       |
| 0.000 normal globe  |                     | 2         | 66.7% | 24        | 82.8% |

# EURASIER

|    | DISORDER     | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--------------|-------------|-----------|-----------------|
| A. | Glaucoma     | Not defined | 2, 3      | NO              |
| B. | Distichiasis | Not defined | 1         | Breeder option  |
| C. | Cataracts    | Not defined | 1         | NO              |

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## Description and Comments

### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure which, when sustained even for a brief period of time, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening examination.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### C. Cataracts

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Strom AR, Hassig M, Iburg TM, et al. Epidemiology of canine glaucoma presented to University of Zurich from 1995 to 2009. Part 1: Congenital and primary glaucoma (4 and 123 cases). *Vet Ophthalmol.* 2011;14:121-126. Epub 2011/03/04.

3. Rosolen SG, Boillot T, Dulaurent T, et al. Morphological, biometrical and biochemical susceptibilities for glaucoma in a healthy Eurasier dog - ECVO 2014 abstract #44. *Vet Ophthalmol.* 2014;17:E23.



# OCULAR DISORDERS REPORT EURASIER

| Diagnostic Name    | TOTAL DOGS EXAMINED  | 1991-2015<br>100 |       | 2016-2020<br>94 |       |
|--------------------|--|------------------|-------|-----------------|-------|
|                    |  | #                | %     | #               | %     |
| <b>EYELIDS</b>     |  |                  |       |                 |       |
| 25.110             | distichiasis   | 34               | 34.0% | 19              | 20.2% |
| <b>CORNEA</b>      |  |                  |       |                 |       |
| 70.700             | corneal dystrophy  | 3                | 3.0%  | 3               | 3.2%  |
| <b>UVEA</b>        |  |                  |       |                 |       |
| 93.710             | persistent pupillary membranes, iris to iris                   | 1                | 1.0%  | 3               | 3.2%  |
| 93.750             | persistent pupillary membranes, lens pigment foci/no strands   | 0                |       | 2               | 2.1%  |
| 93.760             | persistent pupillary membranes, endothelial opacity/no strands | 0                |       | 1               | 1.1%  |
| <b>LENS</b>        |  |                  |       |                 |       |
| 100.210            | cataract. suspect not inherited/significance unknown           | 5                | 5.0%  | 6               | 6.4%  |
| 100.301            | punctate cataract, anterior cortex                             | 0                |       | 1               | 1.1%  |
| 100.302            | punctate cataract, posterior cortex                            | 2                | 2.0%  | 0               |       |
| 100.305            | punctate cataract, posterior sutures                           | 0                |       | 2               | 2.1%  |
| 100.307            | punctate cataract, capsular                                    | 1                | 1.0%  | 0               |       |
| 100.312            | incipient cataract, posterior cortex                           | 0                |       | 3               | 3.2%  |
| 100.315            | incipient cataract, posterior sutures                          | 0                |       | 1               | 1.1%  |
| 100.328            | y-suture tip opacities   | 0                |       | 1               | 1.1%  |
| 100.345            | <i>significant cataracts (summary)</i>                         | 3                | 3.0%  | 8               | 8.5%  |
| <b>VITREOUS</b>    |  |                  |       |                 |       |
| 110.120            | persistent hyaloid artery/remnant                              | 1                | 1.0%  | 0               |       |
| 110.320            | vitreal degeneration   | 0                |       | 1               | 1.1%  |
| <b>OPTIC NERVE</b> |  |                  |       |                 |       |
| 130.110            | micropapilla   | 0                |       | 1               | 1.1%  |
| <b>OTHER</b>       |  |                  |       |                 |       |
| 900.000            | other, unspecified   | 5                | 5.0%  | 0               |       |
| 900.100            | other, not inherited   | 5                | 5.0%  | 0               |       |
| 900.110            | other. suspect not inherited/significance unknown              | 4                | 4.0%  | 0               |       |
| <b>NORMAL</b>      |  |                  |       |                 |       |
| 0.000              | normal globe   | 63               | 63.0% | 56              | 59.6% |

# FIELD SPANIEL

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING<br>ADVICE      | GENETIC TESTS<br>AVAILABLE       |
|----|--|---------------------|-----------|-------------------------|----------------------------------|
| A. | Distichiasis                                     | Not defined         | 1         | Breeder option          |                                  |
| B. | Imperforate lacrimal punctum                     | Not defined         | 1         | Breeder option          |                                  |
| C. | Corneal dystrophy<br>- epithelial/stromal        | Not defined         | 1         | Breeder option          |                                  |
| D. | Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option          |                                  |
|    | - lens pigment foci/no strands                   | Not defined         | 1         | Passes with no notation |                                  |
| E. | Cataract   | Not defined         | 1         | NO                      |                                  |
| F. | Retinal atrophy ( <i>prcd</i> )                  | Autosomal recessive | 1         | NO                      | Mutation in the <i>prcd</i> gene |
| G. | Retinal dysplasia<br>- folds                     | Not defined         | 1         | Breeder option          |                                  |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin, which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Imperforate lacrimal punctum

A developmental anomaly resulting in failure of opening of the lacrimal duct located at the medial lid margins. The lower punctum is more frequently affected. This defect usually results in epiphora, an overflow of tears onto the face.

### C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal

layers; usually inherited and bilateral.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

F. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Field Spaniel is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

G. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT FIELD SPANIEL

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>2,411 |      | 2016-2020<br>738 |      |
|---|---------------------|--------------------|------|------------------|------|
|   |                     | #                  | %    | #                | %    |
| <b>GLOBE</b>  |                     |                    |      |                  |      |
| 0.110 microphthalmia  |                     | 0                  |      | 2                | 0.3% |
| <b>EYELIDS</b>  |                     |                    |      |                  |      |
| 20.160 macropalpebral fissure   |                     | 6                  | 0.2% | 0                |      |
| 21.000 entropion, unspecified   |                     | 10                 | 0.4% | 0                |      |
| 22.000 ectropion, unspecified   |                     | 11                 | 0.5% | 0                |      |
| 25.110 distichiasis   |                     | 156                | 6.5% | 32               | 4.3% |
| <b>NASOLACRIMAL</b>   |                     |                    |      |                  |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 4                  | 0.2% | 13               | 1.8% |
| <b>NICTITANS</b>  |                     |                    |      |                  |      |
| 52.110 prolapsed gland of the third eyelid                            |                     | 1                  | 0.0% | 0                |      |
| <b>CORNEA</b>   |                     |                    |      |                  |      |
| 70.220 pigmentary keratitis   |                     | 1                  | 0.0% | 0                |      |
| 70.700 corneal dystrophy  |                     | 28                 | 1.2% | 6                | 0.8% |
| 70.730 corneal endothelial degeneration                               |                     | 1                  | 0.0% | 0                |      |
| <b>UVEA</b>   |                     |                    |      |                  |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 137                | 5.7% | 68               | 9.2% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 6                  | 0.2% | 0                |      |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 7                  | 0.3% | 1                | 0.1% |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 16                 | 0.7% | 26               | 3.5% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 4                  | 0.2% | 3                | 0.4% |
| <b>LENS</b>   |                     |                    |      |                  |      |
| 100.200 cataract, unspecified   |                     | 3                  | 0.1% | 0                |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 117                | 4.9% | 33               | 4.5% |
| 100.301 punctate cataract, anterior cortex                            |                     | 14                 | 0.6% | 7                | 0.9% |
| 100.302 punctate cataract, posterior cortex                           |                     | 3                  | 0.1% | 0                |      |
| 100.304 punctate cataract, anterior sutures                           |                     | 2                  | 0.1% | 1                | 0.1% |
| 100.305 punctate cataract, posterior sutures                          |                     | 1                  | 0.0% | 0                |      |
| 100.306 punctate cataract, nucleus                                    |                     | 2                  | 0.1% | 0                |      |
| 100.307 punctate cataract, capsular                                   |                     | 7                  | 0.3% | 1                | 0.1% |
| 100.311 incipient cataract, anterior cortex                           |                     | 14                 | 0.6% | 3                | 0.4% |
| 100.312 incipient cataract, posterior cortex                          |                     | 5                  | 0.2% | 3                | 0.4% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 1                  | 0.0% | 0                |      |
| 100.314 incipient cataract, anterior sutures                          |                     | 2                  | 0.1% | 1                | 0.1% |
| 100.315 incipient cataract, posterior sutures                         |                     | 5                  | 0.2% | 0                |      |
| 100.316 incipient cataract, nucleus                                   |                     | 7                  | 0.3% | 1                | 0.1% |
| 100.317 incipient cataract, capsular                                  |                     | 4                  | 0.2% | 1                | 0.1% |
| 100.321 incomplete cataract, anterior cortex                          |                     | 1                  | 0.0% | 0                |      |
| 100.322 incomplete cataract, posterior cortex                         |                     | 1                  | 0.0% | 0                |      |
| 100.328 y-suture tip opacities  |                     | 3                  | 0.1% | 1                | 0.1% |
| 100.330 generalized/complete cataract                                 |                     | 2                  | 0.1% | 1                | 0.1% |
| 100.345 significant cataracts (summary)                               |                     | 77                 | 3.2% | 20               | 2.7% |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>VITREOUS</b>   |             |           |
| 110.120 persistent hyaloid artery/remnant                 | 3 0.1%      | 1 0.1%    |
| 110.135 PHPV/PTVL   | 4 0.2%      | 0         |
| 110.200 vitreous degeneration-anterior chamber            | 0           | 4 0.5%    |
| 110.320 vitreal degeneration                              | 2 0.1%      | 0         |
| <b>FUNDUS</b>   |             |           |
| 97.120 coloboma   | 1 0.0%      | 0         |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 250 10.4%   | 43 5.8%   |
| 120.180 retinal dysplasia, geographic                     | 9 0.4%      | 4 0.5%    |
| 120.190 retinal dysplasia, detached                       | 1 0.0%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 3 0.1%      | 2 0.3%    |
| 120.400 retinal hemorrhage                                | 4 0.2%      | 0         |
| 120.910 retinal detachment without dialysis               | 1 0.0%      | 0         |
| 120.960 retinopathy                                       | 0           | 1 0.1%    |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 1 0.0%      | 2 0.3%    |
| 130.120 optic nerve hypoplasia                            | 1 0.0%      | 0         |
| 130.150 optic disc coloboma                               | 0           | 2 0.3%    |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 47 1.9%     | 0         |
| 900.100 other, not inherited                              | 61 2.5%     | 0         |
| 900.110 other. suspect not inherited/significance unknown | 48 2.0%     | 37 5.0%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,770 73.4% | 515 69.8% |

# FINNISH LAPPHUND

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|--|---------------------|-----------|-----------------|-----------------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option  |                                   |
| B. | Cataract   | Not defined         | 1         | NO              |                                   |
| C. | Retinal atrophy<br>( <i>prcd</i> )               | Autosomal recessive | 2         | NO              | Mutation in the <i>prcd</i> gene  |
| D. | Multifocal retinopathy<br>- <i>cmr3</i>          | Autosomal recessive | 3         | NO              | Mutation in the <i>BEST1</i> gene |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the form of PRA in the Finnish Lapphund is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in

dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

D. Multifocal retinopathy (*cmr3*)

Canine Multifocal Retinopathy type 3 (*cmr3*) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Clinically the disease is similar to that seen in the Bullmastiff and Coton deTulear, but the mutation in the Bestrophin 1 gene (*BEST1* alias *VMD2*) is different. The multifocal retinopathy seen in the Lapponian Herder is caused by a deletion at position 1,388 and a substitution at position 1,466 and is therefore called *cmr3*. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006 Nov;88:551-563. PMID: 16938425
3. Guziewicz KE, Zangerl B, Lindauer SJ, et al. Bestrophin gene mutations cause canine multifocal retinopathy: a novel animal model for best disease. *Invest Ophthalmol Vis Sci*. 2007 May;48:1959-1967.

# OCULAR DISORDERS REPORT FINNISH LAPPHUND

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 1         | 0.2%  | 1         | 0.6%  |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.220          | pigmentary keratitis   | 1         | 0.2%  | 0         |       |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 51        | 9.8%  | 19        | 11.1% |
| 93.720          | persistent pupillary membranes, iris to lens                 | 1         | 0.2%  | 0         |       |
| 93.730          | persistent pupillary membranes, iris to cornea               | 6         | 1.2%  | 0         |       |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 1         | 0.2%  | 5         | 2.9%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 37        | 7.1%  | 8         | 4.7%  |
| 100.301         | punctate cataract, anterior cortex                           | 1         | 0.2%  | 0         |       |
| 100.302         | punctate cataract, posterior cortex                          | 7         | 1.3%  | 1         | 0.6%  |
| 100.303         | punctate cataract, equatorial cortex                         | 0         |       | 1         | 0.6%  |
| 100.305         | punctate cataract, posterior sutures                         | 2         | 0.4%  | 1         | 0.6%  |
| 100.306         | punctate cataract, nucleus                                   | 2         | 0.4%  | 0         |       |
| 100.307         | punctate cataract, capsular                                  | 0         |       | 2         | 1.2%  |
| 100.311         | incipient cataract, anterior cortex                          | 1         | 0.2%  | 1         | 0.6%  |
| 100.312         | incipient cataract, posterior cortex                         | 1         | 0.2%  | 2         | 1.2%  |
| 100.313         | incipient cataract, equatorial cortex                        | 2         | 0.4%  | 1         | 0.6%  |
| 100.315         | incipient cataract, posterior sutures                        | 0         |       | 1         | 0.6%  |
| 100.316         | incipient cataract, nucleus                                  | 0         |       | 1         | 0.6%  |
| 100.328         | y-suture tip opacities                                       | 0         |       | 1         | 0.6%  |
| 100.330         | generalized/complete cataract                                | 1         | 0.2%  | 1         | 0.6%  |
| 100.345         | <i>significant cataracts (summary)</i>                       | 17        | 3.3%  | 13        | 7.6%  |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.120         | persistent hyaloid artery/remnant                            | 0         |       | 1         | 0.6%  |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                                     | 10        | 1.9%  | 1         | 0.6%  |
| 120.310         | generalized progressive retinal atrophy (PRA)                | 1         | 0.2%  | 0         |       |
| 120.960         | retinopathy  | 1         | 0.2%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.000         | other, unspecified   | 10        | 1.9%  | 0         |       |
| 900.100         | other, not inherited   | 14        | 2.7%  | 0         |       |
| 900.110         | other. suspect not inherited/significance unknown            | 9         | 1.7%  | 2         | 1.2%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 427       | 82.1% | 124       | 72.5% |



# FINNISH SPITZ

| DISORDER  | INHERITANCE | REFERENCE | BREEDING<br>ADVICE      |
|---|-------------|-----------|-------------------------|
| A. Persistent pupillary membranes<br>- lens pigment foci/no strands | Not defined | 1         | Passes with no notation |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Finnish Spitz breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT FINNISH SPITZ

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>247 |       | 2016-2020<br>15 |       |
|---|---------------------|------------------|-------|-----------------|-------|
|   |                     | #                | %     | #               | %     |
| <b>EYELIDS</b>  |                     |                  |       |                 |       |
| 20.140 ectopic cilia  |                     | 1                | 0.4%  | 0               |       |
| <b>CORNEA</b>   |                     |                  |       |                 |       |
| 70.700 corneal dystrophy  |                     | 2                | 0.8%  | 0               |       |
| <b>UVEA</b>   |                     |                  |       |                 |       |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 2                | 0.8%  | 0               |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 2                | 0.8%  | 8               | 53.3% |
| <b>LENS</b>   |                     |                  |       |                 |       |
| 100.200 cataract, unspecified                                       |                     | 1                | 0.4%  | 0               |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 33               | 13.4% | 1               | 6.7%  |
| 100.301 punctate cataract, anterior cortex                          |                     | 2                | 0.8%  | 0               |       |
| 100.302 punctate cataract, posterior cortex                         |                     | 1                | 0.4%  | 0               |       |
| 100.304 punctate cataract, anterior sutures                         |                     | 1                | 0.4%  | 0               |       |
| 100.307 punctate cataract, capsular                                 |                     | 2                | 0.8%  | 0               |       |
| 100.311 incipient cataract, anterior cortex                         |                     | 1                | 0.4%  | 0               |       |
| 100.312 incipient cataract, posterior cortex                        |                     | 1                | 0.4%  | 0               |       |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 9                | 3.6%  | 0               |       |
| <b>VITREOUS</b>   |                     |                  |       |                 |       |
| 110.120 persistent hyaloid artery/remnant                           |                     | 4                | 1.6%  | 0               |       |
| 110.320 vitreal degeneration  |                     | 3                | 1.2%  | 1               | 6.7%  |
| <b>RETINA</b>   |                     |                  |       |                 |       |
| 120.170 retinal dysplasia, folds                                    |                     | 2                | 0.8%  | 0               |       |
| 120.310 generalized progressive retinal atrophy (PRA)               |                     | 6                | 2.4%  | 0               |       |
| <b>OTHER</b>  |                     |                  |       |                 |       |
| 900.000 other, unspecified  |                     | 3                | 1.2%  | 0               |       |
| 900.100 other, not inherited  |                     | 8                | 3.2%  | 0               |       |
| 900.110 other. suspect not inherited/significance unknown           |                     | 2                | 0.8%  | 0               |       |
| <b>NORMAL</b>   |                     |                  |       |                 |       |
| 0.000 normal globe  |                     | 195              | 78.9% | 7               | 46.7% |

# FLAT-COATED RETRIEVER

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--------------------------------|-------------|-----------|-------------------------|
| A. | Glaucoma                       | Not defined | 2-4       | NO                      |
| B. | Distichiasis                   | Not defined | 1         | Breeder option          |
| C. | Persistent pupillary membranes |             |           |                         |
|    | - iris to iris                 | Not defined | 1         | Breeder option          |
|    | - lens pigment foci/no strands | Not defined | 1         | Passes with no notation |
| D. | Cataract                       | Not defined | 1         | NO                      |
| E. | Y-suture tip opacity           | Not defined | 1         | Breeder option          |
| F. | Retinopathy                    | Not defined | 1         | Breeder Option          |

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## Description and Comments

### A. Glaucoma (with pectinate ligament abnormality)

Glaucoma is characterized by an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of the intraocular pressure (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine breed eye screening exam.

Flat-Coated Retrievers have been shown to have a higher prevalence of pectinate ligament abnormalities compared with other breeds. There is a significant association between pectinate ligament abnormalities and glaucoma in this breed. The heritability of pectinate ligament abnormalities in Flat-Coated Retrievers is estimated at 0.7. Since glaucoma and pectinate ligament abnormalities are closely associated, glaucoma may also be heritable.

In a recent report, pectinate ligament abnormalities were prevalent and significantly associated with age in a population of Flat-Coated Retrievers in the UK.

Due to the incidence of PLD in the breed and the increased progression observed with age, it may be prudent to perform repeated gonioscopy examinations over time.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin, which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong

recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

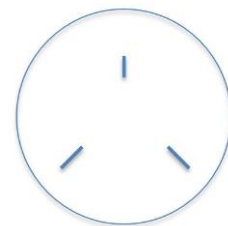
Lens pigment foci/no strands is considered an insignificant finding and therefore is not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region. The exact frequency and significance of cataracts in the breed is not known.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## F. Retinopathy

Patchy focal unilateral or bilateral hyper reflective tapetal lesions most frequently peripheral but occasionally central around a pigmented spot, usually non progressive. Not usually present prior to 3 months of age but usually present by 18 months of age.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Read RA, Wood JL, Lakhani KH, Read RA. Pectinate ligament dysplasia (PLD) and glaucoma in Flat-Coated Retrievers. I. Objectives, technique and results of a PLD survey. *Vet Ophthalmol.* 1998;1:85-90.
3. Read RA, Wood JL, Lakhani KH, Read RA. Pectinate ligament dysplasia (PLD) and glaucoma in Flat-Coated Retrievers. II. Assessment of prevalence and heritability. *Vet Ophthalmol.* 1998;1:91-99.
4. Oliver JA, Ekiri A, Mellersh CS. Prevalence of pectinate ligament dysplasia and associations with age, sex and intraocular pressure in the Basset Hound, Flat-Coated Retriever and Dandie Dinmont Terrier. *Can Genet Epidemiol* 2016 March 12;3:1doi: 10.1186/s40575-016-0033-1.

# OCULAR DISORDERS REPORT FLAT-COATED RETRIEVER

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015<br>8,690 |       | 2016-2020<br>1,922 |       |
|------------------|--|--------------------|-------|--------------------|-------|
|                  |  | #                  | %     | #                  | %     |
| <b>GLOBE</b>     |  |                    |       |                    |       |
| 0.110            | microphthalmia   | 2                  | 0.0%  | 1                  | 0.1%  |
| 10.000           | glaucoma   | 2                  | 0.0%  | 0                  |       |
| <b>EYELIDS</b>   |  |                    |       |                    |       |
| 20.140           | ectopic cilia  | 9                  | 0.1%  | 1                  | 0.1%  |
| 20.160           | macropalpebral fissure   | 2                  | 0.0%  | 0                  |       |
| 21.000           | entropion, unspecified   | 16                 | 0.2%  | 4                  | 0.2%  |
| 22.000           | ectropion, unspecified   | 33                 | 0.4%  | 2                  | 0.1%  |
| 25.110           | distichiasis   | 1,097              | 12.6% | 245                | 12.7% |
| <b>NICTITANS</b> |  |                    |       |                    |       |
| 50.210           | pannus of third eyelid   | 1                  | 0.0%  | 0                  |       |
| 52.110           | prolapsed gland of the third eyelid                            | 4                  | 0.0%  | 0                  |       |
| <b>CORNEA</b>    |  |                    |       |                    |       |
| 70.220           | pigmentary keratitis   | 2                  | 0.0%  | 1                  | 0.1%  |
| 70.700           | corneal dystrophy  | 52                 | 0.6%  | 12                 | 0.6%  |
| 70.730           | corneal endothelial degeneration                               | 3                  | 0.0%  | 0                  |       |
| <b>UVEA</b>      |  |                    |       |                    |       |
| 93.110           | iris hypoplasia  | 2                  | 0.0%  | 0                  |       |
| 93.140           | corneal endothelial pigment without PPM                        | 1                  | 0.0%  | 0                  |       |
| 93.710           | persistent pupillary membranes, iris to iris                   | 208                | 2.4%  | 77                 | 4.0%  |
| 93.720           | persistent pupillary membranes, iris to lens                   | 14                 | 0.2%  | 1                  | 0.1%  |
| 93.740           | persistent pupillary membranes, iris sheets                    | 3                  | 0.0%  | 0                  |       |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands   | 60                 | 0.7%  | 62                 | 3.2%  |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands | 5                  | 0.1%  | 0                  |       |
| 93.810           | uveal melanoma   | 1                  | 0.0%  | 3                  | 0.2%  |
| 93.999           | uveal cysts  | 28                 | 0.3%  | 6                  | 0.3%  |
| <b>LENS</b>      |  |                    |       |                    |       |
| 100.200          | cataract, unspecified  | 16                 | 0.2%  | 0                  |       |
| 100.210          | cataract. suspect not inherited/significance unknown           | 987                | 11.4% | 292                | 15.2% |
| 100.301          | punctate cataract, anterior cortex                             | 74                 | 0.9%  | 25                 | 1.3%  |
| 100.302          | punctate cataract, posterior cortex                            | 17                 | 0.2%  | 4                  | 0.2%  |
| 100.303          | punctate cataract, equatorial cortex                           | 8                  | 0.1%  | 4                  | 0.2%  |
| 100.304          | punctate cataract, anterior sutures                            | 24                 | 0.3%  | 7                  | 0.4%  |
| 100.305          | punctate cataract, posterior sutures                           | 16                 | 0.2%  | 10                 | 0.5%  |
| 100.306          | punctate cataract, nucleus                                     | 10                 | 0.1%  | 4                  | 0.2%  |
| 100.307          | punctate cataract, capsular                                    | 9                  | 0.1%  | 12                 | 0.6%  |
| 100.311          | incipient cataract, anterior cortex                            | 38                 | 0.4%  | 12                 | 0.6%  |
| 100.312          | incipient cataract, posterior cortex                           | 21                 | 0.2%  | 5                  | 0.3%  |
| 100.313          | incipient cataract, equatorial cortex                          | 16                 | 0.2%  | 5                  | 0.3%  |
| 100.314          | incipient cataract, anterior sutures                           | 6                  | 0.1%  | 4                  | 0.2%  |
| 100.315          | incipient cataract, posterior sutures                          | 10                 | 0.1%  | 2                  | 0.1%  |
| 100.316          | incipient cataract, nucleus                                    | 5                  | 0.1%  | 4                  | 0.2%  |
| 100.317          | incipient cataract, capsular                                   | 4                  | 0.0%  | 1                  | 0.1%  |
| 100.321          | incomplete cataract, anterior cortex                           | 0                  |       | 1                  | 0.1%  |
| 100.323          | incomplete cataract, equatorial cortex                         | 1                  | 0.0%  | 0                  |       |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.326 incomplete cataract, nucleus                      | 0                | 1 0.1%           |
| 100.328 y-suture tip opacities                            | 8 0.1%           | 28 1.5%          |
| 100.330 generalized/complete cataract                     | 6 0.1%           | 2 0.1%           |
| 100.345 <i>significant cataracts (summary)</i>            | 289 3.3%         | 131 6.8%         |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 3 0.0%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 13 0.1%          | 7 0.4%           |
| 110.135 PHPV/PTVL   | 5 0.1%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 0                | 2 0.1%           |
| 110.320 vitreal degeneration                              | 1 0.0%           | 3 0.2%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 1 0.0%           | 0                |
| 97.120 coloboma   | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 18 0.2%          | 8 0.4%           |
| 120.180 retinal dysplasia, geographic                     | 12 0.1%          | 3 0.2%           |
| 120.190 retinal dysplasia, detached                       | 0                | 1 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 51 0.6%          | 5 0.3%           |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| 120.920 retinal detachment with dialysis                  | 1 0.0%           | 3 0.2%           |
| 120.960 retinopathy                                       | 14 0.2%          | 18 0.9%          |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 5 0.1%           | 4 0.2%           |
| 130.120 optic nerve hypoplasia                            | 3 0.0%           | 0                |
| 130.150 optic disc coloboma                               | 21 0.2%          | 8 0.4%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 160 1.8%         | 0                |
| 900.100 other, not inherited                              | 267 3.1%         | 6 0.3%           |
| 900.110 other. suspect not inherited/significance unknown | 150 1.7%         | 155 8.1%         |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 6,576 75.7%      | 1,129 58.7%      |

# FRENCH BULLDOG

|    | DISORDER                         | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE          |
|----|----------------------------------|---------------------|-----------|-----------------|----------------------------------|
| A. | Entropion                        | Not defined         | 1         | Breeder option  |                                  |
| B. | Distichiasis                     | Not defined         | 1         | Breeder option  |                                  |
| C. | Imperforate lacrimal punctum     | Not defined         | 1         | Breeder option  |                                  |
| D. | Persistent pupillary membranes   |                     |           |                 |                                  |
|    | - iris to iris                   | Not defined         | 1         | Breeder option  |                                  |
|    | - iris to cornea                 | Not defined         | 1         | NO              |                                  |
|    | - endothelial opacity/no strands | Not defined         | 1         | NO              |                                  |
| E. | Cataract                         | Autosomal recessive | 1, 2      | NO              | Mutation in the <i>HSF4</i> gene |
| F. | Retinal dysplasia - folds        | Not defined         | 1         | Breeder option  |                                  |

## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of the dog. It is difficult to make a strong recommendation with regards to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded and breeding discretion is advised.

### C. Imperforate lacrimal punctum

A developmental anomaly resulting in failure of opening of the lacrimal duct located at the medial lid margins. The lower punctum is more frequently affected. This defect usually results in epiphora, an overflow of tears onto the face.



D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the French Bulldog, the condition is inherited as an autosomal recessive mutation in the *HSF4* gene (*HSF4-1*). A DNA test is available.

F. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

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# OCULAR DISORDERS REPORT FRENCH BULLDOG

| Diagnostic Name   | TOTAL DOGS EXAMINED |      | 1991-2015<br>3,852 |      | 2016-2020<br>2,322 |      |
|---|---------------------|------|--------------------|------|--------------------|------|
|   | #                   | %    | #                  | %    | #                  | %    |
| <b>GLOBE</b>  |                     |      |                    |      |                    |      |
| 0.110 microphthalmia  | 1                   | 0.0% | 1                  | 0.0% | 1                  | 0.0% |
| <b>EYELIDS</b>  |                     |      |                    |      |                    |      |
| 20.140 ectopic cilia  | 1                   | 0.0% | 2                  | 0.1% | 2                  | 0.1% |
| 20.160 macropalpebral fissure   | 3                   | 0.1% | 0                  |      | 0                  |      |
| 21.000 entropion, unspecified   | 40                  | 1.0% | 23                 | 1.0% | 23                 | 1.0% |
| 22.000 ectropion, unspecified   | 6                   | 0.2% | 2                  | 0.1% | 2                  | 0.1% |
| 25.110 distichiasis   | 261                 | 6.8% | 131                | 5.6% | 131                | 5.6% |
| <b>NASOLACRIMAL</b>   |                     |      |                    |      |                    |      |
| 32.110 imperforate lower nasolacrimal punctum                         | 26                  | 0.7% | 49                 | 2.1% | 49                 | 2.1% |
| 40.910 keratoconjunctivitis sicca                                     | 3                   | 0.1% | 1                  | 0.0% | 1                  | 0.0% |
| <b>NICTITANS</b>  |                     |      |                    |      |                    |      |
| 50.210 pannus of third eyelid   | 1                   | 0.0% | 1                  | 0.0% | 1                  | 0.0% |
| 52.110 prolapsed gland of the third eyelid                            | 6                   | 0.2% | 4                  | 0.2% | 4                  | 0.2% |
| <b>CORNEA</b>   |                     |      |                    |      |                    |      |
| 70.210 corneal pannus   | 4                   | 0.1% | 0                  |      | 0                  |      |
| 70.220 pigmentary keratitis   | 20                  | 0.5% | 12                 | 0.5% | 12                 | 0.5% |
| 70.700 corneal dystrophy  | 32                  | 0.8% | 16                 | 0.7% | 16                 | 0.7% |
| 70.730 corneal endothelial degeneration                               | 6                   | 0.2% | 1                  | 0.0% | 1                  | 0.0% |
| <b>UVEA</b>   |                     |      |                    |      |                    |      |
| 93.150 iris coloboma  | 1                   | 0.0% | 2                  | 0.1% | 2                  | 0.1% |
| 93.710 persistent pupillary membranes, iris to iris                   | 98                  | 2.5% | 65                 | 2.8% | 65                 | 2.8% |
| 93.720 persistent pupillary membranes, iris to lens                   | 6                   | 0.2% | 1                  | 0.0% | 1                  | 0.0% |
| 93.730 persistent pupillary membranes, iris to cornea                 | 55                  | 1.4% | 20                 | 0.9% | 20                 | 0.9% |
| 93.740 persistent pupillary membranes, iris sheets                    | 3                   | 0.1% | 0                  |      | 0                  |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   | 8                   | 0.2% | 4                  | 0.2% | 4                  | 0.2% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands | 37                  | 1.0% | 41                 | 1.8% | 41                 | 1.8% |
| 93.810 uveal melanoma   | 1                   | 0.0% | 1                  | 0.0% | 1                  | 0.0% |
| 93.999 uveal cysts  | 9                   | 0.2% | 2                  | 0.1% | 2                  | 0.1% |
| 97.150 chorioretinal coloboma, congenital                             | 1                   | 0.0% | 0                  |      | 0                  |      |
| <b>LENS</b>   |                     |      |                    |      |                    |      |
| 100.210 cataract. suspect not inherited/significance unknown          | 91                  | 2.4% | 41                 | 1.8% | 41                 | 1.8% |
| 100.301 punctate cataract, anterior cortex                            | 11                  | 0.3% | 7                  | 0.3% | 7                  | 0.3% |
| 100.302 punctate cataract, posterior cortex                           | 4                   | 0.1% | 2                  | 0.1% | 2                  | 0.1% |
| 100.303 punctate cataract, equatorial cortex                          | 7                   | 0.2% | 4                  | 0.2% | 4                  | 0.2% |
| 100.305 punctate cataract, posterior sutures                          | 2                   | 0.1% | 1                  | 0.0% | 1                  | 0.0% |
| 100.306 punctate cataract, nucleus                                    | 2                   | 0.1% | 8                  | 0.3% | 8                  | 0.3% |
| 100.307 punctate cataract, capsular                                   | 1                   | 0.0% | 4                  | 0.2% | 4                  | 0.2% |
| 100.311 incipient cataract, anterior cortex                           | 40                  | 1.0% | 6                  | 0.3% | 6                  | 0.3% |
| 100.312 incipient cataract, posterior cortex                          | 14                  | 0.4% | 1                  | 0.0% | 1                  | 0.0% |
| 100.313 incipient cataract, equatorial cortex                         | 17                  | 0.4% | 4                  | 0.2% | 4                  | 0.2% |
| 100.314 incipient cataract, anterior sutures                          | 3                   | 0.1% | 0                  |      | 0                  |      |
| 100.315 incipient cataract, posterior sutures                         | 4                   | 0.1% | 0                  |      | 0                  |      |
| 100.316 incipient cataract, nucleus                                   | 10                  | 0.3% | 6                  | 0.3% | 6                  | 0.3% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.317 incipient cataract, capsular                      | 5 0.1%           | 6 0.3%           |
| 100.321 incomplete cataract, anterior cortex              | 1 0.0%           | 3 0.1%           |
| 100.322 incomplete cataract, posterior cortex             | 1 0.0%           | 2 0.1%           |
| 100.323 incomplete cataract, equatorial cortex            | 0                | 1 0.0%           |
| 100.326 incomplete cataract, nucleus                      | 1 0.0%           | 5 0.2%           |
| 100.330 generalized/complete cataract                     | 18 0.5%          | 1 0.0%           |
| 100.340 resorbing/hypermature cataract                    | 0                | 1 0.0%           |
| 100.345 <i>significant cataracts (summary)</i>            | 141 3.7%         | 62 2.7%          |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 15 0.4%          | 19 0.8%          |
| 110.135 PHPV/PTVL   | 1 0.0%           | 2 0.1%           |
| 110.320 vitreal degeneration                              | 7 0.2%           | 7 0.3%           |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 86 2.2%          | 44 1.9%          |
| 120.180 retinal dysplasia, geographic                     | 10 0.3%          | 10 0.4%          |
| 120.310 generalized progressive retinal atrophy (PRA)     | 1 0.0%           | 0                |
| 120.400 retinal hemorrhage                                | 1 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| 120.920 retinal detachment with dialysis                  | 1 0.0%           | 0                |
| 120.960 retinopathy                                       | 2 0.1%           | 1 0.0%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 0                | 1 0.0%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 65 1.7%          | 0                |
| 900.100 other, not inherited                              | 88 2.3%          | 9 0.4%           |
| 900.110 other. suspect not inherited/significance unknown | 48 1.2%          | 104 4.5%         |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 3,174 82.4%      | 1,748 75.3%      |

# GERMAN PINSCHER

|    | <b>DISORDER</b>  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b>  |
|----|--|--------------------|------------------|-------------------------|
| A. | Distichiasis   | Not defined        | 1                | Breeder Option          |
| B. | Persistent pupillary membranes<br>- lens pigment foci/no strands | Not defined        | 1                | Passes with no notation |
| C. | Cataract   | Not defined        | 1                | NO                      |
| D. | Persistent hyperplastic tunica vasculosa lentis (PHTVL)          | Not defined        | 1-3              | NO                      |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of the dog. It is difficult to make a strong recommendation with regards to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded and breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

There may be more than one type of inherited cataract in German Pinschers. One form is reported in Finland with a later age of onset in which a pedigree analysis suggested autosomal recessive or incomplete dominant inheritance (4). Another form is reported in Germany with an earlier age of onset in which a pedigree analysis suggested autosomal recessive inheritance (5). Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Persistent hyperplastic tunica vasculosa lentis (PHTVL)

Persistent tunica vasculosa lentis results from the failure of regression of the embryologic vascular network which surrounds the developing lens. This disorder has been observed in German Pinschers in Finland and Germany. A pedigree analysis suggested recessive or incomplete dominant inheritance (4).

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2. Leppanen M, Martenson J, Maki K. Results of ophthalmologic screening examinations of German Pinschers in Finland--a retrospective study. *Vet Ophthalmol.* 2001;4:165-169.
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# OCULAR DISORDERS REPORT GERMAN PINSCHER

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|------------------|--|-----------|-------|-----------|-------|
|                  |  | #         | %     | #         | %     |
| <b>EYELIDS</b>   |  |           |       |           |       |
| 25.110           | distichiasis   | 5         | 0.4%  | 9         | 2.1%  |
| <b>NICTITANS</b> |  |           |       |           |       |
| 52.110           | prolapsed gland of the third eyelid                          | 1         | 0.1%  | 0         |       |
| <b>CORNEA</b>    |  |           |       |           |       |
| 70.220           | pigmentary keratitis   | 0         |       | 2         | 0.5%  |
| 70.700           | corneal dystrophy  | 19        | 1.7%  | 1         | 0.2%  |
| <b>UVEA</b>      |  |           |       |           |       |
| 93.710           | persistent pupillary membranes, iris to iris                 | 8         | 0.7%  | 2         | 0.5%  |
| 93.720           | persistent pupillary membranes, iris to lens                 | 5         | 0.4%  | 0         |       |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands | 10        | 0.9%  | 14        | 3.3%  |
| <b>LENS</b>      |  |           |       |           |       |
| 100.210          | cataract. suspect not inherited/significance unknown         | 74        | 6.6%  | 36        | 8.5%  |
| 100.301          | punctate cataract, anterior cortex                           | 15        | 1.3%  | 4         | 0.9%  |
| 100.302          | punctate cataract, posterior cortex                          | 24        | 2.1%  | 4         | 0.9%  |
| 100.303          | punctate cataract, equatorial cortex                         | 0         |       | 2         | 0.5%  |
| 100.304          | punctate cataract, anterior sutures                          | 6         | 0.5%  | 0         |       |
| 100.305          | punctate cataract, posterior sutures                         | 9         | 0.8%  | 0         |       |
| 100.306          | punctate cataract, nucleus                                   | 1         | 0.1%  | 0         |       |
| 100.307          | punctate cataract, capsular                                  | 6         | 0.5%  | 4         | 0.9%  |
| 100.311          | incipient cataract, anterior cortex                          | 19        | 1.7%  | 7         | 1.7%  |
| 100.312          | incipient cataract, posterior cortex                         | 37        | 3.3%  | 7         | 1.7%  |
| 100.313          | incipient cataract, equatorial cortex                        | 7         | 0.6%  | 3         | 0.7%  |
| 100.314          | incipient cataract, anterior sutures                         | 6         | 0.5%  | 0         |       |
| 100.315          | incipient cataract, posterior sutures                        | 8         | 0.7%  | 1         | 0.2%  |
| 100.316          | incipient cataract, nucleus                                  | 5         | 0.4%  | 2         | 0.5%  |
| 100.317          | incipient cataract, capsular                                 | 8         | 0.7%  | 4         | 0.9%  |
| 100.321          | incomplete cataract, anterior cortex                         | 0         |       | 3         | 0.7%  |
| 100.322          | incomplete cataract, posterior cortex                        | 1         | 0.1%  | 4         | 0.9%  |
| 100.325          | incomplete cataract, posterior sutures                       | 0         |       | 2         | 0.5%  |
| 100.327          | incomplete cataract, capsular                                | 0         |       | 1         | 0.2%  |
| 100.328          | y-suture tip opacities                                       | 1         | 0.1%  | 0         |       |
| 100.330          | generalized/complete cataract                                | 8         | 0.7%  | 2         | 0.5%  |
| 100.345          | <i>significant cataracts (summary)</i>                       | 161       | 14.3% | 50        | 11.8% |
| <b>VITREOUS</b>  |  |           |       |           |       |
| 110.120          | persistent hyaloid artery/remnant                            | 2         | 0.2%  | 0         |       |
| 110.135          | PHPV/PTVL  | 4         | 0.4%  | 0         |       |
| 110.320          | vitreal degeneration   | 14        | 1.2%  | 2         | 0.5%  |
| <b>RETINA</b>    |  |           |       |           |       |
| 120.170          | retinal dysplasia, folds                                     | 2         | 0.2%  | 0         |       |
| 120.180          | retinal dysplasia, geographic                                | 1         | 0.1%  | 0         |       |
| 120.400          | retinal hemorrhage   | 1         | 0.1%  | 0         |       |
| 120.960          | retinopathy  | 2         | 0.2%  | 0         |       |

|   | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 10 0.9%          | 5 1.2%           |
| 130.120 optic nerve hypoplasia                            | 6 0.5%           | 1 0.2%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 26 2.3%          | 0                |
| 900.100 other, not inherited                              | 31 2.8%          | 1 0.2%           |
| 900.110 other. suspect not inherited/significance unknown | 17 1.5%          | 33 7.8%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 913 81.2%        | 293 69.4%        |

## GERMAN SHEPHERD DOG

|    | <b>DISORDER</b>  | <b>INHERITANCE</b>                 | <b>REFERENCE</b> | <b>BREEDING<br/>ADVICE</b>                   | <b>GENETIC TESTS<br/>AVAILABLE</b>   |
|----|--|------------------------------------|------------------|--|--------------------------------------|
| A. | Distichiasis   | Not defined                        | 1                | Breeder option                               |                                      |
| B. | Plasmoma/atypical<br>pannus  | Not defined                        | 1                | NO   |                                      |
| C. | Corneal dystrophy<br>- epithelial/stromal  | Not defined                        | 1, 2             | Breeder option                               |                                      |
| D. | Chronic superficial<br>keratitis/pannus  | Not defined                        | 1, 3-9           | NO   |                                      |
| E. | Persistent pupillary<br>membranes<br>- iris to iris<br>- lens pigment foci/no<br>strands | Not defined<br>Not defined         | 1<br>1           | Breeder option<br>Passes with no<br>notation |                                      |
| F. | Cataract<br>- Cortical   | Presumed<br>autosomal<br>recessive | 1, 10            | NO   |                                      |
| G. | Y-suture tip opacity   | Not defined                        | 1                | Breeder option                               |                                      |
| H. | Cone degeneration<br>- hemeralopia/<br>achromatopsia                                     | Autosomal<br>recessive             | 11               | NO   | Mutation in the<br><i>CNGA3</i> gene |
| I. | Retinal dysplasia<br>- folds   | Not defined                        | 1, 12            | Breeder option                               |                                      |
| J. | Micropapilla   | Not defined                        | 1                | Breeder option                               |                                      |
| K. | Limbal melanoma  | Not defined                        | 1, 13            | NO   |                                      |



## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Plasmoma/atypical pannus

Bilateral lymphocytic/plasmocytic infiltration of the nictitating membranes which may occur independent of corneal Pannus.

### C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

### D. Chronic superficial keratitis/pannus

A bilateral inflammatory disease of the cornea which usually starts as a grayish haze to the ventral or ventrolateral cornea, followed by the formation of a vascularized subepithelial growth that begins to spread toward the central cornea; pigmentation follows the vascularization. If severe, vision impairment occurs. Pannus may be associated with plasma cell infiltration of the nictitans which may also occur independent of corneal disease.

The German Shepherd Dog has a higher incidence of pannus than any other breed. The MHC class II risk haplotype has been shown. Although there are likely several other genes and environmental factors that contribute to CSK, a recent paper suggested that MHC class II is a major genetic risk factor. Dogs with the risk haplotype were 2.7 times more likely to develop CSK. Homozygosity of the risk haplotype increased the risk of CSK to over eightfold.

### E. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### F. Cataract

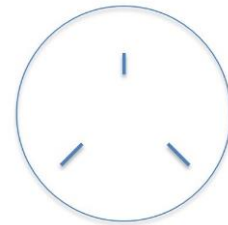
A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume

cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

**Cortical:** Reported by Barnett in Great Britain, opacities are first apparent at 8-12 weeks of age, in the posterior cortex and progress to involve the Y-sutures and nucleus. The equatorial subcapsular cortex is unaffected. No progression is noted after 1-2 years of age. Test breeding suggests an autosomal recessive mode of inheritance.

G. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

H. Cone degeneration - hemeralopia/achromatopsia

Autosomal recessively inherited early degeneration of the cone photoreceptors. Afflicted puppies develop day-blindness and colorblindness. Afflicted dogs remain ophthalmoscopically normal their entire life. Electroretinography is required to definitively diagnose the disorder. A 5-month-old German Shepherd puppy with vision loss during daylight hours was recently identified with a mutation in the *CNGA3* gene.

I. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic,

detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

J. Optic nerve hypoplasia

A congenital defect of the optic nerve which causes blindness and abnormal pupil response in the affected eye. May be unable to differentiate from micropapilla on a routine (dilated) screening ophthalmoscopic exam.

K. Micropapilla

Micropapilla refers to a small optic disc which is not associated with vision impairment. Optic nerve hypoplasia refers to a congenital defect of the optic nerve which causes blindness and abnormal pupil response in the affected eye. It may be difficult to differentiate between micropapilla and optic nerve hypoplasia on a routine (dilated) screening ophthalmoscopic exam.

L. Limbal melanoma

Most limbal melanomas are really epibulbar melanocytomas, but there is a possibility of an extension of an intraocular melanoma extending outward and presenting as a limbal melanoma. An epibulbar melanocytoma originates from the superficial pigment lining the limbus and the lesion may eventually extend into the eye. Metastasis has not been documented and the mass is characterized by large epithelioid cells. The lesion presents as a subconjunctival smooth mass most commonly in the dorsolateral limbal region and extends later into the cornea and posterior on the sclera. Breed predisposition have been noted in the German Shepherd, Labrador and Golden Retriever.

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# OCULAR DISORDERS REPORT GERMAN SHEPHERD DOG

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>4,694 |      | 2016-2020<br>961 |      |
|---------------------|--|--------------------|------|------------------|------|
|                     |  | #                  | %    | #                | %    |
| <b>GLOBE</b>        |  |                    |      |                  |      |
| 0.110               | microphthalmia   | 7                  | 0.1% | 2                | 0.2% |
| 10.000              | glaucoma   | 3                  | 0.1% | 0                |      |
| <b>EYELIDS</b>      |  |                    |      |                  |      |
| 20.140              | ectopic cilia  | 1                  | 0.0% | 0                |      |
| 20.160              | macropalpebral fissure   | 1                  | 0.0% | 0                |      |
| 21.000              | entropion, unspecified   | 3                  | 0.1% | 2                | 0.2% |
| 22.000              | ectropion, unspecified   | 4                  | 0.1% | 0                |      |
| 25.110              | distichiasis   | 53                 | 1.1% | 4                | 0.4% |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 1                  | 0.0% | 0                |      |
| 40.910              | keratoconjunctivitis sicca                                     | 3                  | 0.1% | 0                |      |
| <b>NICTITANS</b>    |  |                    |      |                  |      |
| 50.210              | pannus of third eyelid   | 10                 | 0.2% | 15               | 1.6% |
| 51.100              | third eyelid cartilage anomaly                                 | 3                  | 0.1% | 1                | 0.1% |
| 52.110              | prolapsed gland of the third eyelid                            | 1                  | 0.0% | 0                |      |
| <b>CORNEA</b>       |  |                    |      |                  |      |
| 70.210              | corneal pannus   | 109                | 2.3% | 16               | 1.7% |
| 70.220              | pigmentary keratitis   | 1                  | 0.0% | 1                | 0.1% |
| 70.700              | corneal dystrophy  | 217                | 4.6% | 43               | 4.5% |
| 70.730              | corneal endothelial degeneration                               | 2                  | 0.0% | 0                |      |
| <b>UVEA</b>         |  |                    |      |                  |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 64                 | 1.4% | 16               | 1.7% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 16                 | 0.3% | 0                |      |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 8                  | 0.2% | 3                | 0.3% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 2                  | 0.0% | 1                | 0.1% |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 12                 | 0.3% | 10               | 1.0% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 1                  | 0.0% | 2                | 0.2% |
| 93.810              | uveal melanoma   | 1                  | 0.0% | 2                | 0.2% |
| 93.999              | uveal cysts  | 22                 | 0.5% | 7                | 0.7% |
| <b>LENS</b>         |  |                    |      |                  |      |
| 100.200             | cataract, unspecified  | 28                 | 0.6% | 0                |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 242                | 5.2% | 85               | 8.8% |
| 100.301             | punctate cataract, anterior cortex                             | 27                 | 0.6% | 9                | 0.9% |
| 100.302             | punctate cataract, posterior cortex                            | 14                 | 0.3% | 3                | 0.3% |
| 100.303             | punctate cataract, equatorial cortex                           | 13                 | 0.3% | 0                |      |
| 100.304             | punctate cataract, anterior sutures                            | 1                  | 0.0% | 1                | 0.1% |
| 100.305             | punctate cataract, posterior sutures                           | 14                 | 0.3% | 6                | 0.6% |
| 100.306             | punctate cataract, nucleus                                     | 31                 | 0.7% | 11               | 1.1% |
| 100.307             | punctate cataract, capsular                                    | 8                  | 0.2% | 2                | 0.2% |
| 100.311             | incipient cataract, anterior cortex                            | 34                 | 0.7% | 7                | 0.7% |
| 100.312             | incipient cataract, posterior cortex                           | 30                 | 0.6% | 8                | 0.8% |
| 100.313             | incipient cataract, equatorial cortex                          | 20                 | 0.4% | 3                | 0.3% |
| 100.314             | incipient cataract, anterior sutures                           | 3                  | 0.1% | 2                | 0.2% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.315 incipient cataract, posterior sutures             | 7 0.1%           | 2 0.2%           |
| 100.316 incipient cataract, nucleus                       | 58 1.2%          | 11 1.1%          |
| 100.317 incipient cataract, capsular                      | 2 0.0%           | 5 0.5%           |
| 100.321 incomplete cataract, anterior cortex              | 0                | 1 0.1%           |
| 100.322 incomplete cataract, posterior cortex             | 1 0.0%           | 1 0.1%           |
| 100.323 incomplete cataract, equatorial cortex            | 0                | 1 0.1%           |
| 100.325 incomplete cataract, posterior sutures            | 0                | 1 0.1%           |
| 100.326 incomplete cataract, nucleus                      | 1 0.0%           | 2 0.2%           |
| 100.327 incomplete cataract, capsular                     | 1 0.0%           | 1 0.1%           |
| 100.328 y-suture tip opacities                            | 5 0.1%           | 7 0.7%           |
| 100.330 generalized/complete cataract                     | 21 0.4%          | 5 0.5%           |
| 100.345 <i>significant cataracts (summary)</i>            | 319 6.8%         | 89 9.3%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 8 0.2%           | 1 0.1%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 6 0.1%           | 3 0.3%           |
| 110.135 PHPV/PTVL   | 3 0.1%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 1 0.0%           | 1 0.1%           |
| 110.320 vitreal degeneration                              | 13 0.3%          | 1 0.1%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 89 1.9%          | 13 1.4%          |
| 120.180 retinal dysplasia, geographic                     | 17 0.4%          | 2 0.2%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 20 0.4%          | 1 0.1%           |
| 120.910 retinal detachment without dialysis               | 4 0.1%           | 0                |
| 120.920 retinal detachment with dialysis                  | 1 0.0%           | 1 0.1%           |
| 120.960 retinopathy                                       | 2 0.0%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 25 0.5%          | 12 1.2%          |
| 130.120 optic nerve hypoplasia                            | 35 0.7%          | 2 0.2%           |
| 130.150 optic disc coloboma                               | 3 0.1%           | 1 0.1%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 58 1.2%          | 0                |
| 900.100 other, not inherited                              | 144 3.1%         | 6 0.6%           |
| 900.110 other. suspect not inherited/significance unknown | 75 1.6%          | 60 6.2%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 3,603 76.8%      | 654 68.1%        |

# GERMAN SHORTHAIRED POINTER

|    | DISORDER                             | INHERITANCE         | REFERENCE | BREEDING<br>ADVICE      | GENETIC TESTS<br>AVAILABLE        |
|----|--------------------------------------|---------------------|-----------|-------------------------|-----------------------------------|
| A. | Distichiasis                         | Not defined         | 1         | Breeder option          |                                   |
| B. | Nictitans cartilage anomaly/eversion | Not defined         | 1, 2      | Breeder option          |                                   |
| C. | Persistent pupillary membranes       |                     |           |                         |                                   |
|    | - iris to iris                       | Not defined         | 1         | Breeder option          |                                   |
|    | - lens pigment foci/no strands       | Not defined         | 1         | Passes with no notation |                                   |
| D. | Cataract                             | Not defined         | 1         | NO                      |                                   |
| E. | Persistent hyaloid artery            | Not defined         | 1         | Breeder option          |                                   |
| F. | Retinal dysplasia - folds            | Not defined         | 1         | Breeder option          |                                   |
| G. | Cone degeneration - (achromatopsia)  | Autosomal recessive | 3         | NO                      | Mutation in the <i>CNGB3</i> gene |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Nictitans cartilage anomaly/eversion

A scroll-like curling of the cartilage of the third eyelid, usually everting the margin. This condition may occur in one or both eyes and may cause mild ocular irritation.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Persistent hyaloid artery (PHA)

A congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

F. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. Except for X-linked PRA in the Siberian Husky, in all breeds studied to date, PRA is inherited as an autosomal recessive trait.

G. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

H. Cone degeneration - hemeralopia/achromatopsia

Autosomal recessively inherited early degeneration of the cone photoreceptors. Afflicted puppies develop day-blindness, colorblindness, and photophobia between 8 and 12 weeks of age. Afflicted dogs remain ophthalmoscopically normal their entire life. Electroretinography is required to definitively diagnose the disorder. A missense mutation in the same gene (*CNGB3*) that has been identified in CD-affected Alaskan Malamute-derived dogs has been detected in German Shorthaired Pointers affected with a clinically identical allelic disorder. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Martin CL. Everted membrana nictitans in German Shorthaired Pointers. *J Am Vet Med Assoc.* 1970 Nov 1;157:1229-1232.



3. Sidjanin DJ, Lowe JK, McElwee JL, et al. Canine CNGB3 mutations establish cone degeneration as orthologous to the human achromatopsia locus ACHM3. *Human Molecular Genetics*. 2002;11:1823-1833.

# OCULAR DISORDERS REPORT GERMAN SHORTHAIRED POINTER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>6,095 |      | 2016-2020<br>2,028 |      |
|---------------------|--|--------------------|------|--------------------|------|
|                     |  | #                  | %    | #                  | %    |
| <b>GLOBE</b>        |  |                    |      |                    |      |
| 10.000              | glaucoma   | 1                  | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                    |      |                    |      |
| 20.160              | macropalpebral fissure   | 1                  | 0.0% | 0                  |      |
| 21.000              | entropion, unspecified   | 10                 | 0.2% | 6                  | 0.3% |
| 22.000              | ectropion, unspecified   | 4                  | 0.1% | 1                  | 0.0% |
| 25.110              | distichiasis   | 221                | 3.6% | 106                | 5.2% |
| <b>NASOLACRIMAL</b> |  |                    |      |                    |      |
| 40.910              | keratoconjunctivitis sicca                                     | 1                  | 0.0% | 0                  |      |
| <b>NICTITANS</b>    |  |                    |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 3                  | 0.0% | 0                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 1                  | 0.0% | 1                  | 0.0% |
| <b>CORNEA</b>       |  |                    |      |                    |      |
| 70.210              | corneal pannus   | 1                  | 0.0% | 0                  |      |
| 70.700              | corneal dystrophy  | 20                 | 0.3% | 1                  | 0.0% |
| 70.730              | corneal endothelial degeneration                               | 1                  | 0.0% | 0                  |      |
| <b>UVEA</b>         |  |                    |      |                    |      |
| 93.110              | iris hypoplasia  | 1                  | 0.0% | 1                  | 0.0% |
| 93.140              | corneal endothelial pigment without PPM                        | 1                  | 0.0% | 0                  |      |
| 93.150              | iris coloboma  | 2                  | 0.0% | 0                  |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 412                | 6.8% | 133                | 6.6% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 17                 | 0.3% | 2                  | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 5                  | 0.1% | 4                  | 0.2% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 1                  | 0.0% | 1                  | 0.0% |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 16                 | 0.3% | 29                 | 1.4% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 3                  | 0.0% | 1                  | 0.0% |
| 93.810              | uveal melanoma   | 1                  | 0.0% | 0                  |      |
| 93.999              | uveal cysts  | 6                  | 0.1% | 4                  | 0.2% |
| 97.150              | chorioretinal coloboma, congenital                             | 0                  |      | 1                  | 0.0% |
| <b>LENS</b>         |  |                    |      |                    |      |
| 100.200             | cataract, unspecified  | 9                  | 0.1% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 313                | 5.1% | 75                 | 3.7% |
| 100.301             | punctate cataract, anterior cortex                             | 25                 | 0.4% | 6                  | 0.3% |
| 100.302             | punctate cataract, posterior cortex                            | 48                 | 0.8% | 10                 | 0.5% |
| 100.303             | punctate cataract, equatorial cortex                           | 12                 | 0.2% | 2                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 2                  | 0.0% | 1                  | 0.0% |
| 100.305             | punctate cataract, posterior sutures                           | 11                 | 0.2% | 6                  | 0.3% |
| 100.306             | punctate cataract, nucleus                                     | 15                 | 0.2% | 3                  | 0.1% |
| 100.307             | punctate cataract, capsular                                    | 9                  | 0.1% | 6                  | 0.3% |
| 100.311             | incipient cataract, anterior cortex                            | 18                 | 0.3% | 3                  | 0.1% |
| 100.312             | incipient cataract, posterior cortex                           | 91                 | 1.5% | 16                 | 0.8% |
| 100.313             | incipient cataract, equatorial cortex                          | 20                 | 0.3% | 1                  | 0.0% |
| 100.314             | incipient cataract, anterior sutures                           | 2                  | 0.0% | 0                  |      |
| 100.315             | incipient cataract, posterior sutures                          | 16                 | 0.3% | 3                  | 0.1% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.316 incipient cataract, nucleus                       | 18 0.3%          | 5 0.2%           |
| 100.317 incipient cataract, capsular                      | 12 0.2%          | 6 0.3%           |
| 100.321 incomplete cataract, anterior cortex              | 1 0.0%           | 1 0.0%           |
| 100.322 incomplete cataract, posterior cortex             | 6 0.1%           | 1 0.0%           |
| 100.325 incomplete cataract, posterior sutures            | 1 0.0%           | 0                |
| 100.326 incomplete cataract, nucleus                      | 1 0.0%           | 1 0.0%           |
| 100.328 y-suture tip opacities                            | 1 0.0%           | 8 0.4%           |
| 100.330 generalized/complete cataract                     | 14 0.2%          | 1 0.0%           |
| 100.340 resorbing/hypermature cataract                    | 1 0.0%           | 0                |
| 100.345 <i>significant cataracts (summary)</i>            | 333 5.5%         | 80 3.9%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 2 0.0%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 8 0.1%           | 21 1.0%          |
| 110.135 PHPV/PTVL   | 13 0.2%          | 3 0.1%           |
| 110.200 vitreous degeneration-anterior chamber            | 1 0.0%           | 0                |
| 110.320 vitreal degeneration                              | 22 0.4%          | 9 0.4%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 129 2.1%         | 19 0.9%          |
| 120.180 retinal dysplasia, geographic                     | 24 0.4%          | 3 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 9 0.1%           | 0                |
| 120.920 retinal detachment with dialysis                  | 2 0.0%           | 1 0.0%           |
| 120.960 retinopathy                                       | 4 0.1%           | 4 0.2%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 3 0.0%           | 0                |
| 130.120 optic nerve hypoplasia                            | 4 0.1%           | 1 0.0%           |
| 130.150 optic disc coloboma                               | 1 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 99 1.6%          | 0                |
| 900.100 other, not inherited                              | 134 2.2%         | 5 0.2%           |
| 900.110 other. suspect not inherited/significance unknown | 67 1.1%          | 87 4.3%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 4,949 81.2%      | 1,535 75.7%      |

# GERMAN SPITZ

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|------------------------------------|------------------------|-----------|--------------------|-------------------------------------|
| A. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 1         | NO                 | Mutation in the <i>prcd</i><br>gene |

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## Description and Comments

### A. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the German Spitz is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. However in the American Eskimo Dog the phenotype can be very variable in the age of onset. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

Other forms of retinal degeneration that are not PRCD are recognized in the breed. The currently available genetic test will not detect these other forms of PRA.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the German Spitz breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT GERMAN SPITZ

| Diagnostic Name | TOTAL DOGS EXAMINED                          | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>UVEA</b>     |  | 5         |       | 28        |       |
| 93.710          | persistent pupillary membranes, iris to iris | 1         | 20.0% | 1         | 3.6%  |
| 93.720          | persistent pupillary membranes, iris to lens | 0         |       | 1         | 3.6%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.311         | incipient cataract, anterior cortex          | 0         |       | 1         | 3.6%  |
| 100.345         | significant cataracts (summary)              | 0         |       | 1         | 3.6%  |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.120         | persistent hyaloid artery/remnant            | 0         |       | 5         | 17.9% |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.960         | retinopathy                                  | 1         | 20.0% | 0         |       |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe                                 | 4         | 80.0% | 21        | 75.0% |

# GERMAN WIREHAISED POINTER

(Drathaar, Deutsch Drathaar)

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Distichiasis                                     | Not defined | 1         | Breeder option  |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| C. | Cataract   | Not defined | 1         | NO              |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the German Wirehaired Pointer breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT GERMAN WIREHAired POINTER

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015<br>666 |       | 2016-2020<br>385 |       |
|-----------------|--|------------------|-------|------------------|-------|
|                 |  | #                | %     | #                | %     |
| <b>EYELIDS</b>  |  |                  |       |                  |       |
| 20.160          | macropalpebral fissure                                       | 1                | 0.2%  | 0                |       |
| 25.110          | distichiasis   | 9                | 1.4%  | 6                | 1.6%  |
| <b>CORNEA</b>   |  |                  |       |                  |       |
| 70.730          | corneal endothelial degeneration                             | 0                |       | 1                | 0.3%  |
| <b>UVEA</b>     |  |                  |       |                  |       |
| 93.110          | iris hypoplasia  | 0                |       | 1                | 0.3%  |
| 93.710          | persistent pupillary membranes, iris to iris                 | 8                | 1.2%  | 14               | 3.6%  |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 0                |       | 1                | 0.3%  |
| 93.999          | uveal cysts  | 0                |       | 1                | 0.3%  |
| <b>LENS</b>     |  |                  |       |                  |       |
| 100.200         | cataract, unspecified  | 5                | 0.8%  | 0                |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 18               | 2.7%  | 15               | 3.9%  |
| 100.301         | punctate cataract, anterior cortex                           | 2                | 0.3%  | 0                |       |
| 100.302         | punctate cataract, posterior cortex                          | 5                | 0.8%  | 2                | 0.5%  |
| 100.305         | punctate cataract, posterior sutures                         | 1                | 0.2%  | 2                | 0.5%  |
| 100.312         | incipient cataract, posterior cortex                         | 10               | 1.5%  | 2                | 0.5%  |
| 100.315         | incipient cataract, posterior sutures                        | 1                | 0.2%  | 0                |       |
| 100.316         | incipient cataract, nucleus                                  | 0                |       | 2                | 0.5%  |
| 100.317         | incipient cataract, capsular                                 | 3                | 0.5%  | 0                |       |
| 100.327         | incomplete cataract, capsular                                | 0                |       | 1                | 0.3%  |
| 100.328         | y-suture tip opacities                                       | 0                |       | 2                | 0.5%  |
| 100.330         | generalized/complete cataract                                | 2                | 0.3%  | 0                |       |
| 100.345         | significant cataracts (summary)                              | 29               | 4.4%  | 11               | 2.9%  |
| <b>VITREOUS</b> |  |                  |       |                  |       |
| 110.120         | persistent hyaloid artery/remnant                            | 2                | 0.3%  | 2                | 0.5%  |
| 110.200         | vitreous degeneration-anterior chamber                       | 0                |       | 1                | 0.3%  |
| 110.320         | vitreal degeneration   | 2                | 0.3%  | 1                | 0.3%  |
| <b>RETINA</b>   |  |                  |       |                  |       |
| 120.170         | retinal dysplasia, folds                                     | 3                | 0.5%  | 0                |       |
| 120.180         | retinal dysplasia, geographic                                | 1                | 0.2%  | 1                | 0.3%  |
| 120.190         | retinal dysplasia, detached                                  | 1                | 0.2%  | 0                |       |
| 120.910         | retinal detachment without dialysis                          | 1                | 0.2%  | 0                |       |
| <b>OTHER</b>    |  |                  |       |                  |       |
| 900.000         | other, unspecified   | 9                | 1.4%  | 0                |       |
| 900.100         | other, not inherited   | 8                | 1.2%  | 0                |       |
| 900.110         | other. suspect not inherited/significance unknown            | 14               | 2.1%  | 11               | 2.9%  |
| <b>NORMAL</b>   |  |                  |       |                  |       |
| 0.000           | normal globe   | 589              | 88.4% | 328              | 85.2% |



# GIANT SCHNAUZER

|    | DISORDER   | INHERITANCE                                | REFERENCE | BREEDING ADVICE                           | GENETIC TESTS AVAILABLE  |
|----|--|--|-----------|---|--|
| A. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined                 | 1<br>1    | Breeder option<br>Passes with no notation |  |
| B. | Cataract   | Not defined                                | 1         | NO  |  |
| C. | Y-suture tip opacity   | Not defined                                | 1         | Breeder option                            |  |
| D. | Retinal atrophy ( <i>prcd</i> )<br>Retinal Atrophy                                 | Autosomal recessive<br>Autosomal recessive | 1<br>2    | NO<br>NO                                  | Mutation in the <i>prcd</i> gene<br>Mutation in the <i>NECAP1</i> gene |
| E. | Retinal dysplasia<br>- folds   | Not defined                                | 1         | Breeder option                            |  |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

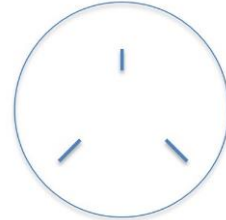
Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

C. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

D. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A genetic test is available.

E. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Hitti, R. J., et al. (2019). "Whole Genome Sequencing of Giant Schnauzer Dogs with Progressive Retinal Atrophy Establishes NECAP1 as a Novel Candidate Gene for Retinal Degeneration." Genes (Basel) 10(5). PMID: 31117272

# OCULAR DISORDERS REPORT

## GIANT SCHNAUZER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 0.110 microphthalmia  |                     | 1         | 0.1% | 0         |      |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 21.000 entropion, unspecified   |                     | 0         |      | 1         | 0.3% |
| 25.110 distichiasis   |                     | 5         | 0.4% | 2         | 0.5% |
| <b>NASOLACRIMAL</b>   |                     |           |      |           |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 0         |      | 2         | 0.5% |
| <b>NICTITANS</b>  |                     |           |      |           |      |
| 51.100 third eyelid cartilage anomaly                                 |                     | 10        | 0.9% | 3         | 0.8% |
| 52.110 prolapsed gland of the third eyelid                            |                     | 2         | 0.2% | 0         |      |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.700 corneal dystrophy  |                     | 1         | 0.1% | 2         | 0.5% |
| 70.730 corneal endothelial degeneration                               |                     | 1         | 0.1% | 0         |      |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 55        | 4.9% | 12        | 3.1% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 4         | 0.4% | 0         |      |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 6         | 0.5% | 0         |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 7         | 0.6% | 11        | 2.8% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1         | 0.1% | 0         |      |
| 93.999 uveal cysts  |                     | 2         | 0.2% | 2         | 0.5% |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.200 cataract, unspecified   |                     | 5         | 0.4% | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 56        | 5.0% | 20        | 5.2% |
| 100.301 punctate cataract, anterior cortex                            |                     | 4         | 0.4% | 1         | 0.3% |
| 100.302 punctate cataract, posterior cortex                           |                     | 8         | 0.7% | 0         |      |
| 100.304 punctate cataract, anterior sutures                           |                     | 1         | 0.1% | 0         |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 2         | 0.2% | 2         | 0.5% |
| 100.306 punctate cataract, nucleus                                    |                     | 1         | 0.1% | 0         |      |
| 100.307 punctate cataract, capsular                                   |                     | 7         | 0.6% | 3         | 0.8% |
| 100.311 incipient cataract, anterior cortex                           |                     | 3         | 0.3% | 1         | 0.3% |
| 100.312 incipient cataract, posterior cortex                          |                     | 24        | 2.1% | 4         | 1.0% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 8         | 0.7% | 1         | 0.3% |
| 100.315 incipient cataract, posterior sutures                         |                     | 4         | 0.4% | 1         | 0.3% |
| 100.316 incipient cataract, nucleus                                   |                     | 2         | 0.2% | 1         | 0.3% |
| 100.317 incipient cataract, capsular                                  |                     | 2         | 0.2% | 3         | 0.8% |
| 100.322 incomplete cataract, posterior cortex                         |                     | 0         |      | 1         | 0.3% |
| 100.328 y-suture tip opacities  |                     | 1         | 0.1% | 5         | 1.3% |
| 100.330 generalized/complete cataract                                 |                     | 2         | 0.2% | 0         |      |
| 100.345 significant cataracts (summary)                               |                     | 74        | 6.6% | 23        | 5.9% |
| 100.375 subluxation/luxation, unspecified                             |                     | 2         | 0.2% | 0         |      |
| <b>VITREOUS</b>   |                     |           |      |           |      |
| 110.120 persistent hyaloid artery/remnant                             |                     | 5         | 0.4% | 4         | 1.0% |
| 110.135 PHPV/PTVL   |                     | 5         | 0.4% | 0         |      |

| <b>VITREOUS CONTINUED</b>                                 | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 110.320 vitreal degeneration                              | 2 0.2%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 27 2.4%          | 2 0.5%           |
| 120.180 retinal dysplasia, geographic                     | 1 0.1%           | 2 0.5%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 8 0.7%           | 0                |
| 120.960 retinopathy                                       | 1 0.1%           | 1 0.3%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 0                | 1 0.3%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 26 2.3%          | 0                |
| 900.100 other, not inherited                              | 19 1.7%          | 1 0.3%           |
| 900.110 other. suspect not inherited/significance unknown | 12 1.1%          | 15 3.9%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 950 84.1%        | 304 78.6%        |

# GLEN OF IMAAL TERRIER

|    | DISORDER                              | INHERITANCE            | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|---------------------------------------|------------------------|-----------|-----------------|--------------------------------------|
| A. | Distichiasis                          | Not defined            | 1         | Breeder option  |                                      |
| B. | Cataract                              | Not defined            | 1         | NO              |                                      |
| C. | Retinal atrophy<br>- generalized      | Not defined            | 1, 2      | NO              |                                      |
| D. | Cone rod dystrophy<br>( <i>crd3</i> ) | Autosomal<br>recessive | 3, 4      | NO              | Mutation in the<br><i>ADAM9</i> gene |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Retinal atrophy-generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

#### D. Cone rod dystrophy

A form of late-onset PRA identified in Glen of Imaal Terriers. Ophthalmoscopic lesions are typically diagnosed by 5 years of age, however lesions may be present as early as 3 years of age in affected dogs. Two distinct phenotypes are observed in affected Glen of Imaal Terriers. The most common phenotype is subtle but generalized tapetal hyperreflectivity and retinal vascular attenuation that progresses over 1 - 2 years after initial examination. The less common phenotype is a focal mid-temporal (area centralis) area of distinct tapetal hyperreflectivity without generalized retinal disease. This lesion may remain unchanged for over a year but will progress to generalized retinal atrophy by 2 - 4 years after initial examination. ERG dysfunction can be observed as early as 15 weeks of age. The disorder is caused by a mutation present in the *ADAM9* gene. A DNA test is available that will unequivocally identify normal, affected, and carrier dogs. The test is accurate only for this mutation and will not identify other forms of PRA.

### References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Kijas JW, Zanger B, Miller B, et al. Cloning of the canine ABCA4 gene and evaluation in canine cone-rod dystrophies and progressive retinal atrophies. *Mol Vis.* 2004;10:223-232.
3. Goldstein O, Mezey JG, Boyko AR, et al. An *ADAM9* mutation in canine cone-rod dystrophy 3 establishes homology with human cone-rod dystrophy 9. *Mol Vis.* 2010;16:1549-1569.
4. Kropatsch R, Petrasch-Parwez E, Seelow D, et al. Generalized progressive retinal atrophy in the Irish Glen of Imaal Terrier is associated with a deletion in the *ADAM9* gene. *Mol Cell Probes.* 2010;24:357-363.

# OCULAR DISORDERS REPORT GLEN OF IMAAL TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>606 |      | 2016-2020<br>189 |      |
|---------------------|--|------------------|------|------------------|------|
|                     |  | #                | %    | #                | %    |
| <b>GLOBE</b>        |  |                  |      |                  |      |
| 0.110               | microphthalmia   | 1                | 0.2% | 0                |      |
| <b>EYELIDS</b>      |  |                  |      |                  |      |
| 21.000              | entropion, unspecified                                       | 2                | 0.3% | 0                |      |
| 25.110              | distichiasis   | 21               | 3.5% | 9                | 4.8% |
| <b>NASOLACRIMAL</b> |  |                  |      |                  |      |
| 32.110              | imperforate lower nasolacrimal punctum                       | 0                |      | 1                | 0.5% |
| <b>CORNEA</b>       |  |                  |      |                  |      |
| 70.220              | pigmentary keratitis   | 0                |      | 1                | 0.5% |
| <b>UVEA</b>         |  |                  |      |                  |      |
| 93.720              | persistent pupillary membranes, iris to lens                 | 1                | 0.2% | 0                |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands | 0                |      | 2                | 1.1% |
| 93.999              | uveal cysts  | 1                | 0.2% | 1                | 0.5% |
| 97.150              | chorioretinal coloboma, congenital                           | 0                |      | 1                | 0.5% |
| <b>LENS</b>         |  |                  |      |                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown         | 55               | 9.1% | 7                | 3.7% |
| 100.301             | punctate cataract, anterior cortex                           | 4                | 0.7% | 1                | 0.5% |
| 100.302             | punctate cataract, posterior cortex                          | 1                | 0.2% | 1                | 0.5% |
| 100.303             | punctate cataract, equatorial cortex                         | 4                | 0.7% | 2                | 1.1% |
| 100.305             | punctate cataract, posterior sutures                         | 0                |      | 1                | 0.5% |
| 100.306             | punctate cataract, nucleus                                   | 2                | 0.3% | 0                |      |
| 100.307             | punctate cataract, capsular                                  | 3                | 0.5% | 1                | 0.5% |
| 100.311             | incipient cataract, anterior cortex                          | 3                | 0.5% | 3                | 1.6% |
| 100.312             | incipient cataract, posterior cortex                         | 0                |      | 1                | 0.5% |
| 100.313             | incipient cataract, equatorial cortex                        | 5                | 0.8% | 1                | 0.5% |
| 100.314             | incipient cataract, anterior sutures                         | 1                | 0.2% | 0                |      |
| 100.315             | incipient cataract, posterior sutures                        | 2                | 0.3% | 0                |      |
| 100.316             | incipient cataract, nucleus                                  | 1                | 0.2% | 0                |      |
| 100.321             | incomplete cataract, anterior cortex                         | 1                | 0.2% | 0                |      |
| 100.322             | incomplete cataract, posterior cortex                        | 1                | 0.2% | 0                |      |
| 100.330             | generalized/complete cataract                                | 1                | 0.2% | 0                |      |
| 100.345             | <i>significant cataracts (summary)</i>                       | 29               | 4.8% | 11               | 5.8% |
| 100.375             | <i>subluxation/luxation, unspecified</i>                     | 3                | 0.5% | 0                |      |
| <b>VITREOUS</b>     |  |                  |      |                  |      |
| 110.120             | persistent hyaloid artery/remnant                            | 1                | 0.2% | 1                | 0.5% |
| 110.320             | vitreal degeneration   | 2                | 0.3% | 0                |      |
| <b>RETINA</b>       |  |                  |      |                  |      |
| 120.170             | retinal dysplasia, folds                                     | 7                | 1.2% | 0                |      |
| 120.180             | retinal dysplasia, geographic                                | 4                | 0.7% | 0                |      |
| 120.310             | generalized progressive retinal atrophy (PRA)                | 23               | 3.8% | 1                | 0.5% |
| 120.960             | retinopathy  | 1                | 0.2% | 0                |      |



|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>OPTIC NERVE</b>  |           |           |
| 130.120 optic nerve hypoplasia                            | 1 0.2%    | 0         |
| 130.150 optic disc coloboma                               | 4 0.7%    | 1 0.5%    |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 12 2.0%   | 0         |
| 900.100 other, not inherited                              | 12 2.0%   | 2 1.1%    |
| 900.110 other. suspect not inherited/significance unknown | 26 4.3%   | 7 3.7%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 488 80.5% | 156 82.5% |

## GOLDEN RETRIEVER

|    | DISORDER   | INHERITANCE                | REFERENCE | BREEDING<br>ADVICE                           | GENETIC TESTS<br>AVAILABLE            |
|----|--|----------------------------|-----------|--|---------------------------------------|
| A. | Microphthalmos   | Autosomal<br>recessive     | 2         | NO   | Mutation in the<br><i>SIX6</i> gene   |
| B. | Entropion  | Not defined                | 1         | Breeder option                               |                                       |
| C. | Distichiasis   | Not defined                | 1         | Breeder option                               |                                       |
| D. | Corneal dystrophy<br>- epithelial/stromal  | Not defined                | 1         | Breeder option                               |                                       |
| E. | Uveal cysts  | Not defined                | 1, 3-5    | Breeder option                               |                                       |
| F. | Pigmentary uveitis   | Not defined                | 1, 3-6    | NO   |                                       |
| G. | Persistent pupillary<br>membranes<br>- iris to iris<br>- lens pigment foci/no<br>strands | Not defined<br>Not defined | 1<br>7    | Breeder option<br>Passes with no<br>notation |                                       |
| H. | Cataract   | Not defined                | 1, 7-12   | NO   |                                       |
| I. | Y-suture tip opacity   | Not defined                | 1         | Breeder option                               |                                       |
| J. | Persistent hyaloid<br>artery   | Not defined                | 1         | Breeder option                               |                                       |
| K. | Vitreous<br>degeneration   | Not defined                | 1         | Breeder option                               |                                       |
| L. | Retinal atrophy<br><i>prcd</i>   | Autosomal<br>recessive     | 1, 12-15  | NO   | Mutation in the <i>prcd</i><br>gene   |
|    | <i>PRA1</i>  | Autosomal<br>recessive     | 13        | NO   | Mutation in the<br><i>SLC4A3</i> gene |
|    | <i>PRA2</i>  | Autosomal<br>recessive     | 3         | NO   | Mutation in the<br><i>TTC8</i> gene   |

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|    | <b>DISORDER</b>                   | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING<br/>ADVICE</b> | <b>GENETIC TESTS<br/>AVAILABLE</b> |
|----|-----------------------------------|--------------------|------------------|----------------------------|------------------------------------|
| M. | Retinal dysplasia<br>- folds      | Not defined        | 1, 16            | Breeder option             |                                    |
| N. | Retinal dysplasia<br>- geographic | Not defined        | 1, 16            | NO                         |                                    |
| O. | Limbal melanoma                   | Not defined        | 17               | NO                         |                                    |

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## **Description and Comments**

### **A. Microphthalmos**

A congenital anomaly in which the globe is abnormally small. Commonly associated with multiple ocular malformations and when severe, may affect vision.

### **B. Entropion**

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. Selection should be directed against entropion and toward a head conformation that reduces or eliminates the likelihood of the defect.

### **C. Distichiasis**

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### **D. Corneal dystrophy - epithelial/stromal**

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

### **E. Uveal cysts**

Fluid filled sacs arising from the posterior surface of the iris, to which they may remain attached or break free and float into the anterior chamber. Usually occur in mature dogs.

This disorder may be observed in any breed but retriever breeds tend to be predisposed. There is usually no effect on vision unless the cysts are heavily clustered and impinge

on the pupillary area. Less frequently, the cysts may rupture and adhere to the cornea or anterior lens capsule. Multiple cysts may occlude the iridocorneal angle and cause glaucoma.

F. Pigmentary uveitis

A unique uveitis observed in the Golden Retriever that is not associated with other ocular or systemic disorders. Adhesions develop between iris and lens and the peripheral iris and cornea. Pigment dispersion (exfoliation) occurs across the anterior lens capsule from the pigmented cells of the posterior iris. Other complications include secondary cataract and obstructive glaucoma. Onset is usually between 5-10 years of age.

G. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

H. Cataract

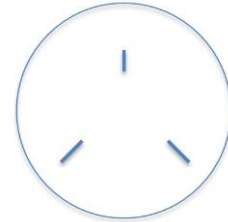
A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

The most common cataract reported in the Golden Retriever is a posterior polar (posterior cortical) cataract. These are generally bilateral, although an occasional unilateral affliction may be observed. These focal opacities will occasionally remain stationary. These cataracts are usually observed between 9 months and 3 years of age. A more generalized cataract is also observed in this breed and is not always associated with the previously mentioned polar cataract. There are also cataract changes involving the Y sutures which may or may not progress.

The existence of cataracts in the Golden Retriever, often with limited clinical significance, presents problems with breeder recognition as the majority of these dogs do not evidence visual impairment. It is strongly recommended that all Golden Retrievers that are used in breeding programs be examined annually as cataract changes have been observed in multiple locations of the lens and variable age of onset.

I. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

J. Persistent hyaloid artery (PHA)

Congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small patent vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

K. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

L. Retinal atrophy

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that one form of PRA in the Golden Retriever is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is

recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

In addition, two other known mutations that cause PRA are present in the breed. Golden Retriever PRA 1 (GR PRA1) is an autosomal recessive trait and is the predominant form in European lines of Golden Retrievers. Golden Retriever PRA 2 (GR PRA2) has also been identified within the breed. Therefore three different DNA tests are available. However these tests will only detect these three mutations.

M. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

N. Retinal dysplasia – geographic

Abnormal development of the retina present at birth. Any irregularly shaped area of abnormal retinal development containing both areas of thinning and areas of elevation representing folds and retinal disorganization.

O. Limbal melanoma

Most limbal melanomas are really epibulbar melanocytomas, but there is a possibility of an extension of an intraocular melanoma extending outward and presenting as a limbal melanoma. An epibulbar melanocytoma originates from the superficial pigment lining the limbus and the lesion may eventually extend into the eye. Metastasis has not been documented and the mass is characterized by large epithelioid cells. The lesion presents as a subconjunctival smooth mass most commonly in the dorsolateral limbal region and extends later into the cornea and posterior on the sclera. Breed predispositions have been noted in the German Shepherd Dog, and Labrador and Golden Retrievers.

**Historical Note:**

Central progressive retinal atrophy was previously a condition listed for this breed. However as the condition is no longer identified in the breed, the condition has been removed. Central progressive retinal atrophy was a progressive retinal degeneration in which photoreceptor death occurred secondary to disease of the underlying pigment epithelium. Progression was slow and some animals never lost vision. CPRA occurred in England, but was uncommon elsewhere.

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# OCULAR DISORDERS REPORT GOLDEN RETRIEVER

| Diagnostic Name   | TOTAL DOGS EXAMINED |  | 1991-2015 |       | 2016-2020 |      |
|---|---------------------|--|-----------|-------|-----------|------|
|   |                     |  | #         | %     | #         | %    |
| <b>GLOBE</b>  |                     |  | 156,875   |       | 43,394    |      |
| 0.110 microphthalmia  |                     |  | 51        | 0.0%  | 7         | 0.0% |
| 10.000 glaucoma   |                     |  | 32        | 0.0%  | 1         | 0.0% |
| <b>EYELIDS</b>  |                     |  |           |       |           |      |
| 20.110 eyelid dermoid   |                     |  | 3         | 0.0%  | 0         |      |
| 20.140 ectopic cilia  |                     |  | 53        | 0.0%  | 5         | 0.0% |
| 20.160 macropalpebral fissure   |                     |  | 22        | 0.0%  | 0         |      |
| 21.000 entropion, unspecified   |                     |  | 366       | 0.2%  | 76        | 0.2% |
| 22.000 ectropion, unspecified   |                     |  | 103       | 0.1%  | 9         | 0.0% |
| 25.110 distichiasis   |                     |  | 16,913    | 10.8% | 3,798     | 8.8% |
| <b>NASOLACRIMAL</b>   |                     |  |           |       |           |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     |  | 30        | 0.0%  | 42        | 0.1% |
| 40.910 keratoconjunctivitis sicca                                     |                     |  | 5         | 0.0%  | 2         | 0.0% |
| <b>NICTITANS</b>  |                     |  |           |       |           |      |
| 50.210 pannus of third eyelid   |                     |  | 2         | 0.0%  | 0         |      |
| 51.100 third eyelid cartilage anomaly                                 |                     |  | 16        | 0.0%  | 5         | 0.0% |
| 52.110 prolapsed gland of the third eyelid                            |                     |  | 41        | 0.0%  | 1         | 0.0% |
| <b>CORNEA</b>   |                     |  |           |       |           |      |
| 70.210 corneal pannus   |                     |  | 11        | 0.0%  | 0         |      |
| 70.220 pigmentary keratitis   |                     |  | 15        | 0.0%  | 13        | 0.0% |
| 70.700 corneal dystrophy  |                     |  | 632       | 0.4%  | 207       | 0.5% |
| 70.730 corneal endothelial degeneration                               |                     |  | 37        | 0.0%  | 5         | 0.0% |
| <b>UVEA</b>   |                     |  |           |       |           |      |
| 90.200 uveitis  |                     |  | 0         |       | 7         | 0.0% |
| 90.250 pigmentary uveitis   |                     |  | 967       | 0.6%  | 503       | 1.2% |
| 93.110 iris hypoplasia  |                     |  | 3         | 0.0%  | 3         | 0.0% |
| 93.140 corneal endothelial pigment without PPM                        |                     |  | 17        | 0.0%  | 0         |      |
| 93.150 iris coloboma  |                     |  | 20        | 0.0%  | 1         | 0.0% |
| 93.180 liris sphincter dysplasia                                      |                     |  | 1         | 0.0%  | 1         | 0.0% |
| 93.710 persistent pupillary membranes, iris to iris                   |                     |  | 3,378     | 2.2%  | 1,279     | 2.9% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     |  | 115       | 0.1%  | 20        | 0.0% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     |  | 83        | 0.1%  | 10        | 0.0% |
| 93.740 persistent pupillary membranes, iris sheets                    |                     |  | 111       | 0.1%  | 1         | 0.0% |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     |  | 416       | 0.3%  | 605       | 1.4% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     |  | 44        | 0.0%  | 20        | 0.0% |
| 93.810 uveal melanoma   |                     |  | 22        | 0.0%  | 17        | 0.0% |
| 93.999 uveal cysts  |                     |  | 7,785     | 5.0%  | 3,531     | 8.1% |
| 97.150 chorioretinal coloboma, congenital                             |                     |  | 1         | 0.0%  | 2         | 0.0% |
| <b>LENS</b>   |                     |  |           |       |           |      |
| 100.200 cataract, unspecified   |                     |  | 952       | 0.6%  | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     |  | 9,430     | 6.0%  | 3,104     | 7.2% |
| 100.300 punctate cataract, unspecified                                |                     |  | 0         |       | 1         | 0.0% |
| 100.301 punctate cataract, anterior cortex                            |                     |  | 708       | 0.5%  | 313       | 0.7% |
| 100.302 punctate cataract, posterior cortex                           |                     |  | 2,239     | 1.4%  | 488       | 1.1% |

| <b>LENS CONTINUED</b>                                 | <b>1991-2015</b> |      | <b>2016-2020</b> |      |
|---|------------------|------|------------------|------|
| 100.303 punctate cataract, equatorial cortex          | 450              | 0.3% | 183              | 0.4% |
| 100.304 punctate cataract, anterior sutures           | 102              | 0.1% | 36               | 0.1% |
| 100.305 punctate cataract, posterior sutures          | 781              | 0.5% | 168              | 0.4% |
| 100.306 punctate cataract, nucleus                    | 209              | 0.1% | 120              | 0.3% |
| 100.307 punctate cataract, capsular                   | 317              | 0.2% | 169              | 0.4% |
| 100.311 incipient cataract, anterior cortex           | 863              | 0.6% | 266              | 0.6% |
| 100.312 incipient cataract, posterior cortex          | 3,117            | 2.0% | 732              | 1.7% |
| 100.313 incipient cataract, equatorial cortex         | 932              | 0.6% | 315              | 0.7% |
| 100.314 incipient cataract, anterior sutures          | 65               | 0.0% | 18               | 0.0% |
| 100.315 incipient cataract, posterior sutures         | 730              | 0.5% | 127              | 0.3% |
| 100.316 incipient cataract, nucleus                   | 328              | 0.2% | 157              | 0.4% |
| 100.317 incipient cataract, capsular                  | 276              | 0.2% | 139              | 0.3% |
| 100.320 incomplete cataract, unspecified              | 0                |      | 3                | 0.0% |
| 100.321 incomplete cataract, anterior cortex          | 31               | 0.0% | 55               | 0.1% |
| 100.322 incomplete cataract, posterior cortex         | 68               | 0.0% | 113              | 0.3% |
| 100.323 incomplete cataract, equatorial cortex        | 13               | 0.0% | 43               | 0.1% |
| 100.324 incomplete cataract, anterior sutures         | 1                | 0.0% | 1                | 0.0% |
| 100.325 incomplete cataract, posterior sutures        | 6                | 0.0% | 21               | 0.0% |
| 100.326 incomplete cataract, nucleus                  | 13               | 0.0% | 37               | 0.1% |
| 100.327 incomplete cataract, capsular                 | 7                | 0.0% | 20               | 0.0% |
| 100.328 y-suture tip opacities                        | 68               | 0.0% | 147              | 0.3% |
| 100.330 generalized/complete cataract                 | 349              | 0.2% | 29               | 0.1% |
| 100.340 resorbing/hypermature cataract                | 2                | 0.0% | 9                | 0.0% |
| 100.345 <i>significant cataracts (summary)</i>        | 12,627           | 8.0% | 3,710            | 8.5% |
| 100.375 <i>subluxation/luxation, unspecified</i>      | 31               | 0.0% | 3                | 0.0% |
| <b>VITREOUS</b>                                       |                  |      |                  |      |
| 110.120 persistent hyaloid artery/remnant             | 150              | 0.1% | 89               | 0.2% |
| 110.135 PHPV/PTVL                                     | 37               | 0.0% | 9                | 0.0% |
| 110.200 vitreous degeneration-anterior chamber        | 4                | 0.0% | 6                | 0.0% |
| 110.320 vitreal degeneration                          | 272              | 0.2% | 87               | 0.2% |
| <b>FUNDUS</b>   |                  |      |                  |      |
| 97.110 choroidal hypoplasia                           | 9                | 0.0% | 0                |      |
| 97.120 coloboma                                       | 8                | 0.0% | 0                |      |
| <b>RETINA</b>   |                  |      |                  |      |
| 120.170 retinal dysplasia, folds                      | 1,962            | 1.3% | 465              | 1.1% |
| 120.180 retinal dysplasia, geographic                 | 786              | 0.5% | 222              | 0.5% |
| 120.190 retinal dysplasia, detached                   | 38               | 0.0% | 5                | 0.0% |
| 120.310 generalized progressive retinal atrophy (PRA) | 173              | 0.1% | 12               | 0.0% |
| 120.400 retinal hemorrhage                            | 18               | 0.0% | 0                |      |
| 120.910 retinal detachment without dialysis           | 28               | 0.0% | 0                |      |
| 120.920 retinal detachment with dialysis              | 3                | 0.0% | 3                | 0.0% |
| 120.960 retinopathy                                   | 33               | 0.0% | 45               | 0.1% |
| <b>OPTIC NERVE</b>                                    |                  |      |                  |      |
| 130.110 micropapilla                                  | 10               | 0.0% | 12               | 0.0% |
| 130.120 optic nerve hypoplasia                        | 37               | 0.0% | 7                | 0.0% |
| 130.150 optic disc coloboma                           | 56               | 0.0% | 12               | 0.0% |

|   | 1991-2015 |       | 2016-2020 |       |
|---|-----------|-------|-----------|-------|
| <b>OTHER</b>  |           |       |           |       |
| 900.000 other, unspecified                                | 1,783     | 1.1%  | 0         |       |
| 900.100 other, not inherited                              | 3,017     | 1.9%  | 84        | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown | 2,066     | 1.3%  | 1,932     | 4.5%  |
| <b>NORMAL</b>   |           |       |           |       |
| 0.000 normal globe  | 118,528   | 75.6% | 28,185    | 65.0% |

# GORDON SETTER

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE         | GENETIC TESTS AVAILABLE             |
|----|--|---------------------|-----------|-------------------------|-------------------------------------|
| A. | Ectropion  | Not defined         | 1         | Breeder option          |                                     |
| B. | Distichiasis   | Not defined         | 1         | Breeder option          |                                     |
| C. | Persistent pupillary membranes                                 | Not defined         | 1         | Breeder option          |                                     |
|    | - iris to iris<br>- lens pigment foci/no strands               | Not defined         | 1         | Passes with no notation |                                     |
| D. | Cataract   | Not defined         | 1         | NO                      |                                     |
| E. | Persistent hyaloid artery                                      | Not defined         | 1         | Breeder option          |                                     |
| F. | Retinal atrophy<br>- rod-cone dysplasia type 4 ( <i>rcd4</i> ) | Autosomal recessive | 2         | NO                      | Mutation in the <i>C2orf71</i> gene |
| G. | Cone degeneration - achromatopsia                              | Not defined         | 3         | NO                      |                                     |
| H. | Retinal dysplasia<br>- folds                                   | Not defined         | 1         | Breeder option          |                                     |

## Description and Comments

### A. Ectropion

A conformational defect resulting in eversion of the eyelids which may cause ocular irritation. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded;

breeding discretion is advised.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Persistent hyaloid artery (PHA)

Congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small patent vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

F. Rod-cone dysplasia, type 4 (*rcd4*)

A form of PRA identified in the Gordon and Irish Setter breeds. Clinical night blindness is observed on average as late as 10 years of age and progresses to total blindness. This form of PRA has been referred to as late-onset PRA (LOPRA). The disorder is caused by a mutation present in the *C2orf71* gene. A DNA test is now available that will unequivocally identify genetically normal, affected and carrier dogs. The test is accurate only for this mutation and will not identify other forms of PRA.

G. Cone degeneration - achromatopsia

Suspected inherited retinopathy characterized by degeneration of the cone receptors and loss of vision in bright light. Age of onset is variable. Ophthalmoscopic examination is normal. The ERG abnormalities are more suggestive of a cone-rod dystrophy. The mode of inheritance and genetic mutation are not yet known.

H. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal

dysplasia is undetermined.

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# OCULAR DISORDERS REPORT GORDON SETTER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>2,217 |      | 2016-2020<br>322 |       |
|---------------------|--|--------------------|------|------------------|-------|
|                     |  | #                  | %    | #                | %     |
| <b>GLOBE</b>        |  |                    |      |                  |       |
| 0.110               | microphthalmia   | 2                  | 0.1% | 0                |       |
| <b>EYELIDS</b>      |  |                    |      |                  |       |
| 20.140              | ectopic cilia  | 1                  | 0.0% | 0                |       |
| 20.160              | macropalpebral fissure   | 9                  | 0.4% | 0                |       |
| 21.000              | entropion, unspecified   | 15                 | 0.7% | 3                | 0.9%  |
| 22.000              | ectropion, unspecified   | 52                 | 2.3% | 4                | 1.2%  |
| 25.110              | distichiasis   | 42                 | 1.9% | 4                | 1.2%  |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 0                  |      | 1                | 0.3%  |
| 40.910              | keratoconjunctivitis sicca                                     | 3                  | 0.1% | 0                |       |
| <b>NICTITANS</b>    |  |                    |      |                  |       |
| 51.100              | third eyelid cartilage anomaly                                 | 1                  | 0.0% | 0                |       |
| <b>CORNEA</b>       |  |                    |      |                  |       |
| 70.210              | corneal pannus   | 3                  | 0.1% | 0                |       |
| 70.700              | corneal dystrophy  | 8                  | 0.4% | 0                |       |
| <b>UVEA</b>         |  |                    |      |                  |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 112                | 5.1% | 40               | 12.4% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 7                  | 0.3% | 1                | 0.3%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 4                  | 0.2% | 0                |       |
| 93.740              | persistent pupillary membranes, iris sheets                    | 2                  | 0.1% | 0                |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 15                 | 0.7% | 17               | 5.3%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 4                  | 0.2% | 0                |       |
| 93.999              | uveal cysts  | 19                 | 0.9% | 2                | 0.6%  |
| <b>LENS</b>         |  |                    |      |                  |       |
| 100.200             | cataract, unspecified  | 9                  | 0.4% | 0                |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 79                 | 3.6% | 16               | 5.0%  |
| 100.301             | punctate cataract, anterior cortex                             | 5                  | 0.2% | 2                | 0.6%  |
| 100.302             | punctate cataract, posterior cortex                            | 9                  | 0.4% | 2                | 0.6%  |
| 100.303             | punctate cataract, equatorial cortex                           | 3                  | 0.1% | 0                |       |
| 100.305             | punctate cataract, posterior sutures                           | 4                  | 0.2% | 0                |       |
| 100.306             | punctate cataract, nucleus                                     | 5                  | 0.2% | 2                | 0.6%  |
| 100.307             | punctate cataract, capsular                                    | 1                  | 0.0% | 0                |       |
| 100.311             | incipient cataract, anterior cortex                            | 7                  | 0.3% | 0                |       |
| 100.312             | incipient cataract, posterior cortex                           | 14                 | 0.6% | 3                | 0.9%  |
| 100.313             | incipient cataract, equatorial cortex                          | 8                  | 0.4% | 0                |       |
| 100.315             | incipient cataract, posterior sutures                          | 1                  | 0.0% | 1                | 0.3%  |
| 100.316             | incipient cataract, nucleus                                    | 4                  | 0.2% | 0                |       |
| 100.317             | incipient cataract, capsular                                   | 4                  | 0.2% | 1                | 0.3%  |
| 100.327             | incomplete cataract, capsular                                  | 1                  | 0.0% | 0                |       |
| 100.328             | y-suture tip opacities   | 2                  | 0.1% | 0                |       |
| 100.330             | generalized/complete cataract                                  | 10                 | 0.5% | 0                |       |
| 100.345             | significant cataracts (summary)                                | 87                 | 3.9% | 11               | 3.4%  |

|   | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 12 0.5%          | 3 0.9%           |
| 110.135 PHPV/PTVL   | 5 0.2%           | 2 0.6%           |
| 110.320 vitreal degeneration                              | 5 0.2%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 32 1.4%          | 7 2.2%           |
| 120.180 retinal dysplasia, geographic                     | 4 0.2%           | 0                |
| 120.190 retinal dysplasia, detached                       | 1 0.0%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 17 0.8%          | 1 0.3%           |
| 120.910 retinal detachment without dialysis               | 2 0.1%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 8 0.4%           | 0                |
| 130.120 optic nerve hypoplasia                            | 8 0.4%           | 0                |
| 130.150 optic disc coloboma                               | 1 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 40 1.8%          | 0                |
| 900.100 other, not inherited                              | 59 2.7%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 31 1.4%          | 12 3.7%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,807 81.5%      | 230 71.4%        |



# **OCULAR DISORDERS REPORT GRAND BASSET GRIFFON VENDEEN**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the GRAND BASSET GRIFFON VENDEEN breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT GRAND BASSET GRIFFON VENDEEN

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015<br>64 |       | 2016-2020<br>67 |       |
|-----------------|--|-----------------|-------|-----------------|-------|
|                 |  | #               | %     | #               | %     |
| <b>EYELIDS</b>  |  |                 |       |                 |       |
| 25.110          | distichiasis   | 1               | 1.6%  | 0               |       |
| <b>UVEA</b>     |  |                 |       |                 |       |
| 93.710          | persistent pupillary membranes, iris to iris                   | 4               | 6.2%  | 3               | 4.5%  |
| 93.730          | persistent pupillary membranes, iris to cornea                 | 5               | 7.8%  | 2               | 3.0%  |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands   | 0               |       | 1               | 1.5%  |
| 93.760          | persistent pupillary membranes, endothelial opacity/no strands | 5               | 7.8%  | 1               | 1.5%  |
| <b>LENS</b>     |  |                 |       |                 |       |
| 100.210         | cataract. suspect not inherited/significance unknown           | 2               | 3.1%  | 6               | 9.0%  |
| 100.311         | incipient cataract, anterior cortex                            | 0               |       | 1               | 1.5%  |
| 100.317         | incipient cataract, capsular                                   | 0               |       | 1               | 1.5%  |
| 100.321         | incomplete cataract, anterior cortex                           | 0               |       | 1               | 1.5%  |
| 100.327         | incomplete cataract, capsular                                  | 1               | 1.6%  | 0               |       |
| 100.345         | <i>significant cataracts (summary)</i>                         | 1               | 1.6%  | 3               | 4.5%  |
| <b>VITREOUS</b> |  |                 |       |                 |       |
| 110.135         | PHPV/PTVL  | 1               | 1.6%  | 0               |       |
| <b>RETINA</b>   |  |                 |       |                 |       |
| 120.170         | retinal dysplasia, folds                                       | 1               | 1.6%  | 0               |       |
| 120.310         | generalized progressive retinal atrophy (PRA)                  | 1               | 1.6%  | 0               |       |
| <b>OTHER</b>    |  |                 |       |                 |       |
| 900.000         | other, unspecified   | 2               | 3.1%  | 0               |       |
| 900.100         | other, not inherited   | 1               | 1.6%  | 0               |       |
| 900.110         | other. suspect not inherited/significance unknown              | 0               |       | 2               | 3.0%  |
| <b>NORMAL</b>   |  |                 |       |                 |       |
| 0.000           | normal globe   | 46              | 71.9% | 51              | 76.1% |

# GREAT DANE

|    | DISORDER   | INHERITANCE                 | REFERENCE | BREEDING ADVICE |
|----|--|-----------------------------|-----------|-----------------|
| A. | Microphthalmia with multiple ocular defects associated with partial albinism | Presumed autosomal dominant | 1, 2      | NO              |
| B. | Glaucoma   | Not defined                 | 1, 3      | NO              |
| C. | Entropion  | Not defined                 | 1         | Breeder option  |
| D. | Ectropion  | Not defined                 | 1         | Breeder option  |
| E. | Distichiasis   | Not defined                 | 1         | Breeder option  |
| F. | Nictitans cartilage anomaly/eversion   | Not defined                 | 1         | Breeder option  |
| G. | Prolapsed gland of the third eyelid  | Not defined                 | 4         | Breeder option  |
| H. | Uveal cysts  | Not defined                 | 1, 5      | Breeder option  |
| I. | Persistent pupillary membranes<br>- iris to iris                             | Not defined                 | 1         | Breeder option  |
| J. | Cataract   | Not defined                 | 1         | NO              |

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## Description and Comments

### A. Microphthalmia with multiple ocular defects associated with partial albinism

Multiple ocular defects are seen associated with partial albinism (white or light coat color) and deafness in Great Danes. The abnormalities are thought to stem from a common developmental defect. Ocular defects are anterior segment dysgenesis, equatorial staphylomas, microphthalmia, cortical cataracts, lens luxation, spherophakia, iris coloboma, and blue irides. An autosomal dominant mode of inheritance is suspected. The hearing loss is attributable to cochlea-saccular degeneration.

### B. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis

and classification of glaucoma requires measurement of the IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine screening exam for certification.

C. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. Entropion and ectropion often occur together in this breed, associated with an abnormally large palpebral fissure.

D. Ectropion

A conformational defect resulting in eversion of the eyelids which may cause ocular irritation. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

E. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

F. Nictitans cartilage anomaly/eversion

A scroll-like curling of the cartilage of the third eyelid, usually everting the margin. This condition may occur in one or both eyes and may cause mild ocular irritation.

G. Prolapsed gland of the third eyelid

Protrusion of the tear gland associated with the third eyelid. The mode of inheritance of this disorder is unknown. The exposed gland may become irritated. Commonly referred to as "cherry eye."

Great Danes were overrepresented in a study of prolapsed gland of the third eyelid. In the study, 83% of the prolapsed glands in Great Danes occurred before 1 year of age. Great Danes were also more likely to develop bilateral prolapsed glands that occurred either simultaneously with the first prolapse or with a short time interval between prolapses.

H. Uveal cysts

Fluid filled sacs arising from the posterior surface of the iris, to which they may remain attached or break free and float into the anterior chamber. Usually occur in mature dogs. In the Great Dane, pigmented cysts may also arise from pigmented epithelial cells of the ciliary body. Ciliary body cysts may predispose to glaucoma in the Great Dane.

I. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

J. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gwin RM, Wyman M, Lim DJ, et al. Multiple ocular defects associated with partial albinism and deafness in the dog. *J Am Anim Hosp Assoc.* 1981;17:401-408.
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# OCULAR DISORDERS REPORT GREAT DANE

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>6,711 |      | 2016-2020<br>2,648 |      |
|---------------------|--|--------------------|------|--------------------|------|
|                     |  | #                  | %    | #                  | %    |
| <b>GLOBE</b>        |  |                    |      |                    |      |
| 0.110               | microphthalmia   | 24                 | 0.4% | 3                  | 0.1% |
| 10.000              | glaucoma   | 2                  | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                    |      |                    |      |
| 20.160              | macropalpebral fissure   | 124                | 1.8% | 0                  |      |
| 21.000              | entropion, unspecified   | 168                | 2.5% | 108                | 4.1% |
| 22.000              | ectropion, unspecified   | 263                | 3.9% | 111                | 4.2% |
| 25.110              | distichiasis   | 366                | 5.5% | 135                | 5.1% |
| <b>NASOLACRIMAL</b> |  |                    |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 6                  | 0.1% | 1                  | 0.0% |
| 40.910              | keratoconjunctivitis sicca                                     | 1                  | 0.0% | 1                  | 0.0% |
| <b>NICTITANS</b>    |  |                    |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 135                | 2.0% | 83                 | 3.1% |
| 52.110              | prolapsed gland of the third eyelid                            | 13                 | 0.2% | 11                 | 0.4% |
| <b>CORNEA</b>       |  |                    |      |                    |      |
| 70.210              | corneal pannus   | 2                  | 0.0% | 0                  |      |
| 70.220              | pigmentary keratitis   | 4                  | 0.1% | 2                  | 0.1% |
| 70.700              | corneal dystrophy  | 28                 | 0.4% | 8                  | 0.3% |
| <b>UVEA</b>         |  |                    |      |                    |      |
| 90.250              | pigmentary uveitis   | 1                  | 0.0% | 1                  | 0.0% |
| 93.110              | iris hypoplasia  | 6                  | 0.1% | 6                  | 0.2% |
| 93.140              | corneal endothelial pigment without PPM                        | 2                  | 0.0% | 0                  |      |
| 93.150              | iris coloboma  | 18                 | 0.3% | 4                  | 0.2% |
| 93.180              | iris sphincter dysplasia                                       | 0                  |      | 2                  | 0.1% |
| 93.710              | persistent pupillary membranes, iris to iris                   | 69                 | 1.0% | 28                 | 1.1% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 15                 | 0.2% | 3                  | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 8                  | 0.1% | 2                  | 0.1% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 4                  | 0.1% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 18                 | 0.3% | 22                 | 0.8% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 1                  | 0.0% | 2                  | 0.1% |
| 93.810              | uveal melanoma   | 3                  | 0.0% | 2                  | 0.1% |
| 93.999              | uveal cysts  | 85                 | 1.3% | 59                 | 2.2% |
| <b>LENS</b>         |  |                    |      |                    |      |
| 100.110             | microphakia, congenital  | 0                  |      | 1                  | 0.0% |
| 100.200             | cataract, unspecified  | 15                 | 0.2% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 241                | 3.6% | 85                 | 3.2% |
| 100.301             | punctate cataract, anterior cortex                             | 25                 | 0.4% | 12                 | 0.5% |
| 100.302             | punctate cataract, posterior cortex                            | 66                 | 1.0% | 27                 | 1.0% |
| 100.303             | punctate cataract, equatorial cortex                           | 17                 | 0.3% | 3                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 4                  | 0.1% | 1                  | 0.0% |
| 100.305             | punctate cataract, posterior sutures                           | 27                 | 0.4% | 4                  | 0.2% |
| 100.306             | punctate cataract, nucleus                                     | 13                 | 0.2% | 2                  | 0.1% |
| 100.307             | punctate cataract, capsular                                    | 12                 | 0.2% | 10                 | 0.4% |
| 100.311             | incipient cataract, anterior cortex                            | 63                 | 0.9% | 22                 | 0.8% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.312 incipient cataract, posterior cortex              | 153              | 2.3%  | 34               | 1.3%  |
| 100.313 incipient cataract, equatorial cortex             | 43               | 0.6%  | 14               | 0.5%  |
| 100.314 incipient cataract, anterior sutures              | 6                | 0.1%  | 0                |       |
| 100.315 incipient cataract, posterior sutures             | 19               | 0.3%  | 8                | 0.3%  |
| 100.316 incipient cataract, nucleus                       | 33               | 0.5%  | 6                | 0.2%  |
| 100.317 incipient cataract, capsular                      | 21               | 0.3%  | 10               | 0.4%  |
| 100.321 incomplete cataract, anterior cortex              | 6                | 0.1%  | 4                | 0.2%  |
| 100.322 incomplete cataract, posterior cortex             | 6                | 0.1%  | 10               | 0.4%  |
| 100.323 incomplete cataract, equatorial cortex            | 0                |       | 3                | 0.1%  |
| 100.325 incomplete cataract, posterior sutures            | 0                |       | 1                | 0.0%  |
| 100.326 incomplete cataract, nucleus                      | 2                | 0.0%  | 2                | 0.1%  |
| 100.327 incomplete cataract, capsular                     | 2                | 0.0%  | 0                |       |
| 100.328 y-suture tip opacities                            | 5                | 0.1%  | 3                | 0.1%  |
| 100.330 generalized/complete cataract                     | 52               | 0.8%  | 3                | 0.1%  |
| 100.345 <i>significant cataracts (summary)</i>            | 590              | 8.8%  | 179              | 6.8%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 9                | 0.1%  | 5                | 0.2%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 11               | 0.2%  | 13               | 0.5%  |
| 110.135 PHPV/PTVL   | 15               | 0.2%  | 2                | 0.1%  |
| 110.200 vitreous degeneration-anterior chamber            | 3                | 0.0%  | 8                | 0.3%  |
| 110.320 vitreal degeneration                              | 36               | 0.5%  | 8                | 0.3%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 1                | 0.0%  | 3                | 0.1%  |
| 97.120 coloboma   | 2                | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 23               | 0.3%  | 5                | 0.2%  |
| 120.180 retinal dysplasia, geographic                     | 3                | 0.0%  | 0                |       |
| 120.190 retinal dysplasia, detached                       | 2                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 7                | 0.1%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 1                | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 1                | 0.0%  | 0                |       |
| 120.960 retinopathy                                       | 2                | 0.0%  | 0                |       |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 1                | 0.0%  | 2                | 0.1%  |
| 130.120 optic nerve hypoplasia                            | 3                | 0.0%  | 1                | 0.0%  |
| 130.150 optic disc coloboma                               | 2                | 0.0%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 60               | 0.9%  | 0                |       |
| 900.100 other, not inherited                              | 137              | 2.0%  | 21               | 0.8%  |
| 900.110 other. suspect not inherited/significance unknown | 89               | 1.3%  | 107              | 4.0%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 5,276            | 78.6% | 1,878            | 70.9% |

# GREAT PYRENEES

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|--|---------------------|-----------|-----------------|-----------------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option  |                                   |
| B. | Cataract   | Not defined         | 1         | NO              |                                   |
| C. | Multifocal retinopathy<br>- <i>cmr1</i>          | Autosomal recessive | 2-4       | Breeder option  | Mutation in the <i>BEST1</i> gene |

## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (*cmr1*) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous sub-retinal fluid, or accumulation of sub-retinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities



despite their abnormal appearing retinas.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff. A DNA test is available.

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# OCULAR DISORDERS REPORT GREAT PYRENEES

| Diagnostic Name   | TOTAL DOGS EXAMINED |  | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|--|-----------|-------|-----------|-------|
|   |                     |  | 1,224     |       | 141       |       |
|   |                     |  | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |  |           |       |           |       |
| 0.110 microphthalmia  |                     |  | 2         | 0.2%  | 0         |       |
| <b>EYELIDS</b>  |                     |  |           |       |           |       |
| 20.160 macropalpebral fissure   |                     |  | 3         | 0.2%  | 0         |       |
| 21.000 entropion, unspecified   |                     |  | 15        | 1.2%  | 0         |       |
| 22.000 ectropion, unspecified   |                     |  | 3         | 0.2%  | 0         |       |
| 25.110 distichiasis   |                     |  | 16        | 1.3%  | 0         |       |
| <b>NASOLACRIMAL</b>   |                     |  |           |       |           |       |
| 32.110 imperforate lower nasolacrimal punctum                         |                     |  | 1         | 0.1%  | 0         |       |
| <b>CORNEA</b>   |                     |  |           |       |           |       |
| 70.210 corneal pannus   |                     |  | 1         | 0.1%  | 0         |       |
| 70.700 corneal dystrophy  |                     |  | 14        | 1.1%  | 3         | 2.1%  |
| 70.730 corneal endothelial degeneration                               |                     |  | 3         | 0.2%  | 0         |       |
| <b>UVEA</b>   |                     |  |           |       |           |       |
| 93.110 iris hypoplasia  |                     |  | 1         | 0.1%  | 0         |       |
| 93.150 iris coloboma  |                     |  | 1         | 0.1%  | 0         |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     |  | 305       | 24.9% | 36        | 25.5% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     |  | 11        | 0.9%  | 2         | 1.4%  |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     |  | 7         | 0.6%  | 0         |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     |  | 0         |       | 1         | 0.7%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     |  | 2         | 0.2%  | 0         |       |
| 93.810 uveal melanoma   |                     |  | 1         | 0.1%  | 0         |       |
| 93.999 uveal cysts  |                     |  | 6         | 0.5%  | 5         | 3.5%  |
| <b>LENS</b>   |                     |  |           |       |           |       |
| 100.200 cataract, unspecified   |                     |  | 3         | 0.2%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     |  | 51        | 4.2%  | 5         | 3.5%  |
| 100.301 punctate cataract, anterior cortex                            |                     |  | 11        | 0.9%  | 1         | 0.7%  |
| 100.302 punctate cataract, posterior cortex                           |                     |  | 12        | 1.0%  | 1         | 0.7%  |
| 100.303 punctate cataract, equatorial cortex                          |                     |  | 6         | 0.5%  | 0         |       |
| 100.304 punctate cataract, anterior sutures                           |                     |  | 3         | 0.2%  | 0         |       |
| 100.305 punctate cataract, posterior sutures                          |                     |  | 3         | 0.2%  | 0         |       |
| 100.306 punctate cataract, nucleus                                    |                     |  | 3         | 0.2%  | 1         | 0.7%  |
| 100.307 punctate cataract, capsular                                   |                     |  | 1         | 0.1%  | 0         |       |
| 100.311 incipient cataract, anterior cortex                           |                     |  | 22        | 1.8%  | 3         | 2.1%  |
| 100.312 incipient cataract, posterior cortex                          |                     |  | 18        | 1.5%  | 3         | 2.1%  |
| 100.313 incipient cataract, equatorial cortex                         |                     |  | 20        | 1.6%  | 3         | 2.1%  |
| 100.315 incipient cataract, posterior sutures                         |                     |  | 5         | 0.4%  | 0         |       |
| 100.316 incipient cataract, nucleus                                   |                     |  | 1         | 0.1%  | 0         |       |
| 100.317 incipient cataract, capsular                                  |                     |  | 4         | 0.3%  | 0         |       |
| 100.321 incomplete cataract, anterior cortex                          |                     |  | 0         |       | 1         | 0.7%  |
| 100.322 incomplete cataract, posterior cortex                         |                     |  | 0         |       | 1         | 0.7%  |
| 100.323 incomplete cataract, equatorial cortex                        |                     |  | 0         |       | 1         | 0.7%  |
| 100.325 incomplete cataract, posterior sutures                        |                     |  | 0         |       | 1         | 0.7%  |
| 100.330 generalized/complete cataract                                 |                     |  | 5         | 0.4%  | 0         |       |
| 100.345 significant cataracts (summary)                               |                     |  | 117       | 9.6%  | 16        | 11.3% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.375 subluxation/luxation, unspecified                 | 1 0.1%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.135 PHPV/PTVL   | 1 0.1%           | 0                |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 2 0.2%           | 0                |
| 97.120 coloboma   | 1 0.1%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 9 0.7%           | 0                |
| 120.180 retinal dysplasia, geographic                     | 15 1.2%          | 1 0.7%           |
| 120.190 retinal dysplasia, detached                       | 2 0.2%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 5 0.4%           | 0                |
| 120.910 retinal detachment without dialysis               | 4 0.3%           | 0                |
| 120.960 retinopathy                                       | 3 0.2%           | 8 5.7%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 6 0.5%           | 0                |
| 130.120 optic nerve hypoplasia                            | 5 0.4%           | 0                |
| 130.150 optic disc coloboma                               | 2 0.2%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 7 0.6%           | 0                |
| 900.100 other, not inherited                              | 35 2.9%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 16 1.3%          | 3 2.1%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 813 66.4%        | 81 57.4%         |

# GREATER SWISS MOUNTAIN DOG

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Distichiasis                                     | Not defined | 1         | Breeder option  |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| C. | Cataract   | Not defined | 1         | NO              |
| D. | Persistent hyaloid artery                        | Not defined | 1         | Breeder option  |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### D. Persistent hyaloid artery (PHA)

Congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small patent vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Greater Swiss Mountain Dog breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT GREATER SWISS MOUNTAIN DOG

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>3,022 |       | 2016-2020<br>656 |       |
|---|---------------------|--------------------|-------|------------------|-------|
|   |                     | #                  | %     | #                | %     |
| <b>GLOBE</b>  |                     |                    |       |                  |       |
| 0.110 microphthalmia  |                     | 1                  | 0.0%  | 0                |       |
| <b>EYELIDS</b>  |                     |                    |       |                  |       |
| 20.140 ectopic cilia  |                     | 1                  | 0.0%  | 0                |       |
| 20.160 macropalpebral fissure   |                     | 1                  | 0.0%  | 0                |       |
| 21.000 entropion, unspecified   |                     | 20                 | 0.7%  | 2                | 0.3%  |
| 22.000 ectropion, unspecified   |                     | 3                  | 0.1%  | 0                |       |
| 25.110 distichiasis   |                     | 1,009              | 33.4% | 152              | 23.2% |
| <b>NICTITANS</b>  |                     |                    |       |                  |       |
| 51.100 third eyelid cartilage anomaly                                 |                     | 5                  | 0.2%  | 0                |       |
| <b>CORNEA</b>   |                     |                    |       |                  |       |
| 70.210 corneal pannus   |                     | 2                  | 0.1%  | 0                |       |
| 70.220 pigmentary keratitis   |                     | 1                  | 0.0%  | 0                |       |
| 70.700 corneal dystrophy  |                     | 13                 | 0.4%  | 2                | 0.3%  |
| 70.730 corneal endothelial degeneration                               |                     | 1                  | 0.0%  | 0                |       |
| <b>UVEA</b>   |                     |                    |       |                  |       |
| 93.150 iris coloboma  |                     | 1                  | 0.0%  | 0                |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 101                | 3.3%  | 22               | 3.4%  |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 6                  | 0.2%  | 0                |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 5                  | 0.2%  | 1                | 0.2%  |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 5                  | 0.2%  | 0                |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 1                  | 0.0%  | 1                | 0.2%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1                  | 0.0%  | 0                |       |
| 93.999 uveal cysts  |                     | 5                  | 0.2%  | 0                |       |
| <b>LENS</b>   |                     |                    |       |                  |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 267                | 8.8%  | 37               | 5.6%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 55                 | 1.8%  | 7                | 1.1%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 51                 | 1.7%  | 12               | 1.8%  |
| 100.303 punctate cataract, equatorial cortex                          |                     | 26                 | 0.9%  | 2                | 0.3%  |
| 100.304 punctate cataract, anterior sutures                           |                     | 2                  | 0.1%  | 1                | 0.2%  |
| 100.305 punctate cataract, posterior sutures                          |                     | 11                 | 0.4%  | 3                | 0.5%  |
| 100.306 punctate cataract, nucleus                                    |                     | 5                  | 0.2%  | 0                |       |
| 100.307 punctate cataract, capsular                                   |                     | 10                 | 0.3%  | 8                | 1.2%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 56                 | 1.9%  | 12               | 1.8%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 86                 | 2.8%  | 23               | 3.5%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 66                 | 2.2%  | 12               | 1.8%  |
| 100.314 incipient cataract, anterior sutures                          |                     | 2                  | 0.1%  | 0                |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 12                 | 0.4%  | 1                | 0.2%  |
| 100.316 incipient cataract, nucleus                                   |                     | 8                  | 0.3%  | 1                | 0.2%  |
| 100.317 incipient cataract, capsular                                  |                     | 11                 | 0.4%  | 2                | 0.3%  |
| 100.321 incomplete cataract, anterior cortex                          |                     | 3                  | 0.1%  | 1                | 0.2%  |
| 100.322 incomplete cataract, posterior cortex                         |                     | 1                  | 0.0%  | 3                | 0.5%  |
| 100.323 incomplete cataract, equatorial cortex                        |                     | 0                  |       | 4                | 0.6%  |
| 100.326 incomplete cataract, nucleus                                  |                     | 1                  | 0.0%  | 0                |       |
| 100.327 incomplete cataract, capsular                                 |                     | 0                  |       | 1                | 0.2%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.328 y-suture tip opacities                            | 1 0.0%           | 2 0.3%           |
| 100.330 generalized/complete cataract                     | 7 0.2%           | 0                |
| 100.345 significant cataracts (summary)                   | 414 13.7%        | 95 14.5%         |
| 100.375 subluxation/luxation, unspecified                 | 3 0.1%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 9 0.3%           | 7 1.1%           |
| 110.135 PHPV/PTVL   | 4 0.1%           | 0                |
| 110.320 vitreal degeneration                              | 3 0.1%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 16 0.5%          | 3 0.5%           |
| 120.180 retinal dysplasia, geographic                     | 7 0.2%           | 0                |
| 120.190 retinal dysplasia, detached                       | 1 0.0%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 3 0.1%           | 2 0.3%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 7 0.2%           | 0                |
| 130.120 optic nerve hypoplasia                            | 5 0.2%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 29 1.0%          | 0                |
| 900.100 other, not inherited                              | 71 2.3%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 23 0.8%          | 22 3.4%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,747 57.8%      | 384 58.5%        |

# GREYHOUND

|    | DISORDER                             | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--------------------------------------|-------------|-----------|-----------------|
| A. | Chronic superficial keratitis/pannus | Not defined | 1, 2      | NO              |
| B. | Cataract                             | Not defined | 1         | NO              |

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## Description and Comments

### A. Chronic superficial keratitis/Pannus

A bilateral disease of the cornea which usually starts as a grayish haze to the ventral or ventrolateral cornea, followed by the formation of a vascularized subepithelial growth that begins to spread toward the central cornea; pigmentation follows the vascularization. If severe, vision impairment occurs. Pannus may be associated with plasma cell infiltration of the nictitans.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Peiffer RL, Jr., Gelatt KN, Gwin RM. Chronic superficial keratitis. *Vet Med Small Anim Clin.* 1977;72:35-37.



# OCULAR DISORDERS REPORT GREYHOUND

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>652 |      | 2016-2020<br>110 |      |
|---|---------------------|------------------|------|------------------|------|
|   |                     | #                | %    | #                | %    |
| <b>GLOBE</b>  |                     |                  |      |                  |      |
| 0.110 microphthalmia  |                     | 1                | 0.2% | 0                |      |
| <b>EYELIDS</b>  |                     |                  |      |                  |      |
| 25.110 distichiasis   |                     | 2                | 0.3% | 0                |      |
| <b>NASOLACRIMAL</b>   |                     |                  |      |                  |      |
| 40.910 keratoconjunctivitis sicca                                     |                     | 1                | 0.2% | 0                |      |
| <b>NICTITANS</b>  |                     |                  |      |                  |      |
| 50.210 pannus of third eyelid   |                     | 0                |      | 2                | 1.8% |
| 51.100 third eyelid cartilage anomaly                                 |                     | 2                | 0.3% | 0                |      |
| <b>CORNEA</b>   |                     |                  |      |                  |      |
| 70.210 corneal pannus   |                     | 21               | 3.2% | 0                |      |
| 70.700 corneal dystrophy  |                     | 5                | 0.8% | 1                | 0.9% |
| 70.730 corneal endothelial degeneration                               |                     | 1                | 0.2% | 0                |      |
| <b>UVEA</b>   |                     |                  |      |                  |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 2                | 0.3% | 0                |      |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 2                | 0.3% | 0                |      |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1                | 0.2% | 0                |      |
| <b>LENS</b>   |                     |                  |      |                  |      |
| 100.200 cataract, unspecified   |                     | 2                | 0.3% | 0                |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 21               | 3.2% | 8                | 7.3% |
| 100.301 punctate cataract, anterior cortex                            |                     | 5                | 0.8% | 0                |      |
| 100.302 punctate cataract, posterior cortex                           |                     | 2                | 0.3% | 0                |      |
| 100.304 punctate cataract, anterior sutures                           |                     | 2                | 0.3% | 0                |      |
| 100.306 punctate cataract, nucleus                                    |                     | 1                | 0.2% | 1                | 0.9% |
| 100.307 punctate cataract, capsular                                   |                     | 1                | 0.2% | 0                |      |
| 100.311 incipient cataract, anterior cortex                           |                     | 6                | 0.9% | 0                |      |
| 100.312 incipient cataract, posterior cortex                          |                     | 8                | 1.2% | 2                | 1.8% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 6                | 0.9% | 0                |      |
| 100.314 incipient cataract, anterior sutures                          |                     | 1                | 0.2% | 0                |      |
| 100.316 incipient cataract, nucleus                                   |                     | 2                | 0.3% | 0                |      |
| 100.317 incipient cataract, capsular                                  |                     | 2                | 0.3% | 0                |      |
| 100.322 incomplete cataract, posterior cortex                         |                     | 0                |      | 1                | 0.9% |
| 100.330 generalized/complete cataract                                 |                     | 1                | 0.2% | 0                |      |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 39               | 6.0% | 4                | 3.6% |
| 100.375 <i>subluxation/luxation, unspecified</i>                      |                     | 2                | 0.3% | 0                |      |
| <b>VITREOUS</b>   |                     |                  |      |                  |      |
| 110.120 persistent hyaloid artery/remnant                             |                     | 1                | 0.2% | 1                | 0.9% |
| 110.320 vitreal degeneration  |                     | 16               | 2.5% | 0                |      |
| <b>RETINA</b>   |                     |                  |      |                  |      |
| 120.170 retinal dysplasia, folds                                      |                     | 3                | 0.5% | 2                | 1.8% |
| 120.180 retinal dysplasia, geographic                                 |                     | 1                | 0.2% | 0                |      |
| 120.310 generalized progressive retinal atrophy (PRA)                 |                     | 6                | 0.9% | 1                | 0.9% |

| <b>RETINA CONTINUED</b>                                   | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 120.920 retinal detachment with dialysis                  | 0                | 1 0.9%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 2 0.3%           | 0                |
| 130.120 optic nerve hypoplasia                            | 2 0.3%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 8 1.2%           | 0                |
| 900.100 other, not inherited                              | 14 2.1%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 25 3.8%          | 9 8.2%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 534 81.9%        | 84 76.4%         |

# **OCULAR DISORDERS REPORT HARRIER**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the HARRIER breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT HARRIER

| Diagnostic Name    | TOTAL DOGS EXAMINED                                  | 1991-2015 |       | 2016-2020 |       |
|--------------------|--|-----------|-------|-----------|-------|
|                    |  | #         | %     | #         | %     |
| <b>EYELIDS</b>     |  |           |       |           |       |
| 21.000             | entropion, unspecified                               | 1         | 0.3%  | 0         |       |
| 25.110             | distichiasis   | 2         | 0.5%  | 0         |       |
| <b>CORNEA</b>      |  |           |       |           |       |
| 70.210             | corneal pannus                                       | 1         | 0.3%  | 0         |       |
| 70.700             | corneal dystrophy                                    | 0         |       | 1         | 2.6%  |
| <b>UVEA</b>        |  |           |       |           |       |
| 93.710             | persistent pupillary membranes, iris to iris         | 12        | 3.0%  | 0         |       |
| 93.730             | persistent pupillary membranes, iris to cornea       | 1         | 0.3%  | 0         |       |
| 93.740             | persistent pupillary membranes, iris sheets          | 1         | 0.3%  | 0         |       |
| <b>LENS</b>        |  |           |       |           |       |
| 100.210            | cataract. suspect not inherited/significance unknown | 8         | 2.0%  | 0         |       |
| 100.302            | punctate cataract, posterior cortex                  | 2         | 0.5%  | 0         |       |
| 100.306            | punctate cataract, nucleus                           | 1         | 0.3%  | 0         |       |
| 100.311            | incipient cataract, anterior cortex                  | 4         | 1.0%  | 0         |       |
| 100.312            | incipient cataract, posterior cortex                 | 3         | 0.8%  | 0         |       |
| 100.322            | incomplete cataract, posterior cortex                | 0         |       | 1         | 2.6%  |
| 100.345            | <i>significant cataracts (summary)</i>               | 10        | 2.5%  | 1         | 2.6%  |
| <b>VITREOUS</b>    |  |           |       |           |       |
| 110.120            | persistent hyaloid artery/remnant                    | 1         | 0.3%  | 0         |       |
| <b>FUNDUS</b>      |  |           |       |           |       |
| 97.120             | coloboma   | 1         | 0.3%  | 0         |       |
| <b>RETINA</b>      |  |           |       |           |       |
| 120.310            | generalized progressive retinal atrophy (PRA)        | 3         | 0.8%  | 0         |       |
| <b>OPTIC NERVE</b> |  |           |       |           |       |
| 130.150            | optic disc coloboma                                  | 1         | 0.3%  | 0         |       |
| <b>OTHER</b>       |  |           |       |           |       |
| 900.000            | other, unspecified                                   | 2         | 0.5%  | 0         |       |
| 900.100            | other, not inherited                                 | 11        | 2.8%  | 0         |       |
| 900.110            | other. suspect not inherited/significance unknown    | 4         | 1.0%  | 1         | 2.6%  |
| <b>NORMAL</b>      |  |           |       |           |       |
| 0.000              | normal globe   | 368       | 92.2% | 36        | 94.7% |

# HAVANA SILK DOG

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Distichiasis                                     | Not defined        | 1                | Breeder option         |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin, which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Havana Silk Dog breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT HAVANA SILK DOG

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015<br>621 |       | 2016-2020<br>100 |       |
|------------------|--|------------------|-------|------------------|-------|
|                  |  | #                | %     | #                | %     |
| <b>EYELIDS</b>   |  |                  |       |                  |       |
| 25.110           | distichiasis   | 32               | 5.2%  | 3                | 3.0%  |
| <b>NICTITANS</b> |  |                  |       |                  |       |
| 52.110           | prolapsed gland of the third eyelid                            | 3                | 0.5%  | 0                |       |
| <b>CORNEA</b>    |  |                  |       |                  |       |
| 70.700           | corneal dystrophy  | 9                | 1.4%  | 2                | 2.0%  |
| <b>UVEA</b>      |  |                  |       |                  |       |
| 93.710           | persistent pupillary membranes, iris to iris                   | 32               | 5.2%  | 2                | 2.0%  |
| 93.740           | persistent pupillary membranes, iris sheets                    | 1                | 0.2%  | 0                |       |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands | 1                | 0.2%  | 0                |       |
| <b>LENS</b>      |  |                  |       |                  |       |
| 100.210          | cataract. suspect not inherited/significance unknown           | 18               | 2.9%  | 4                | 4.0%  |
| 100.301          | punctate cataract, anterior cortex                             | 1                | 0.2%  | 0                |       |
| 100.304          | punctate cataract, anterior sutures                            | 1                | 0.2%  | 0                |       |
| 100.311          | incipient cataract, anterior cortex                            | 2                | 0.3%  | 0                |       |
| 100.312          | incipient cataract, posterior cortex                           | 3                | 0.5%  | 0                |       |
| 100.313          | incipient cataract, equatorial cortex                          | 1                | 0.2%  | 0                |       |
| 100.316          | incipient cataract, nucleus                                    | 1                | 0.2%  | 0                |       |
| 100.330          | generalized/complete cataract                                  | 2                | 0.3%  | 0                |       |
| 100.345          | <i>significant cataracts (summary)</i>                         | 11               | 1.8%  | 0                |       |
| 100.375          | <i>subluxation/luxation, unspecified</i>                       | 1                | 0.2%  | 0                |       |
| <b>VITREOUS</b>  |  |                  |       |                  |       |
| 110.120          | persistent hyaloid artery/remnant                              | 2                | 0.3%  | 0                |       |
| 110.320          | vitreal degeneration   | 7                | 1.1%  | 0                |       |
| <b>RETINA</b>    |  |                  |       |                  |       |
| 120.170          | retinal dysplasia, folds                                       | 1                | 0.2%  | 0                |       |
| <b>OTHER</b>     |  |                  |       |                  |       |
| 900.000          | other, unspecified   | 7                | 1.1%  | 0                |       |
| 900.100          | other, not inherited   | 1                | 0.2%  | 0                |       |
| 900.110          | other. suspect not inherited/significance unknown              | 6                | 1.0%  | 1                | 1.0%  |
| <b>NORMAL</b>    |  |                  |       |                  |       |
| 0.000            | normal globe   | 535              | 86.2% | 88               | 88.0% |

# HAVANESE

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Distichiasis                                     | Not defined        | 1                | Breeder option         |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |
| C. | Cataract   | Not defined        | 1, 2             | NO                     |
| D. | Y-suture tip opacity                             | Not defined        | 1                | Breeder option         |
| E. | Vitreous degeneration                            | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin, which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

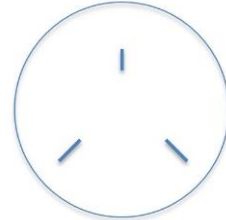
Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region. The exact frequency and significance of cataracts in the breed is not known.

#### D. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

#### E. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

### References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Starr AN, Famula TR, Markward NJ, et al. Hereditary evaluation of multiple developmental abnormalities in the Havanese dog breed. *J Hered.* 2007;98:510-517.



# OCULAR DISORDERS REPORT HAVANESE

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>27,503 |      | 2016-2020<br>6,037 |      |
|---|---------------------|---------------------|------|--------------------|------|
|   |                     | #                   | %    | #                  | %    |
| <b>GLOBE</b>  |                     |                     |      |                    |      |
| 0.110 microphthalmia  |                     | 6                   | 0.0% | 1                  | 0.0% |
| <b>EYELIDS</b>  |                     |                     |      |                    |      |
| 20.140 ectopic cilia  |                     | 10                  | 0.0% | 2                  | 0.0% |
| 21.000 entropion, unspecified   |                     | 18                  | 0.1% | 3                  | 0.0% |
| 22.000 ectropion, unspecified   |                     | 4                   | 0.0% | 0                  |      |
| 25.110 distichiasis   |                     | 1,372               | 5.0% | 296                | 4.9% |
| <b>NASOLACRIMAL</b>   |                     |                     |      |                    |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 8                   | 0.0% | 5                  | 0.1% |
| 40.910 keratoconjunctivitis sicca                                     |                     | 9                   | 0.0% | 1                  | 0.0% |
| <b>NICTITANS</b>  |                     |                     |      |                    |      |
| 51.100 third eyelid cartilage anomaly                                 |                     | 2                   | 0.0% | 1                  | 0.0% |
| 52.110 prolapsed gland of the third eyelid                            |                     | 126                 | 0.5% | 30                 | 0.5% |
| <b>CORNEA</b>   |                     |                     |      |                    |      |
| 70.210 corneal pannus   |                     | 1                   | 0.0% | 1                  | 0.0% |
| 70.220 pigmentary keratitis   |                     | 3                   | 0.0% | 4                  | 0.1% |
| 70.700 corneal dystrophy  |                     | 108                 | 0.4% | 34                 | 0.6% |
| 70.730 corneal endothelial degeneration                               |                     | 3                   | 0.0% | 2                  | 0.0% |
| <b>UVEA</b>   |                     |                     |      |                    |      |
| 90.250 pigmentary uveitis   |                     | 1                   | 0.0% | 0                  |      |
| 93.110 iris hypoplasia  |                     | 0                   |      | 2                  | 0.0% |
| 93.140 corneal endothelial pigment without PPM                        |                     | 3                   | 0.0% | 0                  |      |
| 93.150 iris coloboma  |                     | 1                   | 0.0% | 2                  | 0.0% |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 1,699               | 6.2% | 314                | 5.2% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 28                  | 0.1% | 4                  | 0.1% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 13                  | 0.0% | 1                  | 0.0% |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 18                  | 0.1% | 0                  |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 30                  | 0.1% | 25                 | 0.4% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 4                   | 0.0% | 2                  | 0.0% |
| 93.810 uveal melanoma   |                     | 3                   | 0.0% | 0                  |      |
| 93.999 uveal cysts  |                     | 3                   | 0.0% | 2                  | 0.0% |
| <b>LENS</b>   |                     |                     |      |                    |      |
| 100.200 cataract, unspecified   |                     | 22                  | 0.1% | 0                  |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 1,592               | 5.8% | 365                | 6.0% |
| 100.301 punctate cataract, anterior cortex                            |                     | 116                 | 0.4% | 41                 | 0.7% |
| 100.302 punctate cataract, posterior cortex                           |                     | 93                  | 0.3% | 41                 | 0.7% |
| 100.303 punctate cataract, equatorial cortex                          |                     | 34                  | 0.1% | 5                  | 0.1% |
| 100.304 punctate cataract, anterior sutures                           |                     | 26                  | 0.1% | 9                  | 0.1% |
| 100.305 punctate cataract, posterior sutures                          |                     | 190                 | 0.7% | 72                 | 1.2% |
| 100.306 punctate cataract, nucleus                                    |                     | 16                  | 0.1% | 8                  | 0.1% |
| 100.307 punctate cataract, capsular                                   |                     | 38                  | 0.1% | 21                 | 0.3% |
| 100.311 incipient cataract, anterior cortex                           |                     | 113                 | 0.4% | 27                 | 0.4% |
| 100.312 incipient cataract, posterior cortex                          |                     | 211                 | 0.8% | 39                 | 0.6% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 46                  | 0.2% | 15                 | 0.2% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.314 incipient cataract, anterior sutures              | 14               | 0.1%  | 3                | 0.0%  |
| 100.315 incipient cataract, posterior sutures             | 92               | 0.3%  | 17               | 0.3%  |
| 100.316 incipient cataract, nucleus                       | 20               | 0.1%  | 2                | 0.0%  |
| 100.317 incipient cataract, capsular                      | 47               | 0.2%  | 7                | 0.1%  |
| 100.321 incomplete cataract, anterior cortex              | 4                | 0.0%  | 5                | 0.1%  |
| 100.322 incomplete cataract, posterior cortex             | 9                | 0.0%  | 11               | 0.2%  |
| 100.323 incomplete cataract, equatorial cortex            | 1                | 0.0%  | 0                |       |
| 100.325 incomplete cataract, posterior sutures            | 1                | 0.0%  | 2                | 0.0%  |
| 100.326 incomplete cataract, nucleus                      | 2                | 0.0%  | 1                | 0.0%  |
| 100.327 incomplete cataract, capsular                     | 1                | 0.0%  | 0                |       |
| 100.328 y-suture tip opacities                            | 46               | 0.2%  | 133              | 2.2%  |
| 100.330 generalized/complete cataract                     | 123              | 0.4%  | 6                | 0.1%  |
| 100.340 resorbing/hypermature cataract                    | 2                | 0.0%  | 3                | 0.0%  |
| 100.345 <i>significant cataracts (summary)</i>            | 1,267            | 4.6%  | 468              | 7.8%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 11               | 0.0%  | 3                | 0.0%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 29               | 0.1%  | 4                | 0.1%  |
| 110.135 PHPV/PTVL   | 3                | 0.0%  | 2                | 0.0%  |
| 110.200 vitreous degeneration-anterior chamber            | 13               | 0.0%  | 20               | 0.3%  |
| 110.320 vitreal degeneration                              | 493              | 1.8%  | 71               | 1.2%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 2                | 0.0%  | 0                |       |
| 97.120 coloboma   | 4                | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 138              | 0.5%  | 10               | 0.2%  |
| 120.180 retinal dysplasia, geographic                     | 22               | 0.1%  | 6                | 0.1%  |
| 120.190 retinal dysplasia, detached                       | 1                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 104              | 0.4%  | 8                | 0.1%  |
| 120.400 retinal hemorrhage                                | 1                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 12               | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 0                |       | 4                | 0.1%  |
| 120.960 retinopathy                                       | 12               | 0.0%  | 13               | 0.2%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 1                | 0.0%  | 0                |       |
| 130.120 optic nerve hypoplasia                            | 3                | 0.0%  | 1                | 0.0%  |
| 130.150 optic disc coloboma                               | 7                | 0.0%  | 1                | 0.0%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 257              | 0.9%  | 0                |       |
| 900.100 other, not inherited                              | 558              | 2.0%  | 12               | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown | 200              | 0.7%  | 204              | 3.4%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 22,829           | 83.0% | 4,472            | 74.1% |

# HOKKAIDO DOG

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE           |
|----|---|------------------------|-----------|--------------------|--------------------------------------|
| A. | Choroidal hypoplasia<br>(Collie Eye Anomaly)<br>- staphyloma/coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- optic nerve coloboma | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>NHEJ1</i> gene |

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## Description and Comments

- A. Choroidal hypoplasia (Collie Eye Anomaly)  
- staphyloma/coloboma  
- retinal detachment  
- retinal hemorrhage  
- optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

## References

1. Mizukami K, Chang H, Ota M, et al. Collie eye anomaly in Hokkaido dogs: case study. *Vet Ophthalmol.* 2012;15:128-32.

# OCULAR DISORDERS REPORT HOKKAIDO KEN

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | 2         |       | 17        |       |
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 0         |       | 1         | 5.9%  |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 0         |       | 5         | 29.4% |
| 93.720          | persistent pupillary membranes, iris to lens                 | 0         |       | 1         | 5.9%  |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 0         |       | 1         | 5.9%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 1         | 50.0% | 0         |       |
| 100.311         | incipient cataract, anterior cortex                          | 0         |       | 2         | 11.8% |
| 100.321         | incomplete cataract, anterior cortex                         | 0         |       | 1         | 5.9%  |
| 100.330         | generalized/complete cataract                                | 0         |       | 1         | 5.9%  |
| 100.345         | significant cataracts (summary)                              | 0         |       | 4         | 23.5% |
| <b>FUNDUS</b>   |  |           |       |           |       |
| 97.110          | choroidal hypoplasia   | 0         |       | 5         | 29.4% |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                                     | 0         |       | 1         | 5.9%  |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.100         | other, not inherited   | 1         | 50.0% | 0         |       |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 0         |       | 7         | 41.2% |

# IBIZAN HOUND

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--------------------------------|-------------|-----------|-------------------------|
| A. | Persistent pupillary membranes |             |           |                         |
|    | - iris to iris                 | Not defined | 1         | Breeder option          |
|    | - lens pigment foci/no strands | Not defined | 1         | Passes with no notation |
| B. | Cataract                       | Not defined | 1         | NO                      |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Ibizan Hound breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT IBIZAN HOUND

| Diagnostic Name   | TOTAL DOGS EXAMINED |       | 1991-2015<br>1,278 |       | 2016-2020<br>520 |   |
|---|---------------------|-------|--------------------|-------|------------------|---|
|   | #                   | %     | #                  | %     | #                | % |
| <b>GLOBE</b>  |                     |       |                    |       |                  |   |
| 0.110 microphthalmia  | 3                   | 0.2%  | 1                  | 0.2%  |                  |   |
| <b>EYELIDS</b>  |                     |       |                    |       |                  |   |
| 25.110 distichiasis   | 4                   | 0.3%  | 0                  |       |                  |   |
| <b>NASOLACRIMAL</b>   |                     |       |                    |       |                  |   |
| 40.910 keratoconjunctivitis sicca                                     | 1                   | 0.1%  | 0                  |       |                  |   |
| <b>NICTITANS</b>  |                     |       |                    |       |                  |   |
| 51.100 third eyelid cartilage anomaly                                 | 1                   | 0.1%  | 0                  |       |                  |   |
| 52.110 prolapsed gland of the third eyelid                            | 1                   | 0.1%  | 0                  |       |                  |   |
| <b>CORNEA</b>   |                     |       |                    |       |                  |   |
| 70.700 corneal dystrophy  | 8                   | 0.6%  | 3                  | 0.6%  |                  |   |
| <b>UVEA</b>   |                     |       |                    |       |                  |   |
| 93.140 corneal endothelial pigment without PPM                        | 1                   | 0.1%  | 0                  |       |                  |   |
| 93.150 iris coloboma  | 0                   |       | 1                  | 0.2%  |                  |   |
| 93.710 persistent pupillary membranes, iris to iris                   | 155                 | 12.1% | 57                 | 11.0% |                  |   |
| 93.720 persistent pupillary membranes, iris to lens                   | 1                   | 0.1%  | 1                  | 0.2%  |                  |   |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   | 12                  | 0.9%  | 6                  | 1.2%  |                  |   |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands | 5                   | 0.4%  | 0                  |       |                  |   |
| 93.999 uveal cysts  | 4                   | 0.3%  | 1                  | 0.2%  |                  |   |
| 97.150 chorioretinal coloboma, congenital                             | 0                   |       | 1                  | 0.2%  |                  |   |
| <b>LENS</b>   |                     |       |                    |       |                  |   |
| 100.200 cataract, unspecified   | 4                   | 0.3%  | 0                  |       |                  |   |
| 100.210 cataract. suspect not inherited/significance unknown          | 81                  | 6.3%  | 12                 | 2.3%  |                  |   |
| 100.301 punctate cataract, anterior cortex                            | 3                   | 0.2%  | 2                  | 0.4%  |                  |   |
| 100.302 punctate cataract, posterior cortex                           | 2                   | 0.2%  | 3                  | 0.6%  |                  |   |
| 100.303 punctate cataract, equatorial cortex                          | 1                   | 0.1%  | 0                  |       |                  |   |
| 100.304 punctate cataract, anterior sutures                           | 1                   | 0.1%  | 1                  | 0.2%  |                  |   |
| 100.305 punctate cataract, posterior sutures                          | 1                   | 0.1%  | 5                  | 1.0%  |                  |   |
| 100.306 punctate cataract, nucleus                                    | 7                   | 0.5%  | 0                  |       |                  |   |
| 100.307 punctate cataract, capsular                                   | 2                   | 0.2%  | 4                  | 0.8%  |                  |   |
| 100.311 incipient cataract, anterior cortex                           | 6                   | 0.5%  | 1                  | 0.2%  |                  |   |
| 100.312 incipient cataract, posterior cortex                          | 9                   | 0.7%  | 2                  | 0.4%  |                  |   |
| 100.313 incipient cataract, equatorial cortex                         | 4                   | 0.3%  | 1                  | 0.2%  |                  |   |
| 100.314 incipient cataract, anterior sutures                          | 1                   | 0.1%  | 1                  | 0.2%  |                  |   |
| 100.316 incipient cataract, nucleus                                   | 19                  | 1.5%  | 11                 | 2.1%  |                  |   |
| 100.317 incipient cataract, capsular                                  | 2                   | 0.2%  | 1                  | 0.2%  |                  |   |
| 100.322 incomplete cataract, posterior cortex                         | 1                   | 0.1%  | 0                  |       |                  |   |
| 100.327 incomplete cataract, capsular                                 | 1                   | 0.1%  | 0                  |       |                  |   |
| 100.328 y-suture tip opacities  | 1                   | 0.1%  | 1                  | 0.2%  |                  |   |
| 100.330 generalized/complete cataract                                 | 2                   | 0.2%  | 0                  |       |                  |   |
| 100.345 <i>significant cataracts (summary)</i>                        | 67                  | 5.2%  | 33                 | 6.3%  |                  |   |
| 100.375 <i>subluxation/luxation, unspecified</i>                      | 3                   | 0.2%  | 0                  |       |                  |   |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>VITREOUS</b>   |             |           |
| 110.120 persistent hyaloid artery/remnant                 | 3 0.2%      | 3 0.6%    |
| 110.200 vitreous degeneration-anterior chamber            | 1 0.1%      | 1 0.2%    |
| 110.320 vitreal degeneration                              | 15 1.2%     | 1 0.2%    |
| <b>FUNDUS</b>   |             |           |
| 97.110 choroidal hypoplasia                               | 0           | 1 0.2%    |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 11 0.9%     | 0         |
| 120.180 retinal dysplasia, geographic                     | 2 0.2%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 4 0.3%      | 0         |
| 120.910 retinal detachment without dialysis               | 1 0.1%      | 0         |
| 120.960 retinopathy                                       | 1 0.1%      | 0         |
| <b>OPTIC NERVE</b>  |             |           |
| 130.150 optic disc coloboma                               | 3 0.2%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 24 1.9%     | 0         |
| 900.100 other, not inherited                              | 20 1.6%     | 1 0.2%    |
| 900.110 other. suspect not inherited/significance unknown | 11 0.9%     | 19 3.7%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,021 79.9% | 394 75.8% |

# ICELANDIC SHEEPDOG

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| B. | Cataract   | Not defined | 1         | NO              |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Icelandic Sheepdog breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.



# OCULAR DISORDERS REPORT ICELANDIC SHEEPDOG

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>1,896 |      | 2016-2020<br>920 |      |
|---------------------|--|--------------------|------|------------------|------|
|                     |  | #                  | %    | #                | %    |
| <b>EYELIDS</b>      |  |                    |      |                  |      |
| 21.000              | entropion, unspecified                                       | 5                  | 0.3% | 0                |      |
| 25.110              | distichiasis   | 16                 | 0.8% | 8                | 0.9% |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |      |
| 32.110              | imperforate lower nasolacrimal punctum                       | 0                  |      | 1                | 0.1% |
| <b>NICTITANS</b>    |  |                    |      |                  |      |
| 50.210              | pannus of third eyelid                                       | 0                  |      | 1                | 0.1% |
| <b>CORNEA</b>       |  |                    |      |                  |      |
| 70.210              | corneal pannus   | 0                  |      | 1                | 0.1% |
| 70.220              | pigmentary keratitis   | 0                  |      | 1                | 0.1% |
| 70.700              | corneal dystrophy  | 9                  | 0.5% | 0                |      |
| <b>UVEA</b>         |  |                    |      |                  |      |
| 93.110              | iris hypoplasia  | 2                  | 0.1% | 0                |      |
| 93.710              | persistent pupillary membranes, iris to iris                 | 102                | 5.4% | 24               | 2.6% |
| 93.720              | persistent pupillary membranes, iris to lens                 | 1                  | 0.1% | 0                |      |
| 93.730              | persistent pupillary membranes, iris to cornea               | 3                  | 0.2% | 0                |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands | 1                  | 0.1% | 2                | 0.2% |
| <b>LENS</b>         |  |                    |      |                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown         | 47                 | 2.5% | 32               | 3.5% |
| 100.301             | punctate cataract, anterior cortex                           | 3                  | 0.2% | 5                | 0.5% |
| 100.302             | punctate cataract, posterior cortex                          | 4                  | 0.2% | 2                | 0.2% |
| 100.303             | punctate cataract, equatorial cortex                         | 1                  | 0.1% | 0                |      |
| 100.304             | punctate cataract, anterior sutures                          | 1                  | 0.1% | 1                | 0.1% |
| 100.305             | punctate cataract, posterior sutures                         | 4                  | 0.2% | 5                | 0.5% |
| 100.307             | punctate cataract, capsular                                  | 0                  |      | 2                | 0.2% |
| 100.311             | incipient cataract, anterior cortex                          | 3                  | 0.2% | 0                |      |
| 100.312             | incipient cataract, posterior cortex                         | 13                 | 0.7% | 2                | 0.2% |
| 100.313             | incipient cataract, equatorial cortex                        | 3                  | 0.2% | 0                |      |
| 100.315             | incipient cataract, posterior sutures                        | 8                  | 0.4% | 0                |      |
| 100.317             | incipient cataract, capsular                                 | 2                  | 0.1% | 1                | 0.1% |
| 100.321             | incomplete cataract, anterior cortex                         | 1                  | 0.1% | 3                | 0.3% |
| 100.322             | incomplete cataract, posterior cortex                        | 3                  | 0.2% | 1                | 0.1% |
| 100.328             | y-suture tip opacities                                       | 1                  | 0.1% | 6                | 0.7% |
| 100.330             | generalized/complete cataract                                | 1                  | 0.1% | 0                |      |
| 100.345             | <i>significant cataracts (summary)</i>                       | 48                 | 2.5% | 28               | 3.0% |
| <b>VITREOUS</b>     |  |                    |      |                  |      |
| 110.120             | persistent hyaloid artery/remnant                            | 3                  | 0.2% | 1                | 0.1% |
| 110.320             | vitreal degeneration   | 4                  | 0.2% | 1                | 0.1% |
| <b>RETINA</b>       |  |                    |      |                  |      |
| 120.170             | retinal dysplasia, folds                                     | 9                  | 0.5% | 0                |      |
| 120.180             | retinal dysplasia, geographic                                | 1                  | 0.1% | 0                |      |
| 120.310             | generalized progressive retinal atrophy (PRA)                | 1                  | 0.1% | 0                |      |
| 120.960             | retinopathy  | 0                  |      | 3                | 0.3% |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>OPTIC NERVE</b>  |             |           |
| 130.150 optic disc coloboma                               | 2 0.1%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 25 1.3%     | 0         |
| 900.100 other, not inherited                              | 31 1.6%     | 1 0.1%    |
| 900.110 other. suspect not inherited/significance unknown | 24 1.3%     | 49 5.3%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,730 91.2% | 787 85.5% |

## IRISH RED AND WHITE SETTER

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE             |
|----|---|------------------------|-----------|--------------------|--|
| A. | Retinal atrophy<br>- rod-cone<br>dysplasia, type 1<br>( <i>rcd1</i> ) | Autosomal<br>recessive | 2-21      | NO                 | Mutation of the<br><i>PDE6B</i> gene   |
| B. | Retinal atrophy<br>- rod-cone<br>dysplasia, type 4<br>( <i>rcd4</i> ) | Autosomal<br>recessive | 22        | NO                 | mutation of the<br><i>C2orf71</i> gene |
| C. | Cataract  | Not defined            | 1         | NO                 |  |

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### Description and Comments

A. Retinal atrophy - rod-cone dysplasia, type 1 (*rcd1*)

A form of PRA identified in Irish Setters and Irish Red and White Setters. Clinical night blindness is observed as early as 6 weeks of age progressing to total blindness by one year. It may be diagnosed as early as 24 days with an ERG. Histologically the disease can be detected by 6 weeks. The disorder is caused by a mutation present in exon 21/codon 807 of the *PDE6B* gene. A DNA test is now available that will unequivocally identify genetically normal, affected and carrier dogs. The test is accurate only for this mutation and will not identify other forms of PRA.

B. Retinal atrophy - rod-cone dysplasia, type 4 (*rcd4*)

A form of PRA identified in the Gordon and Irish Setter breeds. Clinical night blindness is observed on average as late as 10 years of age and progresses to total blindness. This form of PRA has been referred to as late-onset PRA (LOPRA). The disorder is caused by a mutation present in the *C2orf71* gene. A DNA test is now available that will unequivocally identify genetically normal, affected and carrier dogs. The test is accurate only for this mutation and will not identify other forms of PRA.

C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
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# OCULAR DISORDERS REPORT IRISH RED & WHITE SETTER

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015<br>441 |       | 2016-2020<br>222 |       |
|-----------------|--|------------------|-------|------------------|-------|
|                 |  | #                | %     | #                | %     |
| <b>EYELIDS</b>  |  |                  |       |                  |       |
| 21.000          | entropion, unspecified   | 1                | 0.2%  | 0                |       |
| 25.110          | distichiasis   | 21               | 4.8%  | 2                | 0.9%  |
| <b>CORNEA</b>   |  |                  |       |                  |       |
| 70.210          | corneal pannus   | 2                | 0.5%  | 0                |       |
| 70.700          | corneal dystrophy  | 1                | 0.2%  | 0                |       |
| 70.730          | corneal endothelial degeneration                               | 0                |       | 1                | 0.5%  |
| <b>UVEA</b>     |  |                  |       |                  |       |
| 93.710          | persistent pupillary membranes, iris to iris                   | 7                | 1.6%  | 1                | 0.5%  |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands   | 1                | 0.2%  | 2                | 0.9%  |
| 93.760          | persistent pupillary membranes, endothelial opacity/no strands | 1                | 0.2%  | 0                |       |
| 93.999          | uveal cysts  | 3                | 0.7%  | 0                |       |
| <b>LENS</b>     |  |                  |       |                  |       |
| 100.210         | cataract. suspect not inherited/significance unknown           | 17               | 3.9%  | 9                | 4.1%  |
| 100.301         | punctate cataract, anterior cortex                             | 3                | 0.7%  | 2                | 0.9%  |
| 100.302         | punctate cataract, posterior cortex                            | 6                | 1.4%  | 2                | 0.9%  |
| 100.304         | punctate cataract, anterior sutures                            | 1                | 0.2%  | 0                |       |
| 100.307         | punctate cataract, capsular                                    | 2                | 0.5%  | 3                | 1.4%  |
| 100.311         | incipient cataract, anterior cortex                            | 4                | 0.9%  | 1                | 0.5%  |
| 100.312         | incipient cataract, posterior cortex                           | 7                | 1.6%  | 1                | 0.5%  |
| 100.315         | incipient cataract, posterior sutures                          | 1                | 0.2%  | 0                |       |
| 100.316         | incipient cataract, nucleus                                    | 1                | 0.2%  | 2                | 0.9%  |
| 100.317         | incipient cataract, capsular                                   | 0                |       | 1                | 0.5%  |
| 100.321         | incomplete cataract, anterior cortex                           | 1                | 0.2%  | 1                | 0.5%  |
| 100.322         | incomplete cataract, posterior cortex                          | 1                | 0.2%  | 1                | 0.5%  |
| 100.345         | <i>significant cataracts (summary)</i>                         | 27               | 6.1%  | 14               | 6.3%  |
| 100.375         | <i>subluxation/luxation, unspecified</i>                       | 1                | 0.2%  | 0                |       |
| <b>VITREOUS</b> |  |                  |       |                  |       |
| 110.135         | PHPV/PTVL  | 1                | 0.2%  | 0                |       |
| 110.200         | vitreous degeneration-anterior chamber                         | 1                | 0.2%  | 0                |       |
| 110.320         | vitreal degeneration   | 4                | 0.9%  | 2                | 0.9%  |
| <b>RETINA</b>   |  |                  |       |                  |       |
| 120.170         | retinal dysplasia, folds                                       | 4                | 0.9%  | 1                | 0.5%  |
| 120.180         | retinal dysplasia, geographic                                  | 2                | 0.5%  | 1                | 0.5%  |
| 120.310         | generalized progressive retinal atrophy (PRA)                  | 3                | 0.7%  | 0                |       |
| 120.960         | retinopathy  | 1                | 0.2%  | 0                |       |
| <b>OTHER</b>    |  |                  |       |                  |       |
| 900.000         | other, unspecified   | 5                | 1.1%  | 0                |       |
| 900.100         | other, not inherited   | 7                | 1.6%  | 0                |       |
| 900.110         | other. suspect not inherited/significance unknown              | 10               | 2.3%  | 20               | 9.0%  |
| <b>NORMAL</b>   |  |                  |       |                  |       |
| 0.000           | normal globe   | 371              | 84.1% | 179              | 80.6% |

# IRISH SETTER

|    | DISORDER   | INHERITANCE                        | REFERENCE | BREEDING<br>ADVICE         | GENETIC TESTS<br>AVAILABLE             |
|----|--|------------------------------------|-----------|----------------------------|--|
| A. | Entropion  | Not defined                        | 1         | Breeder option             |  |
| B. | Distichiasis   | Not defined                        | 1         | Breeder option             |  |
| C. | Persistent pupillary<br>membranes                                  |                                    |           |                            |  |
|    | - iris to iris   | Not defined                        | 1         | Breeder option             |  |
|    | - lens pigment foci/no<br>strands                                  | Not defined                        | 1         | Passes with no<br>notation |  |
| D. | Cataract   | Not defined                        | 1         | NO                         |  |
| E. | Persistent hyaloid<br>artery                                       | Not defined                        | 1         | Breeder option             |  |
| F. | Retinal atrophy -<br>generalized                                   | Presumed<br>autosomal<br>recessive | 1, 2-23   | NO                         |  |
| G. | Retinal atrophy<br>- rod-cone dysplasia,<br>type 1 ( <i>rcd1</i> ) | Autosomal<br>recessive             | 1, 2-21   | NO                         | Mutation of the<br><i>PDE6B</i> gene   |
| H. | Retinal atrophy<br>- rod-cone dysplasia<br>type 4 ( <i>rcd4</i> )  | Autosomal<br>recessive             | 25        | NO                         | Mutation of the<br><i>C2orf71</i> gene |
| I. | Amblyopia with<br>quadriplegia                                     | Autosomal<br>recessive             | 24-25     | NO                         |  |

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## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. In the Irish Setter, the entropion usually involves the lower eyelids.

B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Persistent hyaloid artery (PHA)

A congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small vascular strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

F. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. Except for X-linked PRA in the Siberian Husky, in all breeds studied to date, PRA is inherited as an autosomal recessive trait.

In the Irish Setter, a later form of progressive retinal atrophy has been observed by several ophthalmologists at 4-5 years of age. Cases seen in this category appear to advance more rapidly than those with rod-cone dysplasia.

G. Retinal atrophy - rod-cone dysplasia, type 1 (*rcd1*)

A form of PRA identified in Irish Setters. Clinical night blindness is observed as early as 6 weeks of age progressing to total blindness by one year. It may be diagnosed as early as 24



days with an ERG. Histologically the disease can be detected by 6 weeks. The disorder is caused by a mutation present in exon 21/codon 807 of the *PDE6B* gene. A DNA test is available that will unequivocally identify genetically normal, affected and carrier dogs. The test is accurate only for this mutation and will not identify other forms of PRA.

H. Retinal atrophy - rod-cone dysplasia, type 4 (*rcd4*)

A form of PRA identified in the Gordon and Irish Setter breeds. Clinical night blindness is observed on average as late as 10 years of age and progresses to total blindness. This form of PRA has been referred to as late-onset PRA (LOPRA). The disorder is caused by a mutation present in the *C2orf71* gene. A DNA test is available that will unequivocally identify genetically normal, affected and carrier dogs. The test is accurate only for this mutation and will not identify other forms of PRA.

I. Amblyopia with quadriplegia

A congenital quadriplegia and amblyopia. The main symptoms include inability to stand or walk, amblyopia, tremor, nystagmus and possible seizures. Pathologic lesions are confined to the cerebellum. The condition was shown to be due to a fully penetrant autosomal recessive gene that is post-natally lethal in the homozygote.

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# OCULAR DISORDERS REPORT IRISH SETTER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>2,047 |      | 2016-2020<br>257 |      |
|---------------------|--|--------------------|------|------------------|------|
|                     |  | #                  | %    | #                | %    |
| <b>GLOBE</b>        |  |                    |      |                  |      |
| 0.110               | microphthalmia   | 2                  | 0.1% | 0                |      |
| 10.000              | glaucoma   | 1                  | 0.0% | 0                |      |
| <b>EYELIDS</b>      |  |                    |      |                  |      |
| 20.140              | ectopic cilia  | 1                  | 0.0% | 0                |      |
| 20.160              | macropalpebral fissure   | 2                  | 0.1% | 0                |      |
| 21.000              | entropion, unspecified   | 53                 | 2.6% | 3                | 1.2% |
| 22.000              | ectropion, unspecified   | 9                  | 0.4% | 0                |      |
| 25.110              | distichiasis   | 115                | 5.6% | 15               | 5.8% |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 1                  | 0.0% | 1                | 0.4% |
| 40.910              | keratoconjunctivitis sicca                                     | 1                  | 0.0% | 0                |      |
| <b>NICTITANS</b>    |  |                    |      |                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 3                  | 0.1% | 0                |      |
| <b>CORNEA</b>       |  |                    |      |                  |      |
| 70.210              | corneal pannus   | 1                  | 0.0% | 0                |      |
| 70.220              | pigmentary keratitis   | 1                  | 0.0% | 0                |      |
| 70.700              | corneal dystrophy  | 6                  | 0.3% | 0                |      |
| 70.730              | corneal endothelial degeneration                               | 1                  | 0.0% | 0                |      |
| <b>UVEA</b>         |  |                    |      |                  |      |
| 93.140              | corneal endothelial pigment without PPM                        | 2                  | 0.1% | 0                |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 81                 | 4.0% | 19               | 7.4% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 7                  | 0.3% | 0                |      |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 6                  | 0.3% | 0                |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 21                 | 1.0% | 12               | 4.7% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 3                  | 0.1% | 1                | 0.4% |
| 93.810              | uveal melanoma   | 1                  | 0.0% | 0                |      |
| 93.999              | uveal cysts  | 3                  | 0.1% | 2                | 0.8% |
| <b>LENS</b>         |  |                    |      |                  |      |
| 100.200             | cataract, unspecified  | 31                 | 1.5% | 0                |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 105                | 5.1% | 5                | 1.9% |
| 100.301             | punctate cataract, anterior cortex                             | 8                  | 0.4% | 3                | 1.2% |
| 100.302             | punctate cataract, posterior cortex                            | 10                 | 0.5% | 2                | 0.8% |
| 100.303             | punctate cataract, equatorial cortex                           | 4                  | 0.2% | 0                |      |
| 100.304             | punctate cataract, anterior sutures                            | 1                  | 0.0% | 0                |      |
| 100.305             | punctate cataract, posterior sutures                           | 2                  | 0.1% | 1                | 0.4% |
| 100.306             | punctate cataract, nucleus                                     | 4                  | 0.2% | 0                |      |
| 100.307             | punctate cataract, capsular                                    | 8                  | 0.4% | 2                | 0.8% |
| 100.311             | incipient cataract, anterior cortex                            | 19                 | 0.9% | 1                | 0.4% |
| 100.312             | incipient cataract, posterior cortex                           | 20                 | 1.0% | 1                | 0.4% |
| 100.313             | incipient cataract, equatorial cortex                          | 5                  | 0.2% | 0                |      |
| 100.314             | incipient cataract, anterior sutures                           | 4                  | 0.2% | 0                |      |
| 100.315             | incipient cataract, posterior sutures                          | 4                  | 0.2% | 0                |      |
| 100.316             | incipient cataract, nucleus                                    | 8                  | 0.4% | 1                | 0.4% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.317 incipient cataract, capsular                      | 3 0.1%           | 1 0.4%           |
| 100.321 incomplete cataract, anterior cortex              | 1 0.0%           | 0                |
| 100.322 incomplete cataract, posterior cortex             | 1 0.0%           | 0                |
| 100.325 incomplete cataract, posterior sutures            | 1 0.0%           | 0                |
| 100.328 y-suture tip opacities                            | 0                | 1 0.4%           |
| 100.330 generalized/complete cataract                     | 18 0.9%          | 0                |
| 100.340 resorbing/hypermature cataract                    | 1 0.0%           | 0                |
| 100.345 <i>significant cataracts (summary)</i>            | 153 7.5%         | 13 5.1%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 1 0.0%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 22 1.1%          | 5 1.9%           |
| 110.135 PHPV/PTVL   | 10 0.5%          | 0                |
| 110.320 vitreal degeneration                              | 4 0.2%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 10 0.5%          | 2 0.8%           |
| 120.180 retinal dysplasia, geographic                     | 1 0.0%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 18 0.9%          | 0                |
| 120.960 retinopathy                                       | 0                | 1 0.4%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.120 optic nerve hypoplasia                            | 4 0.2%           | 0                |
| 130.150 optic disc coloboma                               | 1 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 19 0.9%          | 0                |
| 900.100 other, not inherited                              | 38 1.9%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 41 2.0%          | 11 4.3%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,564 76.4%      | 187 72.8%        |

# IRISH WATER SPANIEL

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Distichiasis                                     | Not defined | 1         | Breeder option  |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| C. | Cataract   | Not defined | 1         | NO              |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Irish Water Spaniel breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report

# OCULAR DISORDERS REPORT

## IRISH WATER SPANIEL

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 20.140          | ectopic cilia  | 1         | 0.1%  | 0         |       |
| 21.000          | entropion, unspecified   | 10        | 0.9%  | 0         |       |
| 22.000          | ectropion, unspecified   | 3         | 0.3%  | 0         |       |
| 25.110          | distichiasis   | 277       | 25.6% | 53        | 25.4% |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.700          | corneal dystrophy  | 4         | 0.4%  | 3         | 1.4%  |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.150          | iris coloboma  | 1         | 0.1%  | 0         |       |
| 93.710          | persistent pupillary membranes, iris to iris                   | 40        | 3.7%  | 21        | 10.0% |
| 93.730          | persistent pupillary membranes, iris to cornea                 | 2         | 0.2%  | 0         |       |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands   | 2         | 0.2%  | 1         | 0.5%  |
| 93.760          | persistent pupillary membranes, endothelial opacity/no strands | 1         | 0.1%  | 0         |       |
| 93.999          | uveal cysts  | 2         | 0.2%  | 0         |       |
| <b>LENS</b>     |  |           |       |           |       |
| 100.200         | cataract, unspecified  | 3         | 0.3%  | 0         |       |
| 100.210         | cataract. suspect not inherited/significance unknown           | 97        | 9.0%  | 21        | 10.0% |
| 100.301         | punctate cataract, anterior cortex                             | 13        | 1.2%  | 6         | 2.9%  |
| 100.302         | punctate cataract, posterior cortex                            | 9         | 0.8%  | 1         | 0.5%  |
| 100.303         | punctate cataract, equatorial cortex                           | 4         | 0.4%  | 0         |       |
| 100.305         | punctate cataract, posterior sutures                           | 1         | 0.1%  | 0         |       |
| 100.306         | punctate cataract, nucleus                                     | 0         |       | 1         | 0.5%  |
| 100.311         | incipient cataract, anterior cortex                            | 14        | 1.3%  | 1         | 0.5%  |
| 100.312         | incipient cataract, posterior cortex                           | 23        | 2.1%  | 0         |       |
| 100.313         | incipient cataract, equatorial cortex                          | 10        | 0.9%  | 1         | 0.5%  |
| 100.314         | incipient cataract, anterior sutures                           | 2         | 0.2%  | 0         |       |
| 100.315         | incipient cataract, posterior sutures                          | 2         | 0.2%  | 0         |       |
| 100.316         | incipient cataract, nucleus                                    | 5         | 0.5%  | 1         | 0.5%  |
| 100.317         | incipient cataract, capsular                                   | 5         | 0.5%  | 0         |       |
| 100.326         | incomplete cataract, nucleus                                   | 1         | 0.1%  | 0         |       |
| 100.330         | generalized/complete cataract                                  | 1         | 0.1%  | 0         |       |
| 100.345         | <i>significant cataracts (summary)</i>                         | 93        | 8.6%  | 11        | 5.3%  |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.120         | persistent hyaloid artery/remnant                              | 2         | 0.2%  | 3         | 1.4%  |
| 110.320         | vitreal degeneration   | 2         | 0.2%  | 0         |       |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                                       | 5         | 0.5%  | 1         | 0.5%  |
| 120.180         | retinal dysplasia, geographic                                  | 1         | 0.1%  | 0         |       |
| 120.310         | generalized progressive retinal atrophy (PRA)                  | 5         | 0.5%  | 0         |       |
| 120.910         | retinal detachment without dialysis                            | 1         | 0.1%  | 0         |       |
| 120.920         | retinal detachment with dialysis                               | 0         |       | 2         | 1.0%  |
| 120.960         | retinopathy  | 2         | 0.2%  | 1         | 0.5%  |



|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 20 1.9%   | 0         |
| 900.100 other, not inherited                              | 15 1.4%   | 0         |
| 900.110 other. suspect not inherited/significance unknown | 12 1.1%   | 7 3.3%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 742 68.6% | 109 52.2% |

# IRISH WOLFHOUND

|    | DISORDER                                      | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|---|-------------|-----------|-----------------|
| A. | Distichiasis                                  | Not defined | 1         | Breeder option  |
| B. | Nictitans cartilage anomaly/eversion          | Not defined | 1         | Breeder option  |
| C. | Corneal dystrophy - epithelial/stromal        | Not defined | 1         | Breeder option  |
| D. | Persistent pupillary membranes - iris to iris | Not defined | 1         | Breeder option  |
| E. | Uveal cysts                                   | Not defined | 1         | Breeder option  |
| F. | Cataract                                      | Not defined | 1         | NO              |
| G. | Retinal dysplasia - folds                     | Not defined | 1         | Breeder option  |
| H. | Optic nerve hypoplasia                        | Not defined | 1         | NO              |
| I. | Micropapilla                                  | Not defined | 1         | Breeder option  |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Nictitans cartilage anomaly/eversion

A scroll-like curling of the cartilage of the third eyelid, usually everting the margin. This condition may occur in one or both eyes and may cause mild ocular irritation.

### C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally in the neonatal period. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

E. Uveal cysts

Fluid filled sacs arising from the posterior surface of the iris, to which they may remain attached or break free and float into the anterior chamber. Usually occur in mature dogs.

F. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

G. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

H. Optic nerve hypoplasia

A congenital defect of the optic nerve which causes blindness and abnormal pupil response in the affected eye. May be unable to differentiate from micropapilla on a routine (dilated) screening ophthalmoscopic exam.

I. Micropapilla

A congenital anomaly which results in a small optic disk diameter without vision loss. Contrast with optic nerve hypoplasia, which may have a similar ophthalmoscopic appearance with vision loss.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT IRISH WOLFHOUND

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 0.110 microphthalmia  |                     | 1         | 0.1% | 0         |      |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 20.140 ectopic cilia  |                     | 1         | 0.1% | 0         |      |
| 21.000 entropion, unspecified   |                     | 6         | 0.3% | 0         |      |
| 25.110 distichiasis   |                     | 87        | 4.9% | 29        | 5.4% |
| <b>NASOLACRIMAL</b>   |                     |           |      |           |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 0         |      | 1         | 0.2% |
| <b>NICTITANS</b>  |                     |           |      |           |      |
| 50.210 pannus of third eyelid   |                     | 1         | 0.1% | 0         |      |
| 51.100 third eyelid cartilage anomaly                                 |                     | 17        | 1.0% | 8         | 1.5% |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.220 pigmentary keratitis   |                     | 1         | 0.1% | 0         |      |
| 70.700 corneal dystrophy  |                     | 38        | 2.1% | 3         | 0.6% |
| 70.730 corneal endothelial degeneration                               |                     | 2         | 0.1% | 0         |      |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 19        | 1.1% | 8         | 1.5% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 7         | 0.4% | 0         |      |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 11        | 0.6% | 0         |      |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 5         | 0.3% | 0         |      |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 2         | 0.1% | 0         |      |
| 93.810 uveal melanoma   |                     | 1         | 0.1% | 0         |      |
| 93.999 uveal cysts  |                     | 92        | 5.2% | 48        | 8.9% |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.200 cataract, unspecified   |                     | 12        | 0.7% | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 80        | 4.5% | 35        | 6.5% |
| 100.301 punctate cataract, anterior cortex                            |                     | 10        | 0.6% | 2         | 0.4% |
| 100.302 punctate cataract, posterior cortex                           |                     | 21        | 1.2% | 5         | 0.9% |
| 100.303 punctate cataract, equatorial cortex                          |                     | 2         | 0.1% | 2         | 0.4% |
| 100.304 punctate cataract, anterior sutures                           |                     | 1         | 0.1% | 0         |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 8         | 0.5% | 0         |      |
| 100.306 punctate cataract, nucleus                                    |                     | 5         | 0.3% | 1         | 0.2% |
| 100.307 punctate cataract, capsular                                   |                     | 3         | 0.2% | 2         | 0.4% |
| 100.311 incipient cataract, anterior cortex                           |                     | 11        | 0.6% | 3         | 0.6% |
| 100.312 incipient cataract, posterior cortex                          |                     | 33        | 1.9% | 9         | 1.7% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 8         | 0.5% | 2         | 0.4% |
| 100.314 incipient cataract, anterior sutures                          |                     | 1         | 0.1% | 0         |      |
| 100.315 incipient cataract, posterior sutures                         |                     | 10        | 0.6% | 4         | 0.7% |
| 100.316 incipient cataract, nucleus                                   |                     | 9         | 0.5% | 2         | 0.4% |
| 100.317 incipient cataract, capsular                                  |                     | 1         | 0.1% | 3         | 0.6% |
| 100.322 incomplete cataract, posterior cortex                         |                     | 1         | 0.1% | 1         | 0.2% |
| 100.323 incomplete cataract, equatorial cortex                        |                     | 0         |      | 1         | 0.2% |
| 100.326 incomplete cataract, nucleus                                  |                     | 1         | 0.1% | 0         |      |
| 100.330 generalized/complete cataract                                 |                     | 4         | 0.2% | 1         | 0.2% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.345 <i>significant cataracts (summary)</i>            | 141 7.9%         | 38 7.0%          |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 5 0.3%           | 2 0.4%           |
| 110.320 vitreal degeneration                              | 6 0.3%           | 2 0.4%           |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 25 1.4%          | 8 1.5%           |
| 120.180 retinal dysplasia, geographic                     | 11 0.6%          | 0                |
| 120.190 retinal dysplasia, detached                       | 2 0.1%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 2 0.1%           | 0                |
| 120.400 retinal hemorrhage                                | 1 0.1%           | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.1%           | 0                |
| 120.960 retinopathy                                       | 1 0.1%           | 1 0.2%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 12 0.7%          | 7 1.3%           |
| 130.120 optic nerve hypoplasia                            | 26 1.5%          | 3 0.6%           |
| 130.150 optic disc coloboma                               | 2 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 22 1.2%          | 0                |
| 900.100 other, not inherited                              | 59 3.3%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 34 1.9%          | 28 5.2%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,354 76.3%      | 369 68.3%        |

# ITALIAN GREYHOUND

|    | DISORDER                              | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE                           |
|----|---------------------------------------|------------------------|-----------|--------------------|--|
| A. | Cataract                              | Not defined            | 1         | NO                 |  |
| B. | Vitreous degeneration                 | Not defined            | 1, 2      | Breeder option     |  |
| C. | Retinal atrophy<br>( <i>IG-PRA1</i> ) | Autosomal<br>recessive | 1, 3      | NO                 | A genetic test for<br>susceptibility is<br>available |

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## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Italian Greyhound, posterior subcapsular and cortical cataracts at two to three years of age appear to be the more common location of occurrence, with progression noted in an undetermined percentage of dogs.

### B. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

### C. Retinal atrophy - *IG-PRA1*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Italian Greyhound PRA (*IG-PRA1*) is considered a "late onset" PRA with clinical signs detected between 3-5 years of age. Dogs initially lose night vision followed by decreased vision in bright light conditions. Clinically increases in tapetal reflectivity and retinal vessel attenuation are noted. The risk allele is known, but the genetic mutation has not been determined. The disease has been presumed to be inherited as an autosomal recessive trait. However some affected dogs had only one copy of the risk allele suggesting an autosomal dominant with incomplete penetrance mode of inheritance. A DNA test is available for the risk allele. At least one other form of PRA appears to be present in the

breed and will not be detected with this test.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Krishnan, H., et al. (2020). "Vitreous degeneration and associated ocular abnormalities in the dog." Vet Ophthalmol 23(2): 219-224. PMID: 31464365
3. Goldstein O, Pearce-Kelling, SE, Aguirre GD, Acland GM. Adult onset autosomal recessive hereditary retinal degeneration in Italian Greyhound dogs. *IOVS*, April 2011, Vol 52, 4351.

# OCULAR DISORDERS REPORT ITALIAN GREYHOUND

| Diagnostic Name   | TOTAL DOGS EXAMINED |      | 1991-2015<br>7,573 |      | 2016-2020<br>646 |   |
|---|---------------------|------|--------------------|------|------------------|---|
|   | #                   | %    | #                  | %    | #                | % |
| <b>GLOBE</b>  |                     |      |                    |      |                  |   |
| 0.110 microphthalmia  | 1                   | 0.0% | 1                  | 0.2% |                  |   |
| <b>EYELIDS</b>  |                     |      |                    |      |                  |   |
| 21.000 entropion, unspecified   | 0                   |      | 1                  | 0.2% |                  |   |
| 25.110 distichiasis   | 20                  | 0.3% | 3                  | 0.5% |                  |   |
| <b>NASOLACRIMAL</b>   |                     |      |                    |      |                  |   |
| 32.110 imperforate lower nasolacrimal punctum                         | 3                   | 0.0% | 6                  | 0.9% |                  |   |
| <b>CORNEA</b>   |                     |      |                    |      |                  |   |
| 70.210 corneal pannus   | 7                   | 0.1% | 0                  |      |                  |   |
| 70.220 pigmentary keratitis   | 2                   | 0.0% | 0                  |      |                  |   |
| 70.700 corneal dystrophy  | 19                  | 0.3% | 2                  | 0.3% |                  |   |
| <b>UVEA</b>   |                     |      |                    |      |                  |   |
| 93.110 iris hypoplasia  | 1                   | 0.0% | 0                  |      |                  |   |
| 93.140 corneal endothelial pigment without PPM                        | 3                   | 0.0% | 0                  |      |                  |   |
| 93.150 iris coloboma  | 6                   | 0.1% | 0                  |      |                  |   |
| 93.710 persistent pupillary membranes, iris to iris                   | 51                  | 0.7% | 3                  | 0.5% |                  |   |
| 93.720 persistent pupillary membranes, iris to lens                   | 6                   | 0.1% | 0                  |      |                  |   |
| 93.730 persistent pupillary membranes, iris to cornea                 | 5                   | 0.1% | 0                  |      |                  |   |
| 93.740 persistent pupillary membranes, iris sheets                    | 5                   | 0.1% | 0                  |      |                  |   |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   | 7                   | 0.1% | 1                  | 0.2% |                  |   |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands | 4                   | 0.1% | 1                  | 0.2% |                  |   |
| 93.999 uveal cysts  | 2                   | 0.0% | 1                  | 0.2% |                  |   |
| <b>LENS</b>   |                     |      |                    |      |                  |   |
| 100.200 cataract, unspecified   | 17                  | 0.2% | 0                  |      |                  |   |
| 100.210 cataract. suspect not inherited/significance unknown          | 327                 | 4.3% | 38                 | 5.9% |                  |   |
| 100.301 punctate cataract, anterior cortex                            | 88                  | 1.2% | 11                 | 1.7% |                  |   |
| 100.302 punctate cataract, posterior cortex                           | 85                  | 1.1% | 1                  | 0.2% |                  |   |
| 100.303 punctate cataract, equatorial cortex                          | 26                  | 0.3% | 3                  | 0.5% |                  |   |
| 100.304 punctate cataract, anterior sutures                           | 5                   | 0.1% | 0                  |      |                  |   |
| 100.305 punctate cataract, posterior sutures                          | 16                  | 0.2% | 2                  | 0.3% |                  |   |
| 100.306 punctate cataract, nucleus                                    | 7                   | 0.1% | 0                  |      |                  |   |
| 100.307 punctate cataract, capsular                                   | 11                  | 0.1% | 1                  | 0.2% |                  |   |
| 100.311 incipient cataract, anterior cortex                           | 172                 | 2.3% | 12                 | 1.9% |                  |   |
| 100.312 incipient cataract, posterior cortex                          | 170                 | 2.2% | 16                 | 2.5% |                  |   |
| 100.313 incipient cataract, equatorial cortex                         | 98                  | 1.3% | 7                  | 1.1% |                  |   |
| 100.314 incipient cataract, anterior sutures                          | 7                   | 0.1% | 1                  | 0.2% |                  |   |
| 100.315 incipient cataract, posterior sutures                         | 16                  | 0.2% | 2                  | 0.3% |                  |   |
| 100.316 incipient cataract, nucleus                                   | 14                  | 0.2% | 1                  | 0.2% |                  |   |
| 100.317 incipient cataract, capsular                                  | 16                  | 0.2% | 3                  | 0.5% |                  |   |
| 100.321 incomplete cataract, anterior cortex                          | 7                   | 0.1% | 4                  | 0.6% |                  |   |
| 100.322 incomplete cataract, posterior cortex                         | 7                   | 0.1% | 7                  | 1.1% |                  |   |
| 100.323 incomplete cataract, equatorial cortex                        | 3                   | 0.0% | 2                  | 0.3% |                  |   |
| 100.324 incomplete cataract, anterior sutures                         | 0                   |      | 1                  | 0.2% |                  |   |
| 100.326 incomplete cataract, nucleus                                  | 1                   | 0.0% | 0                  |      |                  |   |
| 100.328 y-suture tip opacities  | 0                   |      | 2                  | 0.3% |                  |   |



| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.330 generalized/complete cataract                     | 49 0.6%          | 1 0.2%           |
| 100.345 significant cataracts (summary)                   | 815 10.8%        | 77 11.9%         |
| 100.375 subluxation/luxation, unspecified                 | 36 0.5%          | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 22 0.3%          | 2 0.3%           |
| 110.135 PHPV/PTVL   | 3 0.0%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 168 2.2%         | 131 20.3%        |
| 110.320 vitreal degeneration                              | 2,493 32.9%      | 107 16.6%        |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 22 0.3%          | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 24 0.3%          | 2 0.3%           |
| 120.180 retinal dysplasia, geographic                     | 4 0.1%           | 0                |
| 120.190 retinal dysplasia, detached                       | 1 0.0%           | 1 0.2%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 244 3.2%         | 7 1.1%           |
| 120.400 retinal hemorrhage                                | 19 0.3%          | 0                |
| 120.910 retinal detachment without dialysis               | 8 0.1%           | 0                |
| 120.920 retinal detachment with dialysis                  | 1 0.0%           | 2 0.3%           |
| 120.960 retinopathy                                       | 6 0.1%           | 3 0.5%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 20 0.3%          | 2 0.3%           |
| 130.120 optic nerve hypoplasia                            | 34 0.4%          | 3 0.5%           |
| 130.150 optic disc coloboma                               | 4 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 63 0.8%          | 0                |
| 900.100 other, not inherited                              | 138 1.8%         | 5 0.8%           |
| 900.110 other. suspect not inherited/significance unknown | 89 1.2%          | 35 5.4%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 4,967 65.6%      | 379 58.7%        |

# JACK RUSSELL TERRIER

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|---|------------------------|-----------|--------------------|---|
| A. | Distichiasis  | Not defined            | 1         | Breeder option     |   |
| B. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined            | 1         | Breeder option     |   |
| C. | Cataract  | Not defined            | 1         | NO                 |   |
| D. | Lens luxation                                       | Autosomal<br>recessive | 1, 2-7    | NO                 | Mutation of the<br><i>ADAMTS17</i> gene |
| E. | Retinal atrophy<br>( <i>prcd</i> )                  | Autosomal<br>recessive | 1         | NO                 | Mutation of the <i>prcd</i><br>gene     |
| F. | Vitreous<br>degeneration                            | Not defined            | 1, 2      | Breeder option     |   |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens

completely (diffuse) or in a localized region.

D. Lens luxation

Partial (subluxation) or complete displacement of the lens from its normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

E. Retinal Atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that one form of PRA in the Jack Russell Terrier is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

F. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Lawson DD. Luxation of the crystalline lens in the dog. *J Small Anim Pract.* 1969;10:461-463.
3. Curtis R, Barnett KC. Primary lens luxation in the dog. *J Small Anim Pract.* 1980;21:657-668.
4. Curtis R, Barnett KC, Lewis SJ. Clinical and pathological observations concerning the aetiology of primary lens luxation in the dog. *Vet Rec.* 1983;112:238-246.
5. Oberbauer AM, Hollingsworth SR, Belanger JM, et al. Inheritance of cataracts and primary lens luxation in Jack Russell Terriers. *Am J Vet Res.* 2008;69:222-227.
6. Farias FH, Johnson GS, Taylor JF, et al. An *ADAMTS17* splice donor site mutation in dogs with primary lens luxation. *Invest Ophthalmol Vis Sci.* 2010;51:4716-4721.

7. Gould D, Pettitt L, McLaughlin B, et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011;14:378-384.

# OCULAR DISORDERS REPORT

## JACK RUSSELL TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>15,323 |      | 2016-2020<br>1,604 |      |
|---------------------|--|---------------------|------|--------------------|------|
|                     |  | #                   | %    | #                  | %    |
| <b>GLOBE</b>        |  |                     |      |                    |      |
| 0.110               | microphthalmia   | 5                   | 0.0% | 0                  |      |
| 10.000              | glaucoma   | 3                   | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                     |      |                    |      |
| 20.140              | ectopic cilia  | 2                   | 0.0% | 0                  |      |
| 20.160              | macropalpebral fissure   | 1                   | 0.0% | 0                  |      |
| 21.000              | entropion, unspecified   | 3                   | 0.0% | 0                  |      |
| 25.110              | distichiasis   | 354                 | 2.3% | 25                 | 1.6% |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |      |
| 40.910              | keratoconjunctivitis sicca                                     | 2                   | 0.0% | 1                  | 0.1% |
| <b>NICTITANS</b>    |  |                     |      |                    |      |
| 52.110              | prolapsed gland of the third eyelid                            | 1                   | 0.0% | 0                  |      |
| <b>CORNEA</b>       |  |                     |      |                    |      |
| 70.210              | corneal pannus   | 1                   | 0.0% | 0                  |      |
| 70.220              | pigmentary keratitis   | 9                   | 0.1% | 0                  |      |
| 70.700              | corneal dystrophy  | 59                  | 0.4% | 9                  | 0.6% |
| 70.730              | corneal endothelial degeneration                               | 9                   | 0.1% | 2                  | 0.1% |
| <b>UVEA</b>         |  |                     |      |                    |      |
| 93.140              | corneal endothelial pigment without PPM                        | 1                   | 0.0% | 0                  |      |
| 93.150              | iris coloboma  | 4                   | 0.0% | 0                  |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 694                 | 4.5% | 64                 | 4.0% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 40                  | 0.3% | 1                  | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 18                  | 0.1% | 1                  | 0.1% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 10                  | 0.1% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 11                  | 0.1% | 6                  | 0.4% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 6                   | 0.0% | 0                  |      |
| 93.999              | uveal cysts  | 7                   | 0.0% | 0                  |      |
| <b>LENS</b>         |  |                     |      |                    |      |
| 100.200             | cataract, unspecified  | 4                   | 0.0% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 534                 | 3.5% | 50                 | 3.1% |
| 100.301             | punctate cataract, anterior cortex                             | 73                  | 0.5% | 13                 | 0.8% |
| 100.302             | punctate cataract, posterior cortex                            | 78                  | 0.5% | 9                  | 0.6% |
| 100.303             | punctate cataract, equatorial cortex                           | 21                  | 0.1% | 2                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 14                  | 0.1% | 1                  | 0.1% |
| 100.305             | punctate cataract, posterior sutures                           | 49                  | 0.3% | 8                  | 0.5% |
| 100.306             | punctate cataract, nucleus                                     | 19                  | 0.1% | 4                  | 0.2% |
| 100.307             | punctate cataract, capsular                                    | 18                  | 0.1% | 4                  | 0.2% |
| 100.311             | incipient cataract, anterior cortex                            | 184                 | 1.2% | 12                 | 0.7% |
| 100.312             | incipient cataract, posterior cortex                           | 373                 | 2.4% | 26                 | 1.6% |
| 100.313             | incipient cataract, equatorial cortex                          | 64                  | 0.4% | 8                  | 0.5% |
| 100.314             | incipient cataract, anterior sutures                           | 8                   | 0.1% | 0                  |      |
| 100.315             | incipient cataract, posterior sutures                          | 133                 | 0.9% | 16                 | 1.0% |
| 100.316             | incipient cataract, nucleus                                    | 30                  | 0.2% | 2                  | 0.1% |
| 100.317             | incipient cataract, capsular                                   | 26                  | 0.2% | 2                  | 0.1% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.321 incomplete cataract, anterior cortex              | 1                | 0.0%  | 3                | 0.2%  |
| 100.322 incomplete cataract, posterior cortex             | 6                | 0.0%  | 7                | 0.4%  |
| 100.323 incomplete cataract, equatorial cortex            | 0                |       | 1                | 0.1%  |
| 100.325 incomplete cataract, posterior sutures            | 0                |       | 1                | 0.1%  |
| 100.328 y-suture tip opacities                            | 5                | 0.0%  | 5                | 0.3%  |
| 100.330 generalized/complete cataract                     | 94               | 0.6%  | 3                | 0.2%  |
| 100.345 <i>significant cataracts (summary)</i>            | 1,200            | 7.8%  | 127              | 7.9%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 81               | 0.5%  | 2                | 0.1%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 18               | 0.1%  | 3                | 0.2%  |
| 110.135 PHPV/PTVL   | 4                | 0.0%  | 0                |       |
| 110.200 vitreous degeneration-anterior chamber            | 1                | 0.0%  | 5                | 0.3%  |
| 110.320 vitreal degeneration                              | 232              | 1.5%  | 15               | 0.9%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.120 coloboma   | 2                | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 58               | 0.4%  | 1                | 0.1%  |
| 120.180 retinal dysplasia, geographic                     | 20               | 0.1%  | 0                |       |
| 120.190 retinal dysplasia, detached                       | 4                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 84               | 0.5%  | 1                | 0.1%  |
| 120.400 retinal hemorrhage                                | 4                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 8                | 0.1%  | 0                |       |
| 120.960 retinopathy                                       | 2                | 0.0%  | 1                | 0.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 7                | 0.0%  | 0                |       |
| 130.120 optic nerve hypoplasia                            | 12               | 0.1%  | 1                | 0.1%  |
| 130.150 optic disc coloboma                               | 1                | 0.0%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 113              | 0.7%  | 0                |       |
| 900.100 other, not inherited                              | 645              | 4.2%  | 3                | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown | 112              | 0.7%  | 71               | 4.4%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 12,682           | 82.8% | 1,285            | 80.1% |

# JAGDTERRIER

|    | DISORDER      | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|---------------|------------------------|-----------|--------------------|---|
| A. | Lens luxation | Autosomal<br>recessive | 1         | NO                 | Mutation of the<br><i>ADAMTS17</i> gene |

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## Description and Comments

### A. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Jagdterrier. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011; 14: 378-384.

# OCULAR DISORDERS REPORT JAGDTERRIER

| Diagnostic Name                     | TOTAL DOGS EXAMINED | 1991-2015 |   | 2016-2020 |        |
|-------------------------------------|---------------------|-----------|---|-----------|--------|
|                                     |                     | #         | % | #         | %      |
| <b>NORMAL</b><br>0.000 normal globe |                     | 0         |   | 2         | 100.0% |



# JAMTHUND

(Swedish Elkhound)

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|------------------------------------|------------------------|-----------|--------------------|-------------------------------------|
| A. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 1         | NO                 | Mutation of the<br><i>prcd</i> gene |

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## Description and Comments

### A. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Jamthund is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Jamthund. The conditions listed above are currently noted solely due to the availability of a genetic test for the disease.

1. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006 Nov;88:551-563. PMID: 16938425

# OCULAR DISORDERS REPORT JAMTHUND

| Diagnostic Name                     | TOTAL DOGS EXAMINED | 1991-2015 |   | 2016-2020 |        |
|-------------------------------------|---------------------|-----------|---|-----------|--------|
|                                     |                     | #         | % | #         | %      |
| <b>NORMAL</b><br>0.000 normal globe |                     | 0         |   | 1         | 100.0% |

# JAPANESE AKITA

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or from sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Japanese Akita breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT JAPANESE AKITA

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 0         |       | 2         | 1.3%  |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.700          | corneal dystrophy  | 0         |       | 5         | 3.2%  |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 2         | 7.7%  | 19        | 12.3% |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 0         |       | 1         | 0.6%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 1         | 3.8%  | 10        | 6.5%  |
| 100.301         | punctate cataract, anterior cortex                           | 1         | 3.8%  | 1         | 0.6%  |
| 100.302         | punctate cataract, posterior cortex                          | 0         |       | 1         | 0.6%  |
| 100.305         | punctate cataract, posterior sutures                         | 0         |       | 3         | 1.9%  |
| 100.317         | incipient cataract, capsular                                 | 0         |       | 3         | 1.9%  |
| 100.328         | y-suture tip opacities                                       | 0         |       | 3         | 1.9%  |
| 100.345         | <i>significant cataracts (summary)</i>                       | 1         | 3.8%  | 11        | 7.1%  |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.120         | persistent hyaloid artery/remnant                            | 0         |       | 5         | 3.2%  |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                                     | 0         |       | 2         | 1.3%  |
| 120.920         | retinal detachment with dialysis                             | 1         | 3.8%  | 0         |       |
| 120.960         | retinopathy  | 0         |       | 2         | 1.3%  |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.100         | other, not inherited   | 1         | 3.8%  | 0         |       |
| 900.110         | other. suspect not inherited/significance unknown            | 1         | 3.8%  | 17        | 11.0% |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 20        | 76.9% | 102       | 65.8% |

## JAPANESE CHIN (JAPANESE SPANIEL)

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE       |
|----|--|---------------------|-----------|--------------------|----------------------------------|
| A. | Entropion  | Not defined         | 1         | Breeder option     |                                  |
| B. | Distichiasis                                     | Not defined         | 1         | Breeder option     |                                  |
| C. | Exposure keratopathy syndrome                    | Not defined         | 1         | Breeder option     |                                  |
| D. | Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option     |                                  |
| E. | Cataract   | Not defined         | 1         | NO                 |                                  |
| F. | Vitreous degeneration                            | Not defined         | 1         | Breeder option     |                                  |
| G. | Retinal atrophy ( <i>prcd</i> )                  | Autosomal recessive | 1, 2      | NO                 | Mutation of the <i>prcd</i> gene |

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### Description and Comments

#### A. Entropion

A conformational defect resulting in an "in-rolling" of one or more of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

#### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

#### C. Exposure keratopathy syndrome

A condition characterized by variable degrees of superficial vascularization, fibrosis and/or pigmentation of the cornea. May be associated with excessive exposure/irritation of the globe due to shallow orbits, lower eyelid medial entropion, lagophthalmos and

macropalpebral fissure.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or from sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

F. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

G. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that one form of PRA in the Japanese Chin is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Zangerl, B., et al. (2006). "Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans." Genomics 88(5): 551-563. PMID: 16938425

# OCULAR DISORDERS REPORT JAPANESE CHIN

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|---------------------|--|-----------|-------|-----------|-------|
|                     |  | #         | %     | #         | %     |
| <b>EYELIDS</b>      |  |           |       |           |       |
| 20.160              | macropalpebral fissure                                       | 13        | 1.2%  | 0         |       |
| 21.000              | entropion, unspecified                                       | 88        | 7.8%  | 21        | 7.2%  |
| 22.000              | ectropion, unspecified                                       | 1         | 0.1%  | 0         |       |
| 25.110              | distichiasis   | 55        | 4.9%  | 8         | 2.7%  |
| <b>NASOLACRIMAL</b> |  |           |       |           |       |
| 32.110              | imperforate lower nasolacrimal punctum                       | 1         | 0.1%  | 0         |       |
| 40.910              | keratoconjunctivitis sicca                                   | 1         | 0.1%  | 2         | 0.7%  |
| <b>NICTITANS</b>    |  |           |       |           |       |
| 52.110              | prolapsed gland of the third eyelid                          | 2         | 0.2%  | 0         |       |
| <b>CORNEA</b>       |  |           |       |           |       |
| 70.210              | corneal pannus   | 9         | 0.8%  | 2         | 0.7%  |
| 70.220              | pigmentary keratitis   | 44        | 3.9%  | 7         | 2.4%  |
| 70.700              | corneal dystrophy  | 2         | 0.2%  | 1         | 0.3%  |
| 70.730              | corneal endothelial degeneration                             | 2         | 0.2%  | 2         | 0.7%  |
| <b>UVEA</b>         |  |           |       |           |       |
| 93.150              | iris coloboma  | 1         | 0.1%  | 1         | 0.3%  |
| 93.710              | persistent pupillary membranes, iris to iris                 | 121       | 10.8% | 25        | 8.6%  |
| 93.720              | persistent pupillary membranes, iris to lens                 | 6         | 0.5%  | 0         |       |
| 93.730              | persistent pupillary membranes, iris to cornea               | 7         | 0.6%  | 0         |       |
| 93.740              | persistent pupillary membranes, iris sheets                  | 6         | 0.5%  | 0         |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands | 1         | 0.1%  | 0         |       |
| 93.999              | uveal cysts  | 0         |       | 1         | 0.3%  |
| <b>LENS</b>         |  |           |       |           |       |
| 100.200             | cataract, unspecified  | 1         | 0.1%  | 0         |       |
| 100.210             | cataract. suspect not inherited/significance unknown         | 53        | 4.7%  | 13        | 4.5%  |
| 100.301             | punctate cataract, anterior cortex                           | 17        | 1.5%  | 5         | 1.7%  |
| 100.302             | punctate cataract, posterior cortex                          | 8         | 0.7%  | 0         |       |
| 100.303             | punctate cataract, equatorial cortex                         | 6         | 0.5%  | 1         | 0.3%  |
| 100.304             | punctate cataract, anterior sutures                          | 4         | 0.4%  | 3         | 1.0%  |
| 100.305             | punctate cataract, posterior sutures                         | 4         | 0.4%  | 2         | 0.7%  |
| 100.306             | punctate cataract, nucleus                                   | 1         | 0.1%  | 1         | 0.3%  |
| 100.307             | punctate cataract, capsular                                  | 2         | 0.2%  | 0         |       |
| 100.311             | incipient cataract, anterior cortex                          | 37        | 3.3%  | 12        | 4.1%  |
| 100.312             | incipient cataract, posterior cortex                         | 24        | 2.1%  | 7         | 2.4%  |
| 100.313             | incipient cataract, equatorial cortex                        | 23        | 2.0%  | 5         | 1.7%  |
| 100.314             | incipient cataract, anterior sutures                         | 1         | 0.1%  | 1         | 0.3%  |
| 100.315             | incipient cataract, posterior sutures                        | 7         | 0.6%  | 1         | 0.3%  |
| 100.316             | incipient cataract, nucleus                                  | 4         | 0.4%  | 3         | 1.0%  |
| 100.317             | incipient cataract, capsular                                 | 11        | 1.0%  | 2         | 0.7%  |
| 100.321             | incomplete cataract, anterior cortex                         | 3         | 0.3%  | 4         | 1.4%  |
| 100.328             | y-suture tip opacities                                       | 0         |       | 1         | 0.3%  |
| 100.330             | generalized/complete cataract                                | 7         | 0.6%  | 1         | 0.3%  |
| 100.345             | significant cataracts (summary)                              | 160       | 14.2% | 49        | 16.8% |
| 100.375             | subluxation/luxation, unspecified                            | 6         | 0.5%  | 0         |       |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>VITREOUS</b>   |           |           |
| 110.120 persistent hyaloid artery/remnant                 | 15 1.3%   | 4 1.4%    |
| 110.135 PHPV/PTVL   | 13 1.2%   | 0         |
| 110.200 vitreous degeneration-anterior chamber            | 4 0.4%    | 1 0.3%    |
| 110.320 vitreal degeneration                              | 50 4.5%   | 15 5.2%   |
| <b>FUNDUS</b>   |           |           |
| 97.120 coloboma   | 1 0.1%    | 0         |
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 2 0.2%    | 1 0.3%    |
| 120.180 retinal dysplasia, geographic                     | 2 0.2%    | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 15 1.3%   | 1 0.3%    |
| 120.910 retinal detachment without dialysis               | 1 0.1%    | 0         |
| 120.920 retinal detachment with dialysis                  | 1 0.1%    | 0         |
| <b>OPTIC NERVE</b>  |           |           |
| 130.110 micropapilla                                      | 1 0.1%    | 0         |
| 130.150 optic disc coloboma                               | 2 0.2%    | 0         |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 28 2.5%   | 0         |
| 900.100 other, not inherited                              | 43 3.8%   | 3 1.0%    |
| 900.110 other. suspect not inherited/significance unknown | 33 2.9%   | 28 9.6%   |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 726 64.6% | 153 52.6% |



# KAI KEN

|    | <b>DISORDER</b>  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b>  |
|----|--|--------------------|------------------|-------------------------|
| A. | Persistent pupillary membranes<br>- lens pigment foci/no strands | Not defined        | 1                | Passes with no notation |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Kai Ken breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT KAI KEN

| TOTAL DOGS EXAMINED   | 1991-2015<br>2 |       | 2016-2020<br>28 |       |
|---|----------------|-------|-----------------|-------|
| Diagnostic Name   | #              | %     | #               | %     |
| <b>UVEA</b>   |                |       |                 |       |
| 93.710 persistent pupillary membranes, iris to iris                 | 0              |       | 1               | 3.6%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands | 1              | 50.0% | 7               | 25.0% |
| <b>LENS</b>   |                |       |                 |       |
| 100.210 cataract. suspect not inherited/significance unknown        | 0              |       | 2               | 7.1%  |
| <b>RETINA</b>   |                |       |                 |       |
| 120.960 retinopathy   | 0              |       | 1               | 3.6%  |
| <b>NORMAL</b>   |                |       |                 |       |
| 0.000 normal globe  | 1              | 50.0% | 18              | 64.3% |

# KARELIAN BEAR DOG

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TEST<br>AVAILABLE           |
|----|------------------------------------|------------------------|-----------|--------------------|-------------------------------------|
| A. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 1-3       | NO                 | Mutation of the<br><i>prcd</i> gene |

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## Description and Comments

### A. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited. A genetic test is available to detect the progressive rod cone degeneration form of PRA caused by a mutation in the *prcd*-gene. A second form of PRA is also present in the Karelian Bear Dog for which the causative mutation is not yet known.

## References

1. ACVO Genetics Committee and Data from OFA All-Breeds Report.
2. Ahonen S, Lohi H, editors. Progressive retinal atrophy in the Karelian Bear Dog: A large animal model for retinitis pigmentosa. ARVO 2014 Annual Meeting; 2014; Orlando, FL. Program number: 3270.
3. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006;88:551-563. Epub 2006/08/30. PMID: 16938425

# OCULAR DISORDERS REPORT KARELIAN BEAR DOG

| Diagnostic Name | TOTAL DOGS EXAMINED                                  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 2         | 1.9%  | 0         |       |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.220          | pigmentary keratitis                                 | 0         |       | 1         | 7.7%  |
| 70.700          | corneal dystrophy                                    | 4         | 3.9%  | 1         | 7.7%  |
| 70.730          | corneal endothelial degeneration                     | 1         | 1.0%  | 0         |       |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris         | 10        | 9.7%  | 2         | 15.4% |
| 93.730          | persistent pupillary membranes, iris to cornea       | 3         | 2.9%  | 0         |       |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown | 1         | 1.0%  | 2         | 15.4% |
| 100.307         | punctate cataract, capsular                          | 2         | 1.9%  | 0         |       |
| 100.311         | incipient cataract, anterior cortex                  | 3         | 2.9%  | 0         |       |
| 100.312         | incipient cataract, posterior cortex                 | 4         | 3.9%  | 0         |       |
| 100.314         | incipient cataract, anterior sutures                 | 0         |       | 1         | 7.7%  |
| 100.317         | incipient cataract, capsular                         | 1         | 1.0%  | 1         | 7.7%  |
| 100.345         | <i>significant cataracts (summary)</i>               | 10        | 9.7%  | 2         | 15.4% |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                             | 4         | 3.9%  | 0         |       |
| 120.310         | generalized progressive retinal atrophy (PRA)        | 1         | 1.0%  | 0         |       |
| 120.960         | retinopathy  | 1         | 1.0%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.000         | other, unspecified                                   | 1         | 1.0%  | 0         |       |
| 900.100         | other, not inherited                                 | 1         | 1.0%  | 0         |       |
| 900.110         | other. suspect not inherited/significance unknown    | 0         |       | 2         | 15.4% |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 79        | 76.7% | 6         | 46.2% |

# KEESHOND

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Distichiasis                                     | Not defined        | 1                | Breeder option         |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |
| C. | Cataract   | Not defined        | 1                | NO                     |
| D. | Y-suture tip opacity                             | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located in the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

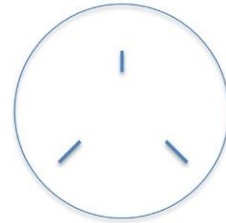
Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

#### D. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Keeshond breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT KEESHOND

| Diagnostic Name   | TOTAL DOGS EXAMINED |  | 1991-2015<br>3,174 |      | 2016-2020<br>634 |       |
|---|---------------------|--|--------------------|------|------------------|-------|
|   |                     |  | #                  | %    | #                | %     |
| <b>GLOBE</b>  |                     |  |                    |      |                  |       |
| 0.110 microphthalmia  |                     |  | 1                  | 0.0% | 0                |       |
| <b>EYELIDS</b>  |                     |  |                    |      |                  |       |
| 21.000 entropion, unspecified   |                     |  | 9                  | 0.3% | 0                |       |
| 25.110 distichiasis   |                     |  | 188                | 5.9% | 23               | 3.6%  |
| <b>NASOLACRIMAL</b>   |                     |  |                    |      |                  |       |
| 32.110 imperforate lower nasolacrimal punctum                         |                     |  | 1                  | 0.0% | 0                |       |
| <b>CORNEA</b>   |                     |  |                    |      |                  |       |
| 70.220 pigmentary keratitis   |                     |  | 0                  |      | 1                | 0.2%  |
| 70.700 corneal dystrophy  |                     |  | 12                 | 0.4% | 0                |       |
| 70.730 corneal endothelial degeneration                               |                     |  | 2                  | 0.1% | 0                |       |
| <b>UVEA</b>   |                     |  |                    |      |                  |       |
| 93.150 iris coloboma  |                     |  | 1                  | 0.0% | 0                |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     |  | 29                 | 0.9% | 3                | 0.5%  |
| 93.720 persistent pupillary membranes, iris to lens                   |                     |  | 2                  | 0.1% | 0                |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     |  | 2                  | 0.1% | 0                |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     |  | 2                  | 0.1% | 0                |       |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     |  | 1                  | 0.0% | 0                |       |
| 93.999 uveal cysts  |                     |  | 2                  | 0.1% | 0                |       |
| <b>LENS</b>   |                     |  |                    |      |                  |       |
| 100.200 cataract, unspecified   |                     |  | 18                 | 0.6% | 0                |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     |  | 274                | 8.6% | 79               | 12.5% |
| 100.301 punctate cataract, anterior cortex                            |                     |  | 12                 | 0.4% | 1                | 0.2%  |
| 100.302 punctate cataract, posterior cortex                           |                     |  | 16                 | 0.5% | 1                | 0.2%  |
| 100.303 punctate cataract, equatorial cortex                          |                     |  | 11                 | 0.3% | 0                |       |
| 100.304 punctate cataract, anterior sutures                           |                     |  | 2                  | 0.1% | 0                |       |
| 100.305 punctate cataract, posterior sutures                          |                     |  | 50                 | 1.6% | 25               | 3.9%  |
| 100.306 punctate cataract, nucleus                                    |                     |  | 1                  | 0.0% | 2                | 0.3%  |
| 100.307 punctate cataract, capsular                                   |                     |  | 1                  | 0.0% | 7                | 1.1%  |
| 100.311 incipient cataract, anterior cortex                           |                     |  | 7                  | 0.2% | 1                | 0.2%  |
| 100.312 incipient cataract, posterior cortex                          |                     |  | 34                 | 1.1% | 2                | 0.3%  |
| 100.313 incipient cataract, equatorial cortex                         |                     |  | 9                  | 0.3% | 0                |       |
| 100.314 incipient cataract, anterior sutures                          |                     |  | 1                  | 0.0% | 0                |       |
| 100.315 incipient cataract, posterior sutures                         |                     |  | 18                 | 0.6% | 7                | 1.1%  |
| 100.316 incipient cataract, nucleus                                   |                     |  | 13                 | 0.4% | 0                |       |
| 100.317 incipient cataract, capsular                                  |                     |  | 2                  | 0.1% | 3                | 0.5%  |
| 100.321 incomplete cataract, anterior cortex                          |                     |  | 0                  |      | 1                | 0.2%  |
| 100.325 incomplete cataract, posterior sutures                        |                     |  | 1                  | 0.0% | 0                |       |
| 100.326 incomplete cataract, nucleus                                  |                     |  | 1                  | 0.0% | 0                |       |
| 100.327 incomplete cataract, capsular                                 |                     |  | 1                  | 0.0% | 1                | 0.2%  |
| 100.328 y-suture tip opacities  |                     |  | 6                  | 0.2% | 47               | 7.4%  |
| 100.330 generalized/complete cataract                                 |                     |  | 7                  | 0.2% | 1                | 0.2%  |
| 100.345 <i>significant cataracts (summary)</i>                        |                     |  | 211                | 6.6% | 99               | 15.6% |
| 100.375 <i>subluxation/luxation, unspecified</i>                      |                     |  | 1                  | 0.0% | 0                |       |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>VITREOUS</b>   |             |           |
| 110.120 persistent hyaloid artery/remnant                 | 1 0.0%      | 1 0.2%    |
| 110.320 vitreal degeneration                              | 7 0.2%      | 4 0.6%    |
| <b>FUNDUS</b>   |             |           |
| 97.120 coloboma   | 1 0.0%      | 0         |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 6 0.2%      | 0         |
| 120.180 retinal dysplasia, geographic                     | 2 0.1%      | 0         |
| 120.190 retinal dysplasia, detached                       | 1 0.0%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 10 0.3%     | 1 0.2%    |
| 120.400 retinal hemorrhage                                | 1 0.0%      | 0         |
| 120.910 retinal detachment without dialysis               | 2 0.1%      | 0         |
| 120.960 retinopathy                                       | 3 0.1%      | 2 0.3%    |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 6 0.2%      | 3 0.5%    |
| 130.120 optic nerve hypoplasia                            | 12 0.4%     | 1 0.2%    |
| 130.150 optic disc coloboma                               | 1 0.0%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 21 0.7%     | 0         |
| 900.100 other, not inherited                              | 44 1.4%     | 3 0.5%    |
| 900.110 other. suspect not inherited/significance unknown | 29 0.9%     | 28 4.4%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 2,555 80.5% | 449 70.8% |



# KERRY BLUE TERRIER

|    | <b>DISORDER</b> | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|-----------------|--------------------|------------------|------------------------|
| A. | Cataract        | Not defined        | 1                | NO                     |

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## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Kerry Blue Terrier breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT

## KERRY BLUE TERRIER

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 11        | 1.5%  | 4         | 4.7%  |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.210          | corneal pannus   | 1         | 0.1%  | 0         |       |
| 70.700          | corneal dystrophy  | 3         | 0.4%  | 1         | 1.2%  |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 10        | 1.4%  | 4         | 4.7%  |
| 93.720          | persistent pupillary membranes, iris to lens                 | 2         | 0.3%  | 0         |       |
| 93.730          | persistent pupillary membranes, iris to cornea               | 1         | 0.1%  | 0         |       |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 0         |       | 2         | 2.3%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.200         | cataract, unspecified  | 6         | 0.8%  | 0         |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 28        | 3.8%  | 3         | 3.5%  |
| 100.301         | punctate cataract, anterior cortex                           | 15        | 2.1%  | 0         |       |
| 100.302         | punctate cataract, posterior cortex                          | 3         | 0.4%  | 0         |       |
| 100.306         | punctate cataract, nucleus                                   | 3         | 0.4%  | 0         |       |
| 100.312         | incipient cataract, posterior cortex                         | 4         | 0.5%  | 0         |       |
| 100.313         | incipient cataract, equatorial cortex                        | 3         | 0.4%  | 0         |       |
| 100.330         | generalized/complete cataract                                | 6         | 0.8%  | 0         |       |
| 100.345         | <i>significant cataracts (summary)</i>                       | 40        | 5.5%  | 0         |       |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.320         | vitreal degeneration   | 10        | 1.4%  | 0         |       |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.310         | generalized progressive retinal atrophy (PRA)                | 2         | 0.3%  | 1         | 1.2%  |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.000         | other, unspecified   | 1         | 0.1%  | 0         |       |
| 900.100         | other, not inherited   | 21        | 2.9%  | 0         |       |
| 900.110         | other. suspect not inherited/significance unknown            | 2         | 0.3%  | 3         | 3.5%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 647       | 88.5% | 72        | 83.7% |

# KOMONDOR

|    | <b>DISORDER</b> | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|-----------------|--------------------|------------------|------------------------|
| A. | Cataract        | Not defined        | 1                | NO                     |

---

## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

Appears to be relatively young age for onset in the Komondor (<4yr) and mainly anterior cortical.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Komondor breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT KOMONDOR

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|------------------|--|-----------|-------|-----------|-------|
|                  |  | #         | %     | #         | %     |
| <b>EYELIDS</b>   |  |           |       |           |       |
| 21.000           | entropion, unspecified   | 1         | 0.3%  | 0         |       |
| 22.000           | ectropion, unspecified   | 1         | 0.3%  | 0         |       |
| 25.110           | distichiasis   | 0         |       | 1         | 2.2%  |
| <b>NICTITANS</b> |  |           |       |           |       |
| 51.100           | third eyelid cartilage anomaly                                 | 1         | 0.3%  | 0         |       |
| <b>CORNEA</b>    |  |           |       |           |       |
| 70.700           | corneal dystrophy  | 1         | 0.3%  | 0         |       |
| <b>UVEA</b>      |  |           |       |           |       |
| 93.710           | persistent pupillary membranes, iris to iris                   | 5         | 1.5%  | 0         |       |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands | 2         | 0.6%  | 0         |       |
| <b>LENS</b>      |  |           |       |           |       |
| 100.200          | cataract, unspecified  | 14        | 4.1%  | 0         |       |
| 100.210          | cataract. suspect not inherited/significance unknown           | 26        | 7.6%  | 3         | 6.5%  |
| 100.303          | punctate cataract, equatorial cortex                           | 2         | 0.6%  | 0         |       |
| 100.306          | punctate cataract, nucleus                                     | 4         | 1.2%  | 0         |       |
| 100.307          | punctate cataract, capsular                                    | 2         | 0.6%  | 0         |       |
| 100.312          | incipient cataract, posterior cortex                           | 3         | 0.9%  | 1         | 2.2%  |
| 100.313          | incipient cataract, equatorial cortex                          | 5         | 1.5%  | 0         |       |
| 100.314          | incipient cataract, anterior sutures                           | 1         | 0.3%  | 0         |       |
| 100.315          | incipient cataract, posterior sutures                          | 3         | 0.9%  | 1         | 2.2%  |
| 100.316          | incipient cataract, nucleus                                    | 5         | 1.5%  | 0         |       |
| 100.326          | incomplete cataract, nucleus                                   | 1         | 0.3%  | 0         |       |
| 100.328          | y-suture tip opacities   | 0         |       | 1         | 2.2%  |
| 100.330          | generalized/complete cataract                                  | 1         | 0.3%  | 0         |       |
| 100.345          | <i>significant cataracts (summary)</i>                         | 41        | 12.0% | 3         | 6.5%  |
| <b>RETINA</b>    |  |           |       |           |       |
| 120.170          | retinal dysplasia, folds                                       | 1         | 0.3%  | 0         |       |
| <b>OTHER</b>     |  |           |       |           |       |
| 900.000          | other, unspecified   | 7         | 2.0%  | 0         |       |
| 900.100          | other, not inherited   | 6         | 1.8%  | 0         |       |
| 900.110          | other. suspect not inherited/significance unknown              | 1         | 0.3%  | 0         |       |
| <b>NORMAL</b>    |  |           |       |           |       |
| 0.000            | normal globe   | 280       | 81.9% | 41        | 89.1% |

# KUVASZ

|    | DISORDER                        | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|---------------------------------|------------------------|-----------|--------------------|-------------------------------------|
| A. | Retinal atrophy ( <i>prcd</i> ) | Autosomal<br>recessive | 1, 2      | NO                 | Mutation of the<br><i>prcd</i> gene |

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## Description and Comments

### A. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the form of PRA in the Kuvasz is *prcd*, which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

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# OCULAR DISORDERS REPORT KUVASZ

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|--|---------------------|-----------|-------|-----------|-------|
|  |                     | #         | %     | #         | %     |
| <b>GLOBE</b>   |                     |           |       |           |       |
| 0.110 microphthalmia   |                     | 2         | 0.4%  | 0         |       |
| <b>EYELIDS</b>   |                     |           |       |           |       |
| 20.140 ectopic cilia   |                     | 1         | 0.2%  | 0         |       |
| 20.160 macropalpebral fissure                                |                     | 1         | 0.2%  | 0         |       |
| 22.000 ectropion, unspecified                                |                     | 2         | 0.4%  | 0         |       |
| 25.110 distichiasis  |                     | 21        | 3.9%  | 0         |       |
| <b>NICTITANS</b>   |                     |           |       |           |       |
| 51.100 third eyelid cartilage anomaly                        |                     | 1         | 0.2%  | 0         |       |
| <b>CORNEA</b>  |                     |           |       |           |       |
| 70.700 corneal dystrophy                                     |                     | 6         | 1.1%  | 0         |       |
| 70.730 corneal endothelial degeneration                      |                     | 1         | 0.2%  | 0         |       |
| <b>UVEA</b>  |                     |           |       |           |       |
| 93.150 iris coloboma   |                     | 2         | 0.4%  | 0         |       |
| 93.710 persistent pupillary membranes, iris to iris          |                     | 23        | 4.2%  | 0         |       |
| 93.720 persistent pupillary membranes, iris to lens          |                     | 3         | 0.6%  | 0         |       |
| 93.730 persistent pupillary membranes, iris to cornea        |                     | 3         | 0.6%  | 0         |       |
| <b>LENS</b>  |                     |           |       |           |       |
| 100.200 cataract, unspecified                                |                     | 2         | 0.4%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 15        | 2.8%  | 1         | 8.3%  |
| 100.301 punctate cataract, anterior cortex                   |                     | 1         | 0.2%  | 0         |       |
| 100.302 punctate cataract, posterior cortex                  |                     | 1         | 0.2%  | 0         |       |
| 100.303 punctate cataract, equatorial cortex                 |                     | 1         | 0.2%  | 0         |       |
| 100.305 punctate cataract, posterior sutures                 |                     | 1         | 0.2%  | 0         |       |
| 100.312 incipient cataract, posterior cortex                 |                     | 1         | 0.2%  | 0         |       |
| 100.313 incipient cataract, equatorial cortex                |                     | 1         | 0.2%  | 0         |       |
| 100.316 incipient cataract, nucleus                          |                     | 3         | 0.6%  | 0         |       |
| 100.330 generalized/complete cataract                        |                     | 5         | 0.9%  | 0         |       |
| 100.345 <i>significant cataracts (summary)</i>               |                     | 16        | 3.0%  | 0         |       |
| <b>VITREOUS</b>  |                     |           |       |           |       |
| 110.320 vitreal degeneration                                 |                     | 1         | 0.2%  | 0         |       |
| <b>RETINA</b>  |                     |           |       |           |       |
| 120.310 generalized progressive retinal atrophy (PRA)        |                     | 4         | 0.7%  | 0         |       |
| <b>OTHER</b>   |                     |           |       |           |       |
| 900.000 other, unspecified                                   |                     | 1         | 0.2%  | 0         |       |
| 900.100 other, not inherited                                 |                     | 12        | 2.2%  | 0         |       |
| 900.110 other. suspect not inherited/significance unknown    |                     | 3         | 0.6%  | 1         | 8.3%  |
| <b>NORMAL</b>  |                     |           |       |           |       |
| 0.000 normal globe   |                     | 454       | 83.8% | 10        | 83.3% |

# LABRADOR RETRIEVER

|    | DISORDER   | INHERITANCE  | REFERENCE              | BREEDING<br>ADVICE                           | GENETIC TESTS<br>AVAILABLE  |
|----|--|--|------------------------|--|---|
| A. | Entropion  | Not defined  | 1-3                    | Breeder option                               |   |
| B. | Ectropion  | Not defined  | 1                      | Breeder option                               |   |
| C. | Distichiasis   | Not defined  | 1                      | Breeder option                               |   |
| D. | Corneal dystrophy<br>- epithelial/stromal<br>- macular                                   | Not defined<br>Autosomal<br>recessive  | 1, 4<br>5              | Breeder option<br>NO                         | Mutation of the<br><i>CHST6</i> gene  |
| E. | Uveal cysts  | Not defined  | 1                      | Breeder option                               |   |
| F. | Persistent pupillary<br>membranes<br>- iris to iris<br>- lens pigment foci/no<br>strands | Not defined<br>Not defined   | 1<br>1                 | Breeder option<br>Passes with<br>no notation |   |
| G. | Cataract   | Presumed<br>dominant with<br>incomplete<br>penetrance<br>Autosomal<br>recessive<br>Not defined | 1-3, 6-8<br><br>9<br>1 | NO<br><br>NO<br>NO                           |   |
| H. | Y-suture tip opacity   | Not defined  | 1                      | Breeder option                               |   |
| I. | Persistent hyaloid<br>artery   | Not defined  | 1                      | Breeder option                               |   |
| J. | Vitreous degeneration  | Not defined  | 1                      | Breeder option                               |   |
| K. | Retinal atrophy<br><br>- ( <i>prcd</i> )<br><br>- generalized                            | Autosomal<br>recessive   | 1, 10-14<br><br>15     | NO<br><br>NO                                 | Mutation of the<br><i>prcd</i> gene<br><br>Mutation of the<br><i>ABCA4</i> gene |

|    | <b>DISORDER</b>   | <b>INHERITANCE</b>   | <b>REFERENCE</b> | <b>BREEDING ADVICE</b>                                | <b>GENETIC TESTS AVAILABLE</b>       |
|----|---|--|------------------|---|--------------------------------------|
| L. | Achromatopsia Type 2 (ACHM - Type 2)                                  | Autosomal recessive  | 16, 17           | NO  | Causative mutation not yet published |
| M. | Retinal dysplasia - folds   | Presumed autosomal recessive                               | 1, 18-27         | NO<br>(Breeder option with Normal DNA test for folds) | Mutation in the COL9A3 gene          |
| N. | Retinal dysplasia - folds/geographic/detached (with skeletal defects) | Autosomal recessive with incomplete dominance for the eyes | 1, 18-26, 28     | NO  | Mutation in the COL9A3 gene          |
| O. | Limbal melanoma   | Not defined  | 29               | NO  |                                      |

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## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. Selection should be directed against entropion and toward a head conformation that reduces or eliminates the likelihood of the defect.

### B. Ectropion

A conformational defect resulting in eversion of the eyelid(s), which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### C. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.



D. Corneal dystrophy - epithelial/stromal/macular

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

In Labrador Retrievers in Europe, macular corneal dystrophy (MCD) has been shown to be caused by accumulations of glycosaminoglycans in the corneal stroma. This form of corneal dystrophy is caused by a mutation in the *CHST6* gene.

E. Uveal cysts

Fluid filled sacs arising from the posterior surface of the iris, to which they may remain attached or break free and float into the anterior chamber. Usually occur in mature dogs.

This disorder may be observed in any breed but retriever breeds tend to be predisposed. There is usually no effect on vision unless the cysts are heavily clustered and impinge on the pupillary area. Less frequently, the cysts may rupture and adhere to the cornea or anterior lens capsule. Multiple cysts may occlude the iridocorneal angle and cause glaucoma.

F. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

In the Labrador Retriever, this is a potentially serious problem as many of the PPMs identified on routine screening examinations bridge from the iris to the cornea and/or from iris sheets bridging the pupils. These forms may cause vision impairment.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

G. Cataract

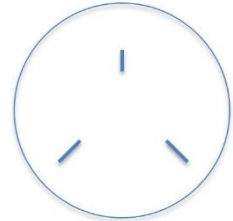
A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

The most frequently reported cataracts in the Labrador Retriever are bilateral or unilateral, focal, posterior polar (posterior cortical)/subcapsular cataracts which usually present between 1-3 years of age. These are generally stationary or very slowly progressive and generally do not interfere with vision. It has been suggested that these cataracts are inherited as dominant with incomplete penetrance, but definitive breeding studies are still required to verify this hypothesis.

A second type of cataract is a progressive cortical cataract which may involve the entire lens. It is not clear whether this is a distinct entity, or an aberrant form of the posterior polar cataract.

#### H. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless misdiagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

#### I. Persistent hyaloid artery (PHA)

A congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small vascular strand (PHA) or as a non-vascular strand that appears gray-white (**persistent hyaloid remnant**).

#### J. Vitreous degeneration

Liquefaction of the vitreous gel, which may predispose to retinal detachment.

#### K. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Labrador Retriever is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

L. Achromatopsia Type 2 (ACHM – Type 2)

A congenital form of day blindness. Visual deficits become apparent between 8-10 weeks of age. Normal vision is present in low light conditions. Clinical examination is normal. Cone responses are absent on an electroretinogram. The causative genetic mutation has been determined, but not yet published. A DNA test is available.

M. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness.

In the Labrador Retriever, the presence of retinal folds may be seen in the heterozygous state described in "R" below, thus the recommendation against breeding.

The breeding advice for Labrador Retrievers and Samoyeds diagnosed with "retinal dysplasia - folds" will be changed from "No" to "Breeder option" if the owner of the dog provides the registering office with results of the DNA test for the affected dog, showing that it is not a carrier of the *COL9A3* mutation.

N. Retinal dysplasia - folds or detachment with skeletal defects

This condition is also known as oculo-skeletal dysplasia (OSD) or dwarfism with retinal dysplasia type 1 (DRD1) in the Labrador Retriever. A similar condition, DRD2, occurs in the Samoyed. The condition is autosomal recessive and homozygous affected dogs have shortened forelimbs ("downhill" conformation) with valgus deformity. They have severe ocular defects including cataract, retinal folds/multifocal retinal dysplasia, vitreal degeneration and retinal detachment. The ocular abnormalities result in blindness in most dogs. Heterozygous dogs can have either a normal ocular exam or have multiple retinal folds, vitreal membranes, or vitreal degeneration suggesting a semi-dominant mechanism with respect to the eyes. It is important to note that generally the retinal folds present in heterozygous dogs tend to cluster around the major superior blood vessels of the central tapetal region. The condition is caused by a 1 base pair insertion of *COL9A3*. A DNA test is available.

O. Limbal melanoma

Most limbal melanomas are really epibulbar melanocytomas, but there is a possibility of an extension of an intraocular melanoma extending outward and presenting as a limbal melanoma. An epibulbar melanocytoma originates from the superficial pigment lining the limbus and the lesion may eventually extend into the eye. Metastasis has not been documented and the mass is characterized by large epithelioid cells. The lesion presents as a subconjunctival smooth mass most commonly in the dorsolateral limbal region and extends later into the cornea and posterior on the sclera. Breed predisposition have been noted in the German Shepherd, Labrador and Golden Retriever.

**Historical Note:**

Central progressive retinal atrophy was previously a condition listed for Labrador Retriever. However as the condition is no longer identified in the breed, the condition has been removed.

Central progressive retinal atrophy was a progressive retinal degeneration in which photoreceptor death occurred secondary to disease of the underlying pigment epithelium. Progression was slow and some animals never lost vision. CPRA occurred in England, but was uncommon elsewhere.

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# OCULAR DISORDERS REPORT LABRADOR RETRIEVER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>229,860 |      | 2016-2020<br>37,648 |      |
|---------------------|--|----------------------|------|---------------------|------|
|                     |  | #                    | %    | #                   | %    |
| <b>GLOBE</b>        |  |                      |      |                     |      |
| 0.110               | microphthalmia   | 59                   | 0.0% | 6                   | 0.0% |
| 10.000              | glaucoma   | 28                   | 0.0% | 2                   | 0.0% |
| <b>EYELIDS</b>      |  |                      |      |                     |      |
| 20.140              | ectopic cilia  | 16                   | 0.0% | 2                   | 0.0% |
| 20.160              | macropalpebral fissure   | 86                   | 0.0% | 0                   |      |
| 21.000              | entropion, unspecified   | 996                  | 0.4% | 185                 | 0.5% |
| 22.000              | ectropion, unspecified   | 485                  | 0.2% | 59                  | 0.2% |
| 25.110              | distichiasis   | 2,267                | 1.0% | 344                 | 0.9% |
| <b>NASOLACRIMAL</b> |  |                      |      |                     |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 22                   | 0.0% | 21                  | 0.1% |
| 40.910              | keratoconjunctivitis sicca                                     | 7                    | 0.0% | 3                   | 0.0% |
| <b>NICTITANS</b>    |  |                      |      |                     |      |
| 51.100              | third eyelid cartilage anomaly                                 | 11                   | 0.0% | 1                   | 0.0% |
| 52.110              | prolapsed gland of the third eyelid                            | 38                   | 0.0% | 1                   | 0.0% |
| <b>CORNEA</b>       |  |                      |      |                     |      |
| 70.210              | corneal pannus   | 9                    | 0.0% | 0                   |      |
| 70.220              | pigmentary keratitis   | 18                   | 0.0% | 7                   | 0.0% |
| 70.700              | corneal dystrophy  | 2,260                | 1.0% | 377                 | 1.0% |
| 70.730              | corneal endothelial degeneration                               | 80                   | 0.0% | 10                  | 0.0% |
| <b>UVEA</b>         |  |                      |      |                     |      |
| 90.250              | pigmentary uveitis   | 2                    | 0.0% | 0                   |      |
| 93.110              | iris hypoplasia  | 6                    | 0.0% | 1                   | 0.0% |
| 93.140              | corneal endothelial pigment without PPM                        | 12                   | 0.0% | 0                   |      |
| 93.150              | iris coloboma  | 11                   | 0.0% | 1                   | 0.0% |
| 93.180              | iris sphincter dysplasia                                       | 0                    |      | 2                   | 0.0% |
| 93.710              | persistent pupillary membranes, iris to iris                   | 6,793                | 3.0% | 1,424               | 3.8% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 144                  | 0.1% | 19                  | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 153                  | 0.1% | 12                  | 0.0% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 176                  | 0.1% | 0                   |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 280                  | 0.1% | 350                 | 0.9% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 28                   | 0.0% | 12                  | 0.0% |
| 93.810              | uveal melanoma   | 45                   | 0.0% | 36                  | 0.1% |
| 93.999              | uveal cysts  | 399                  | 0.2% | 120                 | 0.3% |
| 97.150              | chorioretinal coloboma, congenital                             | 0                    |      | 1                   | 0.0% |
| <b>LENS</b>         |  |                      |      |                     |      |
| 100.200             | cataract, unspecified  | 728                  | 0.3% | 0                   |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 9,963                | 4.3% | 1,770               | 4.7% |
| 100.301             | punctate cataract, anterior cortex                             | 897                  | 0.4% | 248                 | 0.7% |
| 100.302             | punctate cataract, posterior cortex                            | 1,287                | 0.6% | 153                 | 0.4% |
| 100.303             | punctate cataract, equatorial cortex                           | 166                  | 0.1% | 30                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 114                  | 0.0% | 19                  | 0.1% |
| 100.305             | punctate cataract, posterior sutures                           | 710                  | 0.3% | 147                 | 0.4% |
| 100.306             | punctate cataract, nucleus                                     | 174                  | 0.1% | 34                  | 0.1% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |      | <b>2016-2020</b> |      |
|---|------------------|------|------------------|------|
| 100.307 punctate cataract, capsular                       | 239              | 0.1% | 119              | 0.3% |
| 100.311 incipient cataract, anterior cortex               | 699              | 0.3% | 94               | 0.2% |
| 100.312 incipient cataract, posterior cortex              | 1,886            | 0.8% | 270              | 0.7% |
| 100.313 incipient cataract, equatorial cortex             | 488              | 0.2% | 68               | 0.2% |
| 100.314 incipient cataract, anterior sutures              | 59               | 0.0% | 9                | 0.0% |
| 100.315 incipient cataract, posterior sutures             | 456              | 0.2% | 69               | 0.2% |
| 100.316 incipient cataract, nucleus                       | 299              | 0.1% | 39               | 0.1% |
| 100.317 incipient cataract, capsular                      | 235              | 0.1% | 75               | 0.2% |
| 100.320 incomplete cataract, unspecified                  | 0                |      | 1                | 0.0% |
| 100.321 incomplete cataract, anterior cortex              | 13               | 0.0% | 20               | 0.1% |
| 100.322 incomplete cataract, posterior cortex             | 48               | 0.0% | 55               | 0.1% |
| 100.323 incomplete cataract, equatorial cortex            | 14               | 0.0% | 11               | 0.0% |
| 100.324 incomplete cataract, anterior sutures             | 1                | 0.0% | 0                |      |
| 100.325 incomplete cataract, posterior sutures            | 6                | 0.0% | 9                | 0.0% |
| 100.326 incomplete cataract, nucleus                      | 9                | 0.0% | 15               | 0.0% |
| 100.327 incomplete cataract, capsular                     | 6                | 0.0% | 12               | 0.0% |
| 100.328 y-suture tip opacities                            | 124              | 0.1% | 216              | 0.6% |
| 100.330 generalized/complete cataract                     | 350              | 0.2% | 11               | 0.0% |
| 100.340 resorbing/hypermature cataract                    | 1                | 0.0% | 5                | 0.0% |
| 100.345 <i>significant cataracts (summary)</i>            | 9,009            | 3.9% | 1,729            | 4.6% |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 51               | 0.0% | 7                | 0.0% |
| <b>VITREOUS</b>   |                  |      |                  |      |
| 110.120 persistent hyaloid artery/remnant                 | 576              | 0.3% | 129              | 0.3% |
| 110.135 PHPV/PTVL   | 150              | 0.1% | 15               | 0.0% |
| 110.200 vitreous degeneration-anterior chamber            | 9                | 0.0% | 24               | 0.1% |
| 110.320 vitreal degeneration                              | 821              | 0.4% | 149              | 0.4% |
| <b>FUNDUS</b>   |                  |      |                  |      |
| 97.110 choroidal hypoplasia                               | 14               | 0.0% | 0                |      |
| 97.120 coloboma   | 11               | 0.0% | 0                |      |
| <b>RETINA</b>   |                  |      |                  |      |
| 120.170 retinal dysplasia, folds                          | 5,030            | 2.2% | 400              | 1.1% |
| 120.180 retinal dysplasia, geographic                     | 1,984            | 0.9% | 170              | 0.5% |
| 120.190 retinal dysplasia, detached                       | 182              | 0.1% | 13               | 0.0% |
| 120.310 generalized progressive retinal atrophy (PRA)     | 985              | 0.4% | 16               | 0.0% |
| 120.400 retinal hemorrhage                                | 34               | 0.0% | 0                |      |
| 120.910 retinal detachment without dialysis               | 73               | 0.0% | 0                |      |
| 120.920 retinal detachment with dialysis                  | 6                | 0.0% | 7                | 0.0% |
| 120.960 retinopathy                                       | 58               | 0.0% | 41               | 0.1% |
| <b>OPTIC NERVE</b>  |                  |      |                  |      |
| 130.110 micropapilla                                      | 102              | 0.0% | 21               | 0.1% |
| 130.120 optic nerve hypoplasia                            | 86               | 0.0% | 5                | 0.0% |
| 130.150 optic disc coloboma                               | 43               | 0.0% | 6                | 0.0% |
| <b>OTHER</b>  |                  |      |                  |      |
| 900.000 other, unspecified                                | 1,697            | 0.7% | 0                |      |
| 900.100 other, not inherited                              | 4,315            | 1.9% | 50               | 0.1% |
| 900.110 other. suspect not inherited/significance unknown | 1,949            | 0.8% | 1,420            | 3.8% |

|                                     | 1991-2015     | 2016-2020    |
|-------------------------------------|---------------|--------------|
| <b>NORMAL</b><br>0.000 normal globe | 198,107 86.2% | 30,139 80.1% |



# LAGOTTO ROMAGNOLO

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE       |
|----|--|---------------------|-----------|--------------------|----------------------------------|
| A. | Distichiasis                                     | Not defined         | 1         | Breeder option     |                                  |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option     |                                  |
| C. | Cataract   | Not defined         | 1         | NO                 |                                  |
| D. | Retinal atrophy<br>( <i>prcd</i> )               | Autosomal recessive | 1         | NO                 | Mutation of the <i>prcd</i> gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Retinal atrophy – *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Lagotto Romagnolo is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

There are no references providing detailed descriptions of hereditary conditions of the Lagotto Romagnolo breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT LAGOTTO ROMAGNOLO

| Diagnostic Name    | TOTAL DOGS EXAMINED  | 1991-2015<br>287 |       | 2016-2020<br>877 |       |
|--------------------|--|------------------|-------|------------------|-------|
|                    |  | #                | %     | #                | %     |
| <b>EYELIDS</b>     |  |                  |       |                  |       |
| 25.110             | distichiasis   | 26               | 9.1%  | 79               | 9.0%  |
| <b>NICTITANS</b>   |  |                  |       |                  |       |
| 51.100             | third eyelid cartilage anomaly                                 | 1                | 0.3%  | 0                |       |
| 52.110             | prolapsed gland of the third eyelid                            | 1                | 0.3%  | 0                |       |
| <b>UVEA</b>        |  |                  |       |                  |       |
| 93.710             | persistent pupillary membranes, iris to iris                   | 12               | 4.2%  | 39               | 4.4%  |
| 93.750             | persistent pupillary membranes, lens pigment foci/no strands   | 3                | 1.0%  | 9                | 1.0%  |
| 93.760             | persistent pupillary membranes, endothelial opacity/no strands | 0                |       | 1                | 0.1%  |
| 93.999             | uveal cysts  | 0                |       | 1                | 0.1%  |
| <b>LENS</b>        |  |                  |       |                  |       |
| 100.210            | cataract. suspect not inherited/significance unknown           | 7                | 2.4%  | 20               | 2.3%  |
| 100.301            | punctate cataract, anterior cortex                             | 1                | 0.3%  | 5                | 0.6%  |
| 100.302            | punctate cataract, posterior cortex                            | 0                |       | 3                | 0.3%  |
| 100.303            | punctate cataract, equatorial cortex                           | 1                | 0.3%  | 4                | 0.5%  |
| 100.305            | punctate cataract, posterior sutures                           | 1                | 0.3%  | 0                |       |
| 100.306            | punctate cataract, nucleus                                     | 0                |       | 1                | 0.1%  |
| 100.311            | incipient cataract, anterior cortex                            | 0                |       | 2                | 0.2%  |
| 100.313            | incipient cataract, equatorial cortex                          | 2                | 0.7%  | 1                | 0.1%  |
| 100.315            | incipient cataract, posterior sutures                          | 0                |       | 1                | 0.1%  |
| 100.316            | incipient cataract, nucleus                                    | 0                |       | 1                | 0.1%  |
| 100.321            | incomplete cataract, anterior cortex                           | 1                | 0.3%  | 2                | 0.2%  |
| 100.322            | incomplete cataract, posterior cortex                          | 1                | 0.3%  | 1                | 0.1%  |
| 100.323            | incomplete cataract, equatorial cortex                         | 0                |       | 2                | 0.2%  |
| 100.326            | incomplete cataract, nucleus                                   | 1                | 0.3%  | 0                |       |
| 100.328            | y-suture tip opacities   | 0                |       | 3                | 0.3%  |
| 100.345            | <i>significant cataracts (summary)</i>                         | 8                | 2.8%  | 26               | 3.0%  |
| <b>VITREOUS</b>    |  |                  |       |                  |       |
| 110.120            | persistent hyaloid artery/remnant                              | 0                |       | 6                | 0.7%  |
| <b>RETINA</b>      |  |                  |       |                  |       |
| 120.170            | retinal dysplasia, folds                                       | 2                | 0.7%  | 4                | 0.5%  |
| 120.310            | generalized progressive retinal atrophy (PRA)                  | 0                |       | 1                | 0.1%  |
| 120.960            | retinopathy  | 0                |       | 1                | 0.1%  |
| <b>OPTIC NERVE</b> |  |                  |       |                  |       |
| 130.110            | micropapilla   | 0                |       | 6                | 0.7%  |
| <b>OTHER</b>       |  |                  |       |                  |       |
| 900.000            | other, unspecified   | 3                | 1.0%  | 0                |       |
| 900.100            | other, not inherited   | 0                |       | 1                | 0.1%  |
| 900.110            | other. suspect not inherited/significance unknown              | 6                | 2.1%  | 15               | 1.7%  |
| <b>NORMAL</b>      |  |                  |       |                  |       |
| 0.000              | normal globe   | 239              | 83.3% | 703              | 80.2% |

# LAKELAND TERRIER

|    | DISORDER   | INHERITANCE                | REFERENCE | BREEDING ADVICE                           | GENETIC TESTS AVAILABLE              |
|----|--|----------------------------|-----------|---|--------------------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined | 1<br>1    | Breeder Option<br>Passes with no notation |                                      |
| B. | Lens luxation  | Autosomal recessive        | 2         | NO  | Mutation of the <i>ADAMTS17</i> gene |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### B. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011; 14: 378-384.

# OCULAR DISORDERS REPORT LAKELAND TERRIER

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015<br>228 |       | 2016-2020<br>38 |       |
|-----------------|--|------------------|-------|-----------------|-------|
|                 |  | #                | %     | #               | %     |
| <b>EYELIDS</b>  |  |                  |       |                 |       |
| 25.110          | distichiasis   | 8                | 3.5%  | 4               | 10.5% |
| <b>CORNEA</b>   |  |                  |       |                 |       |
| 70.700          | corneal dystrophy  | 0                |       | 1               | 2.6%  |
| 70.730          | corneal endothelial degeneration                             | 2                | 0.9%  | 0               |       |
| <b>UVEA</b>     |  |                  |       |                 |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 33               | 14.5% | 5               | 13.2% |
| 93.720          | persistent pupillary membranes, iris to lens                 | 2                | 0.9%  | 0               |       |
| 93.730          | persistent pupillary membranes, iris to cornea               | 4                | 1.8%  | 0               |       |
| 93.740          | persistent pupillary membranes, iris sheets                  | 1                | 0.4%  | 0               |       |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 8                | 3.5%  | 6               | 15.8% |
| <b>LENS</b>     |  |                  |       |                 |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 5                | 2.2%  | 0               |       |
| 100.305         | punctate cataract, posterior sutures                         | 0                |       | 1               | 2.6%  |
| 100.311         | incipient cataract, anterior cortex                          | 3                | 1.3%  | 0               |       |
| 100.312         | incipient cataract, posterior cortex                         | 4                | 1.8%  | 0               |       |
| 100.328         | y-suture tip opacities                                       | 0                |       | 1               | 2.6%  |
| 100.330         | generalized/complete cataract                                | 3                | 1.3%  | 0               |       |
| 100.345         | <i>significant cataracts (summary)</i>                       | 10               | 4.4%  | 2               | 5.3%  |
| <b>RETINA</b>   |  |                  |       |                 |       |
| 120.180         | retinal dysplasia, geographic                                | 1                | 0.4%  | 0               |       |
| <b>OTHER</b>    |  |                  |       |                 |       |
| 900.000         | other, unspecified   | 2                | 0.9%  | 0               |       |
| 900.100         | other, not inherited   | 6                | 2.6%  | 0               |       |
| <b>NORMAL</b>   |  |                  |       |                 |       |
| 0.000           | normal globe   | 174              | 76.3% | 25              | 65.8% |

# LANCASHIRE HEELER

|    | DISORDER  | INHERITANCE            | REFEREN<br>CE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|---|------------------------|---------------|--------------------|---|
| A. | Persistent pupillary membrane<br>- iris to iris   | Not defined            | 1             | Breeder option     |   |
| B. | Lens luxation   | Autosomal<br>recessive | 2-4           | NO                 | Mutation of the<br><i>ADAMTS17</i> gene |
| C. | Choroidal hypoplasia<br>(Collie Eye Anomaly)<br>- staphyloma/coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- optic nerve coloboma | Autosomal<br>recessive | 5-7           | NO                 | Deletion in the<br><i>NHEJ1</i> gene    |
| D. | Retinal atrophy ( <i>prcd</i> )   | Autosomal<br>recessive | 1             | NO                 | Mutation of the<br><i>prcd</i> gene     |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or from sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

### C. Choroidal hypoplasia (Collie Eye Anomaly)

- staphyloma/coloboma
- retinal detachment
- retinal hemorrhage
- optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve

(coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

D. Retinal Atrophy – *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Lancashire Heeler is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Sargan DR, Withers D, Pettitt L, et al. Mapping the mutation causing lens luxation in several terrier breeds. *J Hered.* 2007;98:534-538.
3. Farias FH, Johnson GS, Taylor JF, et al. An ADAMTS17 splice donor site mutation in dogs with primary lens luxation. *Invest Ophthalmol Vis Sci.* 2010;51:4716-4721.
4. Gould D, Pettitt L, McLaughlin B, et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011;14:378-384.
5. Bedford PG. Collie eye anomaly in the Lancashire Heeler. *Vet Rec.* 1998;143:354-356.
6. Parker HG, Kukekova AV, Akey DT, et al. Breed relationships facilitate fine-mapping studies: a 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. *Gen Res.* 2007;17:1562-1571.
7. Lowe JK, Kukekova AV, Kirkness EF, et al. Linkage mapping of the primary disease locus for Collie eye anomaly. *Genomics.* 2003;82:86-95.

# OCULAR DISORDERS REPORT LANCASHIRE HEELER

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 1         | 0.7%  | 0         |       |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.700          | corneal dystrophy  | 0         |       | 1         | 3.6%  |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 58        | 41.1% | 4         | 14.3% |
| 93.720          | persistent pupillary membranes, iris to lens                 | 1         | 0.7%  | 0         |       |
| 93.730          | persistent pupillary membranes, iris to cornea               | 2         | 1.4%  | 0         |       |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 0         |       | 3         | 10.7% |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 1         | 0.7%  | 0         |       |
| 100.317         | incipient cataract, capsular                                 | 1         | 0.7%  | 0         |       |
| 100.345         | <i>significant cataracts (summary)</i>                       | 1         | 0.7%  | 0         |       |
| 100.375         | <i>subluxation/luxation, unspecified</i>                     | 1         | 0.7%  | 0         |       |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.120         | persistent hyaloid artery/remnant                            | 2         | 1.4%  | 0         |       |
| 110.200         | vitreous degeneration-anterior chamber                       | 1         | 0.7%  | 0         |       |
| 110.320         | vitreal degeneration   | 4         | 2.8%  | 0         |       |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                                     | 1         | 0.7%  | 0         |       |
| 120.310         | generalized progressive retinal atrophy (PRA)                | 1         | 0.7%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.110         | other. suspect not inherited/significance unknown            | 0         |       | 2         | 7.1%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 93        | 66.0% | 20        | 71.4% |



# LAPPONIAN HERDER

|    | DISORDER                             | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|--------------------------------------|---------------------|-----------|-----------------|-----------------------------------|
| A. | Retinal atrophy ( <i>prcd</i> )      | Autosomal recessive | 1         | NO              | Mutation of the <i>prcd</i> gene  |
| B. | Multifocal retinopathy - <i>cmr3</i> | Autosomal recessive | 2         | NO              | Mutation of the <i>BEST1</i> gene |

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## Description and Comments

### A. Retinal atrophy – *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Lapponian Herder is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

### B. Multifocal retinopathy (*cmr3*)

Canine Multifocal Retinopathy type 3 (*cmr3*) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Clinically the disease is similar to that seen in the Bullmastiff and Coton deTulear, but the mutation in the Bestrophin 1 gene (*BEST1* alias *VMD2*) is different. The multifocal retinopathy seen in the Lapponian Herder is caused by a deletion at position 1,388 and a

substitution at position 1,466 and is therefore called cmr3. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Lapponian Herder. The conditions listed above are currently noted solely due to the availability of a genetic test for the disease.

1. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006;88:551-563. PMID: 16938425
2. Zangerl B, Wickstrom K, Slavik J, et al. Assessment of canine BEST1 variations identifies new mutations and establishes an independent bestrophinopathy model (cmr3). *Mol Vis*. 2010;16:2791-2804.

# OCULAR DISORDERS REPORT LAPPONIAN HERDER

| Diagnostic Name                     | TOTAL DOGS EXAMINED | 1991-2015 |   | 2016-2020 |        |
|-------------------------------------|---------------------|-----------|---|-----------|--------|
|                                     |                     | #         | % | #         | %      |
| <b>NORMAL</b><br>0.000 normal globe |                     | 0         |   | 1         | 100.0% |

# **OCULAR DISORDERS REPORT LARGE MUNSTERLANDER**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the LARGE MUNSTERLANDER breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT LARGE MUNSTERLANDER

| Diagnostic Name                     | TOTAL DOGS EXAMINED | 1991-2015 |   | 2016-2020 |        |
|-------------------------------------|---------------------|-----------|---|-----------|--------|
|                                     |                     | #         | % | #         | %      |
| <b>NORMAL</b><br>0.000 normal globe |                     | 0         |   | 1         | 100.0% |

# LEONBERGER

|    | DISORDER                             | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--------------------------------------|-------------|-----------|-------------------------|
| A. | Ectropion                            | Not defined | 1         | Breeder option          |
| B. | Entropion                            | Not defined | 1, 2      | Breeder option          |
| C. | Distichiasis                         | Not defined | 1         | Breeder option          |
| D. | Nictitans cartilage anomaly/eversion | Not defined | 1         | Breeder option          |
| E. | Uveal cysts                          | Not defined | 1         | Breeder option          |
| F. | Persistent pupillary membranes       |             |           |                         |
|    | - iris to iris                       | Not defined | 1         | Breeder option          |
|    | - lens pigment foci/no strands       | Not defined |           | Passes with no notation |
| G. | Cataract                             | Not defined | 1         | NO                      |
| H. | Retinal dysplasia – folds            | Not defined | 1         | Breeder option          |

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## Description and Comments

### A. Ectropion

A conformational defect resulting in eversion of the eyelid(s), which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### C. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds.

Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

D. Nictitans cartilage anomaly/eversion

A scroll-like curling of the cartilage of the third eyelid, usually everting the margin. This condition may occur in one or both eyes and may cause mild ocular irritation.

E. Uveal cysts

Fluid filled sacs arising from the posterior surface of the iris, to which they may remain attached or break free and float into the anterior chamber. Usually occur in mature dogs.

There is usually no effect on vision unless the cysts are heavily clustered and impinge on the pupillary area. Less frequently, the cysts may rupture and adhere to the cornea or anterior lens capsule. Multiple cysts may occlude the iridocorneal angle and cause glaucoma.

F. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

G. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

H. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Heinrich CL, Lakhani KH, Featherstone HJ, et al. Cataract in the UK Leonberger population. *Vet Ophthalmol.* 2006 Sep-Oct;9:350-356.



# OCULAR DISORDERS REPORT LEONBERGER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|---------------------|--|-----------|-------|-----------|-------|
|                     |  | #         | %     | #         | %     |
| <b>EYELIDS</b>      |  |           |       |           |       |
| 20.160              | macropalpebral fissure   | 35        | 1.9%  | 0         |       |
| 21.000              | entropion, unspecified   | 58        | 3.2%  | 31        | 4.7%  |
| 22.000              | ectropion, unspecified   | 26        | 1.4%  | 11        | 1.7%  |
| 25.110              | distichiasis   | 45        | 2.5%  | 13        | 2.0%  |
| <b>NASOLACRIMAL</b> |  |           |       |           |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 1         | 0.1%  | 0         |       |
| <b>NICTITANS</b>    |  |           |       |           |       |
| 51.100              | third eyelid cartilage anomaly                                 | 23        | 1.3%  | 13        | 2.0%  |
| 52.110              | prolapsed gland of the third eyelid                            | 1         | 0.1%  | 2         | 0.3%  |
| <b>CORNEA</b>       |  |           |       |           |       |
| 70.700              | corneal dystrophy  | 5         | 0.3%  | 0         |       |
| <b>UVEA</b>         |  |           |       |           |       |
| 93.110              | iris hypoplasia  | 1         | 0.1%  | 1         | 0.2%  |
| 93.710              | persistent pupillary membranes, iris to iris                   | 392       | 21.4% | 156       | 23.7% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 2         | 0.1%  | 0         |       |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 1         | 0.1%  | 0         |       |
| 93.740              | persistent pupillary membranes, iris sheets                    | 1         | 0.1%  | 0         |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 10        | 0.5%  | 8         | 1.2%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 1         | 0.1%  | 1         | 0.2%  |
| 93.810              | uveal melanoma   | 1         | 0.1%  | 0         |       |
| 93.999              | uveal cysts  | 15        | 0.8%  | 10        | 1.5%  |
| <b>LENS</b>         |  |           |       |           |       |
| 100.200             | cataract, unspecified  | 2         | 0.1%  | 0         |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 154       | 8.4%  | 52        | 7.9%  |
| 100.301             | punctate cataract, anterior cortex                             | 22        | 1.2%  | 12        | 1.8%  |
| 100.302             | punctate cataract, posterior cortex                            | 23        | 1.3%  | 8         | 1.2%  |
| 100.303             | punctate cataract, equatorial cortex                           | 3         | 0.2%  | 0         |       |
| 100.304             | punctate cataract, anterior sutures                            | 3         | 0.2%  | 1         | 0.2%  |
| 100.305             | punctate cataract, posterior sutures                           | 12        | 0.7%  | 7         | 1.1%  |
| 100.306             | punctate cataract, nucleus                                     | 6         | 0.3%  | 10        | 1.5%  |
| 100.307             | punctate cataract, capsular                                    | 4         | 0.2%  | 7         | 1.1%  |
| 100.311             | incipient cataract, anterior cortex                            | 9         | 0.5%  | 9         | 1.4%  |
| 100.312             | incipient cataract, posterior cortex                           | 30        | 1.6%  | 10        | 1.5%  |
| 100.313             | incipient cataract, equatorial cortex                          | 1         | 0.1%  | 0         |       |
| 100.314             | incipient cataract, anterior sutures                           | 5         | 0.3%  | 1         | 0.2%  |
| 100.315             | incipient cataract, posterior sutures                          | 8         | 0.4%  | 3         | 0.5%  |
| 100.316             | incipient cataract, nucleus                                    | 19        | 1.0%  | 10        | 1.5%  |
| 100.317             | incipient cataract, capsular                                   | 3         | 0.2%  | 7         | 1.1%  |
| 100.321             | incomplete cataract, anterior cortex                           | 0         |       | 1         | 0.2%  |
| 100.322             | incomplete cataract, posterior cortex                          | 1         | 0.1%  | 2         | 0.3%  |
| 100.326             | incomplete cataract, nucleus                                   | 0         |       | 1         | 0.2%  |
| 100.328             | y-suture tip opacities   | 3         | 0.2%  | 5         | 0.8%  |
| 100.330             | generalized/complete cataract                                  | 4         | 0.2%  | 0         |       |
| 100.345             | significant cataracts (summary)                                | 158       | 8.6%  | 94        | 14.3% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.375 subluxation/luxation, unspecified                 | 5 0.3%           | 3 0.5%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 3 0.2%           | 6 0.9%           |
| 110.135 PHPV/PTVL   | 4 0.2%           | 1 0.2%           |
| 110.200 vitreous degeneration-anterior chamber            | 0                | 1 0.2%           |
| 110.320 vitreal degeneration                              | 6 0.3%           | 2 0.3%           |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 8 0.4%           | 9 1.4%           |
| 120.180 retinal dysplasia, geographic                     | 4 0.2%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 5 0.3%           | 0                |
| 120.960 retinopathy                                       | 1 0.1%           | 1 0.2%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 1 0.1%           | 0                |
| 130.120 optic nerve hypoplasia                            | 2 0.1%           | 0                |
| 130.150 optic disc coloboma                               | 1 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 32 1.7%          | 0                |
| 900.100 other, not inherited                              | 53 2.9%          | 1 0.2%           |
| 900.110 other. suspect not inherited/significance unknown | 22 1.2%          | 38 5.8%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,208 65.9%      | 335 51.0%        |

# LHASA APSO

|    | DISORDER     | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--------------|-------------|-----------|-----------------|
| A. | Distichiasis | Not defined | 1         | Breeder option  |
| B. | Cataract     | Not defined | 2         | NO              |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gelatt KN, Mackay EO. Prevalence of primary breed-related cataracts in the dog in North America. *Vet Ophthalmol.* 2005;8:101-111.

# OCULAR DISORDERS REPORT LHASA APSO

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>794 |      | 2016-2020<br>90 |      |
|---------------------|--|------------------|------|-----------------|------|
|                     |  | #                | %    | #               | %    |
| <b>GLOBE</b>        |  |                  |      |                 |      |
| 0.110               | microphthalmia   | 1                | 0.1% | 0               |      |
| <b>EYELIDS</b>      |  |                  |      |                 |      |
| 20.160              | macropalpebral fissure                                       | 3                | 0.4% | 0               |      |
| 21.000              | entropion, unspecified                                       | 11               | 1.4% | 3               | 3.3% |
| 25.110              | distichiasis   | 32               | 4.0% | 2               | 2.2% |
| <b>NASOLACRIMAL</b> |  |                  |      |                 |      |
| 32.110              | imperforate lower nasolacrimal punctum                       | 1                | 0.1% | 0               |      |
| 40.910              | keratoconjunctivitis sicca                                   | 3                | 0.4% | 0               |      |
| <b>NICTITANS</b>    |  |                  |      |                 |      |
| 51.100              | third eyelid cartilage anomaly                               | 1                | 0.1% | 0               |      |
| 52.110              | prolapsed gland of the third eyelid                          | 4                | 0.5% | 0               |      |
| <b>CORNEA</b>       |  |                  |      |                 |      |
| 70.210              | corneal pannus   | 8                | 1.0% | 0               |      |
| 70.220              | pigmentary keratitis   | 20               | 2.5% | 3               | 3.3% |
| 70.700              | corneal dystrophy  | 16               | 2.0% | 0               |      |
| <b>UVEA</b>         |  |                  |      |                 |      |
| 93.110              | iris hypoplasia  | 1                | 0.1% | 0               |      |
| 93.710              | persistent pupillary membranes, iris to iris                 | 10               | 1.3% | 0               |      |
| 93.730              | persistent pupillary membranes, iris to cornea               | 1                | 0.1% | 0               |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands | 0                |      | 2               | 2.2% |
| 93.999              | uveal cysts  | 1                | 0.1% | 0               |      |
| <b>LENS</b>         |  |                  |      |                 |      |
| 100.200             | cataract, unspecified  | 6                | 0.8% | 0               |      |
| 100.210             | cataract. suspect not inherited/significance unknown         | 27               | 3.4% | 2               | 2.2% |
| 100.301             | punctate cataract, anterior cortex                           | 6                | 0.8% | 1               | 1.1% |
| 100.302             | punctate cataract, posterior cortex                          | 5                | 0.6% | 0               |      |
| 100.303             | punctate cataract, equatorial cortex                         | 3                | 0.4% | 0               |      |
| 100.306             | punctate cataract, nucleus                                   | 1                | 0.1% | 0               |      |
| 100.311             | incipient cataract, anterior cortex                          | 13               | 1.6% | 3               | 3.3% |
| 100.312             | incipient cataract, posterior cortex                         | 14               | 1.8% | 0               |      |
| 100.313             | incipient cataract, equatorial cortex                        | 3                | 0.4% | 1               | 1.1% |
| 100.314             | incipient cataract, anterior sutures                         | 4                | 0.5% | 0               |      |
| 100.315             | incipient cataract, posterior sutures                        | 2                | 0.3% | 0               |      |
| 100.316             | incipient cataract, nucleus                                  | 3                | 0.4% | 0               |      |
| 100.322             | incomplete cataract, posterior cortex                        | 0                |      | 1               | 1.1% |
| 100.330             | generalized/complete cataract                                | 18               | 2.3% | 1               | 1.1% |
| 100.345             | significant cataracts (summary)                              | 78               | 9.8% | 7               | 7.8% |
| 100.375             | subluxation/luxation, unspecified                            | 1                | 0.1% | 0               |      |
| <b>VITREOUS</b>     |  |                  |      |                 |      |
| 110.200             | vitreal degeneration-anterior chamber                        | 1                | 0.1% | 1               | 1.1% |
| 110.320             | vitreal degeneration   | 9                | 1.1% | 0               |      |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>FUNDUS</b>   |           |           |
| 97.110 choroidal hypoplasia                               | 1 0.1%    | 0         |
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 4 0.5%    | 3 3.3%    |
| 120.180 retinal dysplasia, geographic                     | 3 0.4%    | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 7 0.9%    | 0         |
| <b>OPTIC NERVE</b>  |           |           |
| 130.110 micropapilla                                      | 1 0.1%    | 0         |
| 130.120 optic nerve hypoplasia                            | 2 0.3%    | 0         |
| 130.150 optic disc coloboma                               | 1 0.1%    | 0         |
| <b>OTHER</b>  |           |           |
| 900.100 other, not inherited                              | 12 1.5%   | 0         |
| 900.110 other. suspect not inherited/significance unknown | 20 2.5%   | 4 4.4%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 609 76.7% | 65 72.2%  |

# LOUISIANA CATAHOULA LEOPARD DOG

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| B. | Cataract   | Not defined | 1         | NO              |
| C. | Iris coloboma                                    | Not defined | 1         | NO              |

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**It is recommended that this breed be examined prior to pharmacological dilation to best facilitate identification of iris coloboma and persistent pupillary membranes.**

## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Iris coloboma

A congenital abnormality in iris development usually characterized by a full-thickness defect in iris tissue, commonly (though not exclusively) located at the 6 o'clock position associated with failure of closure of the optic fissure. A partial-thickness defect in iris tissue should be recorded as iris hypoplasia on the OFA form.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT

## LOUISIANA CATAHOULA LEOPARD DOG

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|--|---------------------|-----------|-------|-----------|-------|
|  |                     | #         | %     | #         | %     |
| <b>GLOBE</b>   |                     |           |       |           |       |
| 0.110 microphthalmia   |                     | 5         | 1.3%  | 0         |       |
| <b>EYELIDS</b>   |                     |           |       |           |       |
| 25.110 distichiasis  |                     | 3         | 0.8%  | 0         |       |
| <b>CORNEA</b>  |                     |           |       |           |       |
| 70.700 corneal dystrophy                                     |                     | 1         | 0.3%  | 0         |       |
| <b>UVEA</b>  |                     |           |       |           |       |
| 93.110 iris hypoplasia                                       |                     | 3         | 0.8%  | 1         | 1.4%  |
| 93.150 iris coloboma   |                     | 12        | 3.2%  | 1         | 1.4%  |
| 93.710 persistent pupillary membranes, iris to iris          |                     | 38        | 10.2% | 6         | 8.5%  |
| 93.720 persistent pupillary membranes, iris to lens          |                     | 1         | 0.3%  | 0         |       |
| 93.730 persistent pupillary membranes, iris to cornea        |                     | 0         |       | 1         | 1.4%  |
| 97.150 chorioretinal coloboma, congenital                    |                     | 1         | 0.3%  | 0         |       |
| <b>LENS</b>  |                     |           |       |           |       |
| 100.200 cataract, unspecified                                |                     | 1         | 0.3%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 5         | 1.3%  | 2         | 2.8%  |
| 100.302 punctate cataract, posterior cortex                  |                     | 1         | 0.3%  | 1         | 1.4%  |
| 100.306 punctate cataract, nucleus                           |                     | 0         |       | 1         | 1.4%  |
| 100.311 incipient cataract, anterior cortex                  |                     | 4         | 1.1%  | 2         | 2.8%  |
| 100.312 incipient cataract, posterior cortex                 |                     | 2         | 0.5%  | 1         | 1.4%  |
| 100.313 incipient cataract, equatorial cortex                |                     | 2         | 0.5%  | 0         |       |
| 100.316 incipient cataract, nucleus                          |                     | 0         |       | 1         | 1.4%  |
| 100.322 incomplete cataract, posterior cortex                |                     | 0         |       | 2         | 2.8%  |
| 100.330 generalized/complete cataract                        |                     | 0         |       | 1         | 1.4%  |
| 100.345 <i>significant cataracts (summary)</i>               |                     | 10        | 2.7%  | 9         | 12.7% |
| <b>VITREOUS</b>  |                     |           |       |           |       |
| 110.120 persistent hyaloid artery/remnant                    |                     | 2         | 0.5%  | 0         |       |
| 110.320 vitreal degeneration                                 |                     | 2         | 0.5%  | 0         |       |
| <b>FUNDUS</b>  |                     |           |       |           |       |
| 97.110 choroidal hypoplasia                                  |                     | 1         | 0.3%  | 1         | 1.4%  |
| 97.120 coloboma  |                     | 2         | 0.5%  | 0         |       |
| <b>RETINA</b>  |                     |           |       |           |       |
| 120.170 retinal dysplasia, folds                             |                     | 9         | 2.4%  | 0         |       |
| 120.910 retinal detachment without dialysis                  |                     | 2         | 0.5%  | 0         |       |
| 120.920 retinal detachment with dialysis                     |                     | 1         | 0.3%  | 0         |       |
| <b>OPTIC NERVE</b>   |                     |           |       |           |       |
| 130.150 optic disc coloboma                                  |                     | 2         | 0.5%  | 2         | 2.8%  |
| <b>OTHER</b>   |                     |           |       |           |       |
| 900.100 other, not inherited                                 |                     | 4         | 1.1%  | 0         |       |
| 900.110 other. suspect not inherited/significance unknown    |                     | 12        | 3.2%  | 3         | 4.2%  |

|                                     | 1991-2015 | 2016-2020 |
|-------------------------------------|-----------|-----------|
| <b>NORMAL</b><br>0.000 normal globe | 304 81.7% | 54 76.1%  |



# LOWCHEN

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--------------------------------|-------------|-----------|-------------------------|
| A. | Distichiasis                   | Not defined | 1         | Breeder option          |
| B. | Persistent pupillary membranes |             |           |                         |
|    | - iris to iris                 | Not defined | 1         | Breeder option          |
|    | - lens pigment foci/no strands | Not defined | 1         | Passes with no notation |
| C. | Cataract                       | Not defined | 1         | NO                      |
| D. | Vitreous degeneration          | Not defined | 1         | Breeder option          |
| E. | Retinal atrophy                | Not defined | 1         | NO                      |
|    | - generalized                  |             |           |                         |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely

(diffuse) or in a localized region.

D. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment and/or glaucoma.

E. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Lowchen breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT LOWCHEN

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>1,676 |      | 2016-2020<br>390 |       |
|---------------------|--|--------------------|------|------------------|-------|
|                     |  | #                  | %    | #                | %     |
| <b>EYELIDS</b>      |  |                    |      |                  |       |
| 20.140              | ectopic cilia  | 1                  | 0.1% | 0                |       |
| 21.000              | entropion, unspecified   | 1                  | 0.1% | 0                |       |
| 25.110              | distichiasis   | 78                 | 4.7% | 24               | 6.2%  |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |       |
| 40.910              | keratoconjunctivitis sicca                                     | 0                  |      | 1                | 0.3%  |
| <b>CORNEA</b>       |  |                    |      |                  |       |
| 70.210              | corneal pannus   | 1                  | 0.1% | 0                |       |
| 70.730              | corneal endothelial degeneration                               | 2                  | 0.1% | 0                |       |
| <b>UVEA</b>         |  |                    |      |                  |       |
| 93.150              | iris coloboma  | 1                  | 0.1% | 0                |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 130                | 7.8% | 41               | 10.5% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 3                  | 0.2% | 0                |       |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 2                  | 0.1% | 1                | 0.3%  |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 7                  | 0.4% | 10               | 2.6%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 1                  | 0.1% | 0                |       |
| 93.999              | uveal cysts  | 1                  | 0.1% | 1                | 0.3%  |
| <b>LENS</b>         |  |                    |      |                  |       |
| 100.200             | cataract, unspecified  | 21                 | 1.3% | 0                |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 56                 | 3.3% | 11               | 2.8%  |
| 100.301             | punctate cataract, anterior cortex                             | 8                  | 0.5% | 1                | 0.3%  |
| 100.302             | punctate cataract, posterior cortex                            | 12                 | 0.7% | 0                |       |
| 100.303             | punctate cataract, equatorial cortex                           | 4                  | 0.2% | 0                |       |
| 100.304             | punctate cataract, anterior sutures                            | 1                  | 0.1% | 0                |       |
| 100.305             | punctate cataract, posterior sutures                           | 5                  | 0.3% | 1                | 0.3%  |
| 100.306             | punctate cataract, nucleus                                     | 2                  | 0.1% | 0                |       |
| 100.307             | punctate cataract, capsular                                    | 1                  | 0.1% | 1                | 0.3%  |
| 100.311             | incipient cataract, anterior cortex                            | 21                 | 1.3% | 3                | 0.8%  |
| 100.312             | incipient cataract, posterior cortex                           | 24                 | 1.4% | 3                | 0.8%  |
| 100.313             | incipient cataract, equatorial cortex                          | 5                  | 0.3% | 3                | 0.8%  |
| 100.314             | incipient cataract, anterior sutures                           | 2                  | 0.1% | 0                |       |
| 100.315             | incipient cataract, posterior sutures                          | 4                  | 0.2% | 0                |       |
| 100.316             | incipient cataract, nucleus                                    | 1                  | 0.1% | 1                | 0.3%  |
| 100.317             | incipient cataract, capsular                                   | 2                  | 0.1% | 0                |       |
| 100.321             | incomplete cataract, anterior cortex                           | 1                  | 0.1% | 1                | 0.3%  |
| 100.322             | incomplete cataract, posterior cortex                          | 1                  | 0.1% | 0                |       |
| 100.323             | incomplete cataract, equatorial cortex                         | 1                  | 0.1% | 0                |       |
| 100.328             | y-suture tip opacities   | 0                  |      | 1                | 0.3%  |
| 100.330             | generalized/complete cataract                                  | 16                 | 1.0% | 1                | 0.3%  |
| 100.345             | significant cataracts (summary)                                | 132                | 7.9% | 16               | 4.1%  |
| 100.375             | subluxation/luxation, unspecified                              | 2                  | 0.1% | 0                |       |
| <b>VITREOUS</b>     |  |                    |      |                  |       |
| 110.120             | persistent hyaloid artery/remnant                              | 3                  | 0.2% | 3                | 0.8%  |
| 110.135             | PHPV/PTVL  | 1                  | 0.1% | 0                |       |
| 110.200             | vitreous degeneration-anterior chamber                         | 1                  | 0.1% | 0                |       |

| <b>VITREOUS CONTINUED</b>                                 | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 110.320 vitreal degeneration                              | 51 3.0%          | 3 0.8%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 2 0.1%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 3 0.2%           | 0                |
| 120.180 retinal dysplasia, geographic                     | 0                | 1 0.3%           |
| 120.190 retinal dysplasia, detached                       | 1 0.1%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 38 2.3%          | 3 0.8%           |
| 120.910 retinal detachment without dialysis               | 2 0.1%           | 0                |
| 120.960 retinopathy                                       | 4 0.2%           | 2 0.5%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 1 0.1%           | 0                |
| 130.150 optic disc coloboma                               | 1 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 13 0.8%          | 0                |
| 900.100 other, not inherited                              | 39 2.3%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 7 0.4%           | 8 2.1%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,343 80.1%      | 283 72.6%        |

# LUCAS TERRIER

|    | DISORDER      | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|---------------|------------------------|-----------|--------------------|---|
| A. | Lens luxation | Autosomal<br>recessive | 1         | NO                 | Mutation of the<br><i>ADAMTS17</i> gene |

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## Description and Comments

### A. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Lucas Terrier. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011; 14: 378-384.

# MALTESE

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Entropion  | Not defined        | 1                | Breeder option         |
| B. | Distichiasis                                     | Not defined        | 1                | Breeder option         |
| C. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |
| D. | Cataract   | Not defined        | 1, 2             | NO                     |
| E. | Vitreous degeneration                            | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Entropion

A conformational defect resulting in inversion of the eyelid margin which may cause ocular irritation. It is likely that entropion is influenced by several factors defining the skin and other structures, which make up the eyelids, orbital contents, and conformation of the skull.

### B. Distichiasis

The presence of abnormally oriented eyelashes, frequently protruding from Meibomian gland ductal openings.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### E. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gelatt KN and Mackay EO. Prevalence of primary breed-related cataracts in the dog in North America. *Vet Ophthalmol.* 2005 Mar-Apr;8:101-111.

# OCULAR DISORDERS REPORT MALTESE

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |      |
|---|---------------------|-----------|-------|-----------|------|
|   |                     | #         | %     | #         | %    |
| <b>GLOBE</b>  |                     |           |       |           |      |
| 0.110 microphthalmia  |                     | 1         | 0.3%  | 0         |      |
| <b>EYELIDS</b>  |                     |           |       |           |      |
| 21.000 entropion, unspecified                                       |                     | 4         | 1.3%  | 7         | 2.6% |
| 25.110 distichiasis   |                     | 12        | 3.8%  | 3         | 1.1% |
| <b>NASOLACRIMAL</b>   |                     |           |       |           |      |
| 32.110 imperforate lower nasolacrimal punctum                       |                     | 1         | 0.3%  | 0         |      |
| 40.910 keratoconjunctivitis sicca                                   |                     | 2         | 0.6%  | 0         |      |
| <b>NICTITANS</b>  |                     |           |       |           |      |
| 52.110 prolapsed gland of the third eyelid                          |                     | 2         | 0.6%  | 2         | 0.7% |
| <b>CORNEA</b>   |                     |           |       |           |      |
| 70.220 pigmentary keratitis   |                     | 0         |       | 4         | 1.5% |
| 70.700 corneal dystrophy  |                     | 2         | 0.6%  | 0         |      |
| <b>UVEA</b>   |                     |           |       |           |      |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 18        | 5.8%  | 2         | 0.7% |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 0         |       | 1         | 0.4% |
| 93.999 uveal cysts  |                     | 1         | 0.3%  | 0         |      |
| <b>LENS</b>   |                     |           |       |           |      |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 17        | 5.4%  | 0         |      |
| 100.301 punctate cataract, anterior cortex                          |                     | 2         | 0.6%  | 3         | 1.1% |
| 100.302 punctate cataract, posterior cortex                         |                     | 4         | 1.3%  | 1         | 0.4% |
| 100.303 punctate cataract, equatorial cortex                        |                     | 2         | 0.6%  | 0         |      |
| 100.304 punctate cataract, anterior sutures                         |                     | 1         | 0.3%  | 0         |      |
| 100.305 punctate cataract, posterior sutures                        |                     | 2         | 0.6%  | 0         |      |
| 100.306 punctate cataract, nucleus                                  |                     | 1         | 0.3%  | 0         |      |
| 100.307 punctate cataract, capsular                                 |                     | 1         | 0.3%  | 1         | 0.4% |
| 100.311 incipient cataract, anterior cortex                         |                     | 9         | 2.9%  | 3         | 1.1% |
| 100.312 incipient cataract, posterior cortex                        |                     | 9         | 2.9%  | 1         | 0.4% |
| 100.313 incipient cataract, equatorial cortex                       |                     | 2         | 0.6%  | 0         |      |
| 100.314 incipient cataract, anterior sutures                        |                     | 0         |       | 1         | 0.4% |
| 100.315 incipient cataract, posterior sutures                       |                     | 1         | 0.3%  | 1         | 0.4% |
| 100.316 incipient cataract, nucleus                                 |                     | 2         | 0.6%  | 0         |      |
| 100.317 incipient cataract, capsular                                |                     | 1         | 0.3%  | 0         |      |
| 100.321 incomplete cataract, anterior cortex                        |                     | 0         |       | 1         | 0.4% |
| 100.322 incomplete cataract, posterior cortex                       |                     | 0         |       | 1         | 0.4% |
| 100.323 incomplete cataract, equatorial cortex                      |                     | 0         |       | 1         | 0.4% |
| 100.328 y-suture tip opacities                                      |                     | 1         | 0.3%  | 1         | 0.4% |
| 100.330 generalized/complete cataract                               |                     | 4         | 1.3%  | 0         |      |
| 100.345 significant cataracts (summary)                             |                     | 42        | 13.4% | 15        | 5.6% |
| <b>VITREOUS</b>   |                     |           |       |           |      |
| 110.120 persistent hyaloid artery/remnant                           |                     | 1         | 0.3%  | 0         |      |
| 110.200 vitreous degeneration-anterior chamber                      |                     | 0         |       | 1         | 0.4% |
| 110.320 vitreal degeneration  |                     | 8         | 2.6%  | 6         | 2.2% |



|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 3 1.0%    | 1 0.4%    |
| 120.180 retinal dysplasia, geographic                     | 5 1.6%    | 0         |
| 120.190 retinal dysplasia, detached                       | 0         | 1 0.4%    |
| 120.310 generalized progressive retinal atrophy (PRA)     | 4 1.3%    | 1 0.4%    |
| 120.920 retinal detachment with dialysis                  | 0         | 1 0.4%    |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 8 2.6%    | 0         |
| 900.100 other, not inherited                              | 6 1.9%    | 0         |
| 900.110 other. suspect not inherited/significance unknown | 3 1.0%    | 10 3.7%   |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 230 73.5% | 222 82.5% |

# MANCHESTER TERRIER

## Standard & Toy Varieties

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|---|------------------------|-----------|--------------------|-------------------------------------|
| A. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined            | 1         | Breeder option     |                                     |
| B. | Cataract  | Not defined            | 1         | NO                 |                                     |
| C. | Retinal atrophy<br>( <i>prcd</i> )                  | Autosomal<br>recessive | 1         | NO                 | Mutation of the<br><i>prcd</i> gene |

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### Description and Comments

#### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

#### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

#### C. Retinal Atrophy – *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Manchester Terrier is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with

signs of night blindness followed by day blindness. A DNA test is available.

## **References**

There are no references providing detailed descriptions of hereditary ocular conditions of the Manchester Terrier breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT MANCHESTER TERRIER

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 1         | 0.5%  | 0         |       |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                   | 12        | 6.2%  | 5         | 3.8%  |
| 93.730          | persistent pupillary membranes, iris to cornea                 | 1         | 0.5%  | 0         |       |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands   | 4         | 2.1%  | 3         | 2.3%  |
| 93.760          | persistent pupillary membranes, endothelial opacity/no strands | 2         | 1.0%  | 0         |       |
| 93.999          | uveal cysts  | 1         | 0.5%  | 0         |       |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown           | 6         | 3.1%  | 6         | 4.5%  |
| 100.301         | punctate cataract, anterior cortex                             | 1         | 0.5%  | 2         | 1.5%  |
| 100.302         | punctate cataract, posterior cortex                            | 2         | 1.0%  | 2         | 1.5%  |
| 100.303         | punctate cataract, equatorial cortex                           | 1         | 0.5%  | 0         |       |
| 100.305         | punctate cataract, posterior sutures                           | 1         | 0.5%  | 1         | 0.8%  |
| 100.307         | punctate cataract, capsular                                    | 0         |       | 1         | 0.8%  |
| 100.311         | incipient cataract, anterior cortex                            | 2         | 1.0%  | 0         |       |
| 100.312         | incipient cataract, posterior cortex                           | 2         | 1.0%  | 3         | 2.3%  |
| 100.313         | incipient cataract, equatorial cortex                          | 1         | 0.5%  | 0         |       |
| 100.317         | incipient cataract, capsular                                   | 2         | 1.0%  | 1         | 0.8%  |
| 100.328         | y-suture tip opacities   | 0         |       | 1         | 0.8%  |
| 100.345         | <i>significant cataracts (summary)</i>                         | 12        | 6.2%  | 11        | 8.3%  |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.135         | PHPV/PTVL  | 3         | 1.6%  | 0         |       |
| 110.320         | vitreal degeneration   | 7         | 3.6%  | 0         |       |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                                       | 1         | 0.5%  | 1         | 0.8%  |
| 120.960         | retinopathy  | 1         | 0.5%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.000         | other, unspecified   | 6         | 3.1%  | 0         |       |
| 900.110         | other. suspect not inherited/significance unknown              | 2         | 1.0%  | 5         | 3.8%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 165       | 85.5% | 107       | 80.5% |

# MAREMMA SHEEPDOG

|    | <b>DISORDER</b>                      | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--------------------------------------|--------------------|------------------|------------------------|
| A. | Entropion                            | Not defined        | 1                | Breeder option         |
| B. | Corneal dystrophy                    | Not defined        | 1                | Breeder option         |
| C. | Chronic superficial keratitis/pannus | Not defined        | 1                | NO                     |
| D. | Cataract                             | Not defined        | 1                | NO                     |
| E. | Retinal dysplasia - folds            | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

### C. Chronic superficial keratitis/pannus

A bilateral inflammatory disease of the cornea which usually starts as a grayish haze to the ventral or ventrolateral cornea, followed by the formation of a vascularized subepithelial growth that begins to spread toward the central cornea; pigmentation follows the vascularization. If severe, vision impairment occurs. Pannus may be associated with plasma cell infiltration of the nictitans. This has been reported in the Italian population of the breed.

### D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region. This has been reported in the Italian population of the breed.

E. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined. This has been reported in the Italian population of the breed.

## References

1. Guandalini A, Di Girolamo N, Santillo D, Andreani V, Corvi R, Bandini M, and Peruccio C. (2017) Epidemiology of ocular disorders presumed to be inherited in three large Italian dog breeds in Italy. *Vet Ophthalmol*, 20: 420-426. doi:10.1111/vop.12442.

# OCULAR DISORDERS REPORT MAREMMA SHEEPDOG

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015<br>13 |       | 2016-2020<br>18 |       |
|--|---------------------|-----------------|-------|-----------------|-------|
|  |                     | #               | %     | #               | %     |
| <b>UVEA</b>  |                     |                 |       |                 |       |
| 93.710 persistent pupillary membranes, iris to iris          |                     | 2               | 15.4% | 0               |       |
| <b>LENS</b>  |                     |                 |       |                 |       |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 2               | 15.4% | 1               | 5.6%  |
| 100.301 punctate cataract, anterior cortex                   |                     | 0               |       | 1               | 5.6%  |
| 100.328 y-suture tip opacities                               |                     | 0               |       | 1               | 5.6%  |
| 100.345 significant cataracts (summary)                      |                     | 0               |       | 2               | 11.1% |
| <b>VITREOUS</b>  |                     |                 |       |                 |       |
| 110.320 vitreal degeneration                                 |                     | 1               | 7.7%  | 0               |       |
| <b>OTHER</b>   |                     |                 |       |                 |       |
| 900.000 other, unspecified                                   |                     | 1               | 7.7%  | 0               |       |
| 900.110 other. suspect not inherited/significance unknown    |                     | 0               |       | 1               | 5.6%  |
| <b>NORMAL</b>  |                     |                 |       |                 |       |
| 0.000 normal globe   |                     | 9               | 69.2% | 15              | 83.3% |

# MARKIESJE

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|------------------------------------|------------------------|-----------|--------------------|-------------------------------------|
| A. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 1         | NO                 | Mutation of the <i>prcd</i><br>gene |

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## Description and Comments

### A. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Markiesje is *prcd*, which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Markiesje breed. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006 Nov;88:551-563. PMID: 16938425



# MASTIFF

(English)

|    | DISORDER                             | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|--------------------------------------|---------------------|-----------|-----------------|-----------------------------------|
| A. | Entropion                            | Not defined         | 1         | Breeder option  |                                   |
| B. | Ectropion                            | Not defined         | 1         | Breeder option  |                                   |
| C. | Distichiasis                         | Not defined         | 1         | Breeder option  |                                   |
| D. | Uveal cysts                          | Not defined         | 1         | Breeder option  |                                   |
| E. | Persistent pupillary membranes       |                     |           |                 |                                   |
|    | - iris to iris                       | Not defined         | 1         | Breeder option  |                                   |
|    | - iris to cornea                     | Not defined         | 1         | NO              |                                   |
|    | - endothelial opacity/no strands     | Not defined         | 4         | NO              |                                   |
| F. | Cataract                             | Not defined         | 1         | NO              |                                   |
| G. | Retinal atrophy ( <i>RHO</i> )       | Autosomal dominant  | 1, 2      | NO              | Mutation of the <i>RHO</i> gene   |
| H. | Multifocal retinopathy - <i>cmr1</i> | Autosomal recessive | 3         | Breeder option  | Mutation of the <i>BEST1</i> gene |
| I. | Retinal dysplasia - folds            | Not defined         | 1         | Breeder option  |                                   |

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## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. Entropion in the Mastiff is severe and may require multiple surgical corrections.

### B. Ectropion

A conformational defect resulting in eversion of the eyelids, which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin

covering the head and face, the orbital contents, and the conformation of the skull.

C. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

D. Uveal cysts

Fluid filled sacs arising from the posterior surface of the iris, to which they may remain attached or break free and float into the anterior chamber. Usually occur in mature dogs.

There is usually no effect on vision unless the cysts are heavily clustered and impinge on the pupillary area. Less frequently, the cysts may rupture and adhere to the cornea or anterior lens capsule. Multiple cysts may occlude the iridocorneal angle and cause glaucoma.

E. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

In the Mastiff, the strands most often bridge from the iris to the cornea and may potentially cause vision impairment. Thus, the strong recommendations against breeding animals with any form of this abnormality.

F. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

G. Retinal atrophy - *RHO*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. The ERG is normal at 3-6 months of age, but abnormal by 13 months of age. Increased exposure to bright light causes more rapid loss of neurons. PRA in the Mastiff is inherited as an autosomal dominant trait. The mutation is a single nucleotide transversion of the *RHO* gene. A DNA test is available.

## H. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (cmr1) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff. A DNA test is available.

## I. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Kijas JW, Miller BJ, Pearce-Kelling SE, et al. Canine models of ocular disease: outcross breedings define a dominant disorder present in the English mastiff and bull mastiff dog breeds. *J Hered.* 2003;94:27-30.
3. Miyadera K, Acland GM, Aguirre GD. Genetic and phenotypic variations of inherited retinal diseases in dogs: the power of within- and across-breed studies. *Mamm Genome.* 2012;23:40-61.
4. Guziewicz KE, Zangerl B, Lindauer SJ, et al. Bestrophin gene mutations cause canine multifocal retinopathy: a novel animal model for best disease. *Invest Ophthalmol Vis Sci.* 2007;48:1959-1967.

# OCULAR DISORDERS REPORT MASTIFF

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 0.110 microphthalmia  |                     | 19        | 0.2% | 3         | 0.4% |
| 10.000 glaucoma   |                     | 2         | 0.0% | 0         |      |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 20.160 macropalpebral fissure   |                     | 344       | 3.9% | 0         |      |
| 21.000 entropion, unspecified   |                     | 387       | 4.4% | 48        | 6.3% |
| 22.000 ectropion, unspecified   |                     | 622       | 7.1% | 62        | 8.1% |
| 25.110 distichiasis   |                     | 92        | 1.0% | 4         | 0.5% |
| <b>NASOLACRIMAL</b>   |                     |           |      |           |      |
| 40.910 keratoconjunctivitis sicca                                     |                     | 4         | 0.0% | 3         | 0.4% |
| <b>NICTITANS</b>  |                     |           |      |           |      |
| 51.100 third eyelid cartilage anomaly                                 |                     | 11        | 0.1% | 1         | 0.1% |
| 52.110 prolapsed gland of the third eyelid                            |                     | 18        | 0.2% | 2         | 0.3% |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.210 corneal pannus   |                     | 3         | 0.0% | 0         |      |
| 70.220 pigmentary keratitis   |                     | 3         | 0.0% | 1         | 0.1% |
| 70.700 corneal dystrophy  |                     | 37        | 0.4% | 3         | 0.4% |
| 70.730 corneal endothelial degeneration                               |                     | 51        | 0.6% | 1         | 0.1% |
| <b>UVEA</b>   |                     |           |      |           |      |
| 90.250 pigmentary uveitis   |                     | 1         | 0.0% | 0         |      |
| 93.140 corneal endothelial pigment without PPM                        |                     | 7         | 0.1% | 0         |      |
| 93.150 iris coloboma  |                     | 3         | 0.0% | 0         |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 274       | 3.1% | 25        | 3.3% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 59        | 0.7% | 3         | 0.4% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 455       | 5.2% | 20        | 2.6% |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 19        | 0.2% | 0         |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 7         | 0.1% | 3         | 0.4% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 40        | 0.5% | 13        | 1.7% |
| 93.810 uveal melanoma   |                     | 3         | 0.0% | 0         |      |
| 93.999 uveal cysts  |                     | 94        | 1.1% | 12        | 1.6% |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.200 cataract, unspecified   |                     | 19        | 0.2% | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 402       | 4.6% | 36        | 4.7% |
| 100.301 punctate cataract, anterior cortex                            |                     | 64        | 0.7% | 8         | 1.1% |
| 100.302 punctate cataract, posterior cortex                           |                     | 12        | 0.1% | 3         | 0.4% |
| 100.303 punctate cataract, equatorial cortex                          |                     | 6         | 0.1% | 0         |      |
| 100.304 punctate cataract, anterior sutures                           |                     | 12        | 0.1% | 0         |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 10        | 0.1% | 1         | 0.1% |
| 100.306 punctate cataract, nucleus                                    |                     | 12        | 0.1% | 3         | 0.4% |
| 100.307 punctate cataract, capsular                                   |                     | 15        | 0.2% | 5         | 0.7% |
| 100.311 incipient cataract, anterior cortex                           |                     | 69        | 0.8% | 7         | 0.9% |
| 100.312 incipient cataract, posterior cortex                          |                     | 42        | 0.5% | 2         | 0.3% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 20        | 0.2% | 3         | 0.4% |
| 100.314 incipient cataract, anterior sutures                          |                     | 8         | 0.1% | 0         |      |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.315 incipient cataract, posterior sutures             | 6 0.1%           | 1 0.1%           |
| 100.316 incipient cataract, nucleus                       | 36 0.4%          | 8 1.1%           |
| 100.317 incipient cataract, capsular                      | 10 0.1%          | 2 0.3%           |
| 100.321 incomplete cataract, anterior cortex              | 1 0.0%           | 2 0.3%           |
| 100.322 incomplete cataract, posterior cortex             | 0                | 1 0.1%           |
| 100.326 incomplete cataract, nucleus                      | 1 0.0%           | 1 0.1%           |
| 100.327 incomplete cataract, capsular                     | 1 0.0%           | 0                |
| 100.328 y-suture tip opacities                            | 1 0.0%           | 3 0.4%           |
| 100.330 generalized/complete cataract                     | 40 0.5%          | 1 0.1%           |
| 100.340 resorbing/hypermature cataract                    | 0                | 1 0.1%           |
| 100.345 <i>significant cataracts (summary)</i>            | 385 4.4%         | 52 6.8%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 5 0.1%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 9 0.1%           | 1 0.1%           |
| 110.135 PHPV/PTVL   | 5 0.1%           | 0                |
| 110.320 vitreal degeneration                              | 11 0.1%          | 0                |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 653 7.4%         | 33 4.3%          |
| 120.180 retinal dysplasia, geographic                     | 51 0.6%          | 1 0.1%           |
| 120.190 retinal dysplasia, detached                       | 5 0.1%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 151 1.7%         | 0                |
| 120.910 retinal detachment without dialysis               | 4 0.0%           | 0                |
| 120.920 retinal detachment with dialysis                  | 1 0.0%           | 0                |
| 120.960 retinopathy                                       | 8 0.1%           | 2 0.3%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 4 0.0%           | 0                |
| 130.120 optic nerve hypoplasia                            | 2 0.0%           | 0                |
| 130.150 optic disc coloboma                               | 4 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 59 0.7%          | 0                |
| 900.100 other, not inherited                              | 164 1.9%         | 5 0.7%           |
| 900.110 other. suspect not inherited/significance unknown | 92 1.0%          | 28 3.7%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 6,007 68.1%      | 490 64.4%        |

## MI-KI

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE             |
|----|---|------------------------|-----------|-----------------|-------------------------------------|
| A. | Distichiasis  | Not defined            | 1         | Breeder option  |                                     |
| B. | Corneal dystrophy<br>- epithelial/stromal           | Not defined            | 1         | Breeder option  |                                     |
| C. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined            | 1         | Breeder option  |                                     |
| D. | Cataract  | Not defined            | 1         | NO              |                                     |
| E. | Y-suture tip opacity                                | Not defined            | 1         | Breeder option  |                                     |
| F. | Vitreous degeneration                               | Not defined            | 1         | Breeder option  |                                     |
| G. | Retinal atrophy ( <i>prcd</i> )                     | Autosomal<br>recessive | 1         | NO              | Mutation in the<br><i>prcd</i> gene |

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### Description and Comments

#### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make strong recommendations with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

#### B. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral. In the Mi-Ki, lesions are circular or semicircular central crystalline deposits in the anterior corneal stroma that appear between 2 and 5 years of age. It may be associated with exophthalmos and lagophthalmos common in these dogs.

#### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose

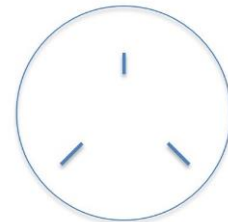
the greatest threat to vision and when severe, vision impairment or blindness may occur.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

F. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

G. Retinal atrophy – *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Mi-Ki is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Mi-Ki breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.



# OCULAR DISORDERS REPORT

## MI-KI

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|---------------------|--|-----------|-------|-----------|-------|
|                     |  | #         | %     | #         | %     |
| <b>EYELIDS</b>      |  |           |       |           |       |
| 20.140              | ectopic cilia  | 1         | 0.1%  | 0         |       |
| 20.160              | macropalpebral fissure                                       | 2         | 0.1%  | 0         |       |
| 21.000              | entropion, unspecified                                       | 10        | 0.7%  | 1         | 0.2%  |
| 25.110              | distichiasis   | 188       | 13.9% | 65        | 15.4% |
| <b>NASOLACRIMAL</b> |  |           |       |           |       |
| 40.910              | keratoconjunctivitis sicca                                   | 4         | 0.3%  | 0         |       |
| <b>NICTITANS</b>    |  |           |       |           |       |
| 52.110              | prolapsed gland of the third eyelid                          | 1         | 0.1%  | 2         | 0.5%  |
| <b>CORNEA</b>       |  |           |       |           |       |
| 70.210              | corneal pannus   | 1         | 0.1%  | 0         |       |
| 70.220              | pigmentary keratitis   | 3         | 0.2%  | 2         | 0.5%  |
| 70.700              | corneal dystrophy  | 24        | 1.8%  | 4         | 0.9%  |
| 70.730              | corneal endothelial degeneration                             | 1         | 0.1%  | 0         |       |
| <b>UVEA</b>         |  |           |       |           |       |
| 93.710              | persistent pupillary membranes, iris to iris                 | 171       | 12.6% | 26        | 6.2%  |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands | 2         | 0.1%  | 1         | 0.2%  |
| <b>LENS</b>         |  |           |       |           |       |
| 100.200             | cataract, unspecified  | 1         | 0.1%  | 0         |       |
| 100.210             | cataract. suspect not inherited/significance unknown         | 117       | 8.6%  | 23        | 5.5%  |
| 100.301             | punctate cataract, anterior cortex                           | 5         | 0.4%  | 1         | 0.2%  |
| 100.302             | punctate cataract, posterior cortex                          | 5         | 0.4%  | 0         |       |
| 100.303             | punctate cataract, equatorial cortex                         | 0         |       | 1         | 0.2%  |
| 100.305             | punctate cataract, posterior sutures                         | 25        | 1.8%  | 1         | 0.2%  |
| 100.306             | punctate cataract, nucleus                                   | 0         |       | 1         | 0.2%  |
| 100.311             | incipient cataract, anterior cortex                          | 3         | 0.2%  | 4         | 0.9%  |
| 100.312             | incipient cataract, posterior cortex                         | 6         | 0.4%  | 0         |       |
| 100.313             | incipient cataract, equatorial cortex                        | 11        | 0.8%  | 1         | 0.2%  |
| 100.314             | incipient cataract, anterior sutures                         | 1         | 0.1%  | 0         |       |
| 100.315             | incipient cataract, posterior sutures                        | 21        | 1.6%  | 0         |       |
| 100.316             | incipient cataract, nucleus                                  | 1         | 0.1%  | 1         | 0.2%  |
| 100.317             | incipient cataract, capsular                                 | 0         |       | 1         | 0.2%  |
| 100.322             | incomplete cataract, posterior cortex                        | 0         |       | 1         | 0.2%  |
| 100.327             | incomplete cataract, capsular                                | 0         |       | 1         | 0.2%  |
| 100.328             | y-suture tip opacities                                       | 8         | 0.6%  | 1         | 0.2%  |
| 100.330             | generalized/complete cataract                                | 1         | 0.1%  | 0         |       |
| 100.345             | significant cataracts (summary)                              | 88        | 6.5%  | 14        | 3.3%  |
| <b>VITREOUS</b>     |  |           |       |           |       |
| 110.120             | persistent hyaloid artery/remnant                            | 1         | 0.1%  | 1         | 0.2%  |
| 110.135             | PHPV/PTVL  | 1         | 0.1%  | 0         |       |
| 110.200             | vitreal degeneration-anterior chamber                        | 4         | 0.3%  | 7         | 1.7%  |
| 110.320             | vitreal degeneration   | 123       | 9.1%  | 22        | 5.2%  |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>FUNDUS</b>   |           |           |
| 97.110 choroidal hypoplasia                               | 1 0.1%    | 0         |
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 11 0.8%   | 2 0.5%    |
| 120.180 retinal dysplasia, geographic                     | 8 0.6%    | 1 0.2%    |
| 120.310 generalized progressive retinal atrophy (PRA)     | 5 0.4%    | 2 0.5%    |
| 120.920 retinal detachment with dialysis                  | 1 0.1%    | 1 0.2%    |
| 120.960 retinopathy                                       | 6 0.4%    | 6 1.4%    |
| <b>OPTIC NERVE</b>  |           |           |
| 130.110 micropapilla                                      | 2 0.1%    | 1 0.2%    |
| 130.120 optic nerve hypoplasia                            | 2 0.1%    | 0         |
| 130.150 optic disc coloboma                               | 2 0.1%    | 0         |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 24 1.8%   | 0         |
| 900.100 other, not inherited                              | 55 4.1%   | 2 0.5%    |
| 900.110 other. suspect not inherited/significance unknown | 33 2.4%   | 31 7.3%   |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 863 63.7% | 263 62.3% |

## MINIATURE AMERICAN SHEPHERD (AKC)/ MINIATURE AUSTRALIAN SHEPHERD

It is recommended that this breed be examined prior to pharmacological dilation to best facilitate identification of iris coloboma.

|    | DISORDER                                      | INHERITANCE   | REFERENCE | BREEDING<br>ADVICE | GENETIC<br>TESTS<br>AVAILABLE     |
|----|---|---|-----------|--------------------|-----------------------------------|
| A. | Microphthalmia with multiple ocular defects   | Presumed autosomal recessive with incomplete penetrance | 1-6       | NO                 |                                   |
| B. | Distichiasis                                  | Not defined   | 1         | Breeder option     |                                   |
| C. | Corneal dystrophy - epithelial/stromal        | Not defined   | 1         | Breeder option     |                                   |
| D. | Iris hypoplasia                               | Not defined   | 1         | Breeder option     |                                   |
| E. | Persistent pupillary membranes - iris to iris | Not defined   | 1         | Breeder option     |                                   |
| F. | Cataract                                      | Autosomal co-dominant                                   | 1, 7, 8   | NO                 | Mutation of the <i>HSF4</i> gene  |
| G. | Retinal atrophy ( <i>prcd</i> )               | Autosomal recessive                                     | 1, 9      | NO                 | Mutation of the <i>prcd</i> gene  |
| H. | Cone degeneration - day blindness             | Autosomal recessive                                     | 10        | NO                 | Mutation of the <i>CNGB3</i> gene |
| I. | Multifocal retinopathy - <i>cmr1</i>          | Autosomal recessive                                     | 11        | Breeder option     | Mutation of the <i>BEST1</i> gene |

|    | <b>DISORDER</b>   | <b>INHERITANCE</b>     | <b>REFERENCE</b> | <b>BREEDING<br/>ADVICE</b> | <b>GENETIC TESTS<br/>AVAILABLE</b>   |
|----|---|------------------------|------------------|----------------------------|--------------------------------------|
| J. | Choroidal hypoplasia<br>(Collie Eye Anomaly)<br>- optic nerve coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- staphyloma/coloboma | Autosomal<br>recessive | 1, 12-15         | NO                         | Mutation of the<br><i>NHEJ1</i> gene |
| K. | Coloboma/staphyloma<br>without microphthalmia   | Not defined            | 1                | NO                         |                                      |

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## Description and Comments

### A. Microphthalmia with multiple ocular defects

Microphthalmia is a congenital defect characterized by a small eye with associated defects of the cornea, iris (coloboma), anterior chamber, lens (cataract) and/or retina (dysplasia). In the Australian Shepherd, microphthalmia has long been suspected to be associated with merled coat coloration but a definitive genetic relationship has not been established. The eyes of affected homozygous merle (usually white) dogs have extreme forms of this entity and are usually blind at birth. Affected heterozygous merle-coated dogs demonstrate less severe manifestations.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### C. Corneal Dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

### D. Iris hypoplasia

A congenital abnormality in iris development usually characterized by a reduced quantity of tissue identified as a partial-thickness defect in iris tissue. Full-thickness iris hypoplasia is rare and should be recorded as an iris coloboma on the OFA form.

### E. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress

normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

F. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Australian Shepherd, a mutation in *HSF4* (heat shock transcription factor 4), the HSF4-2 mutation, has been shown to increase the likelihood of cataract formation. The mutation is inherited in a co-dominant manner. Dogs with one copy of the mutation develop bilateral posterior cataracts and homozygotes develop a nuclear cataract that typically progresses to a mature cataract. A DNA test is available for this mutation. Other genetic factors can contribute to cataract formation in this breed and will not be detected by this test.

G. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Miniature American/Australian Shepherd is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

H. Cone degeneration - day blindness or hemeralopia

Autosomal recessively inherited early degeneration of the cone photoreceptors. Affected puppies develop day blindness, color blindness, and photophobia between 8 and 12 weeks of age. Affected dogs remain ophthalmoscopically normal their entire life. Electroretinography is required to definitively diagnose the disorder. Genetically, the condition results from a mutation in the *CNGB3* gene. A DNA test is available.

I. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (*cmr1*) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal

detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff.

- J. Choroidal hypoplasia (Collie Eye Anomaly)
- staphyloma/coloboma
  - retinal detachment
  - retinal hemorrhage
  - optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

- K. Coloboma/staphyloma (unassociated with microphthalmia)

A coloboma is a congenital defect which may affect the iris, choroid or optic disc. Iris colobomas are seen as notches in the pupillary margin. Scleral ectasia is defined as a congenital thinning and secondary distention of the sclera; when lined by uveal tissue it is called a staphyloma. These may be anteriorly located, apparent as a bulge beneath the upper eyelid or posteriorly located, requiring visualization with an ophthalmoscope. These conditions may or may not be genetically related to the same anomalies seen associated with microphthalmia (entity "A" above).

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# OCULAR DISORDERS REPORT

## MINIATURE AMERICAN(AKC)/MINIATURE AUSTRALIAN SHEPHERD

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>13,491 |      | 2016-2020<br>5,368 |       |
|---------------------|--|---------------------|------|--------------------|-------|
|                     |  | #                   | %    | #                  | %     |
| <b>GLOBE</b>        |  |                     |      |                    |       |
| 0.110               | microphthalmia   | 18                  | 0.1% | 9                  | 0.2%  |
| 10.000              | glaucoma   | 1                   | 0.0% | 0                  |       |
| <b>EYELIDS</b>      |  |                     |      |                    |       |
| 20.140              | ectopic cilia  | 0                   |      | 1                  | 0.0%  |
| 25.110              | distichiasis   | 639                 | 4.7% | 145                | 2.7%  |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 1                   | 0.0% | 2                  | 0.0%  |
| 40.910              | keratoconjunctivitis sicca                                     | 1                   | 0.0% | 1                  | 0.0%  |
| <b>NICTITANS</b>    |  |                     |      |                    |       |
| 51.100              | third eyelid cartilage anomaly                                 | 2                   | 0.0% | 0                  |       |
| <b>CORNEA</b>       |  |                     |      |                    |       |
| 70.220              | pigmentary keratitis   | 2                   | 0.0% | 0                  |       |
| 70.700              | corneal dystrophy  | 92                  | 0.7% | 116                | 2.2%  |
| 70.730              | corneal endothelial degeneration                               | 5                   | 0.0% | 1                  | 0.0%  |
| <b>UVEA</b>         |  |                     |      |                    |       |
| 90.250              | pigmentary uveitis   | 1                   | 0.0% | 0                  |       |
| 93.110              | iris hypoplasia  | 51                  | 0.4% | 83                 | 1.5%  |
| 93.150              | iris coloboma  | 259                 | 1.9% | 111                | 2.1%  |
| 93.180              | liris sphincter dysplasia                                      | 3                   | 0.0% | 11                 | 0.2%  |
| 93.710              | persistent pupillary membranes, iris to iris                   | 1,229               | 9.1% | 707                | 13.2% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 19                  | 0.1% | 17                 | 0.3%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 7                   | 0.1% | 2                  | 0.0%  |
| 93.740              | persistent pupillary membranes, iris sheets                    | 9                   | 0.1% | 0                  |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 2                   | 0.0% | 0                  |       |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 3                   | 0.0% | 1                  | 0.0%  |
| 93.810              | uveal melanoma   | 1                   | 0.0% | 0                  |       |
| 93.999              | uveal cysts  | 0                   |      | 1                  | 0.0%  |
| 97.150              | chorioretinal coloboma, congenital                             | 5                   | 0.0% | 2                  | 0.0%  |
| <b>LENS</b>         |  |                     |      |                    |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 149                 | 1.1% | 61                 | 1.1%  |
| 100.301             | punctate cataract, anterior cortex                             | 19                  | 0.1% | 9                  | 0.2%  |
| 100.302             | punctate cataract, posterior cortex                            | 8                   | 0.1% | 0                  |       |
| 100.303             | punctate cataract, equatorial cortex                           | 6                   | 0.0% | 1                  | 0.0%  |
| 100.304             | punctate cataract, anterior sutures                            | 3                   | 0.0% | 0                  |       |
| 100.305             | punctate cataract, posterior sutures                           | 8                   | 0.1% | 8                  | 0.1%  |
| 100.306             | punctate cataract, nucleus                                     | 4                   | 0.0% | 6                  | 0.1%  |
| 100.307             | punctate cataract, capsular                                    | 7                   | 0.1% | 2                  | 0.0%  |
| 100.311             | incipient cataract, anterior cortex                            | 22                  | 0.2% | 11                 | 0.2%  |
| 100.312             | incipient cataract, posterior cortex                           | 25                  | 0.2% | 5                  | 0.1%  |
| 100.313             | incipient cataract, equatorial cortex                          | 7                   | 0.1% | 5                  | 0.1%  |
| 100.315             | incipient cataract, posterior sutures                          | 1                   | 0.0% | 1                  | 0.0%  |
| 100.316             | incipient cataract, nucleus                                    | 5                   | 0.0% | 2                  | 0.0%  |
| 100.317             | incipient cataract, capsular                                   | 6                   | 0.0% | 8                  | 0.1%  |



| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.321 incomplete cataract, anterior cortex              | 0                |       | 2                | 0.0%  |
| 100.322 incomplete cataract, posterior cortex             | 1                | 0.0%  | 2                | 0.0%  |
| 100.323 incomplete cataract, equatorial cortex            | 0                |       | 2                | 0.0%  |
| 100.327 incomplete cataract, capsular                     | 1                | 0.0%  | 1                | 0.0%  |
| 100.328 y-suture tip opacities                            | 1                | 0.0%  | 12               | 0.2%  |
| 100.330 generalized/complete cataract                     | 5                | 0.0%  | 1                | 0.0%  |
| 100.345 <i>significant cataracts (summary)</i>            | 129              | 1.0%  | 78               | 1.5%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 1                | 0.0%  | 0                |       |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 40               | 0.3%  | 46               | 0.9%  |
| 110.135 PHPV/PTVL   | 13               | 0.1%  | 2                | 0.0%  |
| 110.200 vitreous degeneration-anterior chamber            | 5                | 0.0%  | 7                | 0.1%  |
| 110.320 vitreal degeneration                              | 70               | 0.5%  | 12               | 0.2%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 24               | 0.2%  | 13               | 0.2%  |
| 97.120 coloboma   | 8                | 0.1%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 45               | 0.3%  | 12               | 0.2%  |
| 120.180 retinal dysplasia, geographic                     | 1                | 0.0%  | 0                |       |
| 120.190 retinal dysplasia, detached                       | 1                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 28               | 0.2%  | 1                | 0.0%  |
| 120.910 retinal detachment without dialysis               | 1                | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 0                |       | 1                | 0.0%  |
| 120.960 retinopathy                                       | 2                | 0.0%  | 3                | 0.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 65               | 0.5%  | 14               | 0.3%  |
| 130.120 optic nerve hypoplasia                            | 19               | 0.1%  | 4                | 0.1%  |
| 130.150 optic disc coloboma                               | 23               | 0.2%  | 8                | 0.1%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 129              | 1.0%  | 0                |       |
| 900.100 other, not inherited                              | 180              | 1.3%  | 4                | 0.1%  |
| 900.110 other. suspect not inherited/significance unknown | 85               | 0.6%  | 146              | 2.7%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 11,433           | 84.7% | 4,003            | 74.6% |

# MINIATURE BULL TERRIER

|    | DISORDER                       | INHERITANCE         | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE           |
|----|--------------------------------|---------------------|-----------|--------------------|--------------------------------------|
| A. | Persistent pupillary membranes |                     |           |                    |                                      |
|    | - iris to iris                 | Not defined         | 1         | Breeder option     |                                      |
|    | - iris to lens                 | Not defined         | 1         | NO                 |                                      |
|    | - iris to cornea               | Not defined         | 1         | NO                 |                                      |
| B. | Cataract                       | Not defined         | 1         | NO                 |                                      |
| C. | Lens luxation                  | Autosomal recessive | 2, 3      | NO                 | Mutation of the <i>ADAMTS17</i> gene |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Although the total number of Miniature Bull Terriers presented for OFA/CERF examination is not large, the incidence of PPM in this breed is approximately 10% in recent years. Some of these PPM's have been iris to cornea and iris to lens. Considerable discretion should be used before breeding a dog with the latter more severe forms of PPM.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

Two loci with potentially enhancing effects on the ADAMTS17 mutation are associated with primary lens luxation (PLL) in Australian Miniature Bull Terriers. PLL associated allele of the BICF2G630420272 SNP increases the risk of PLL in the presence of the ADAMTS17 mutation. Candidate genes in the two regions of interest included CPE on chromosome 15 and CTCF on chromosome 1. The ADAMTS17 mutation is also associated with abnormal foot and nail shapes, pedal hyperkeratosis, and persistent pupillary membranes. Association of the ADAMTS17 mutation with possible pedal skeletal abnormalities in the Miniature Bull Terriers supports primary lens luxation in this breed and Marchesani syndrome-like disease in humans as being homologous diseases.

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# OCULAR DISORDERS REPORT MINIATURE BULL TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>1,228 |      | 2016-2020<br>105 |      |
|---|---------------------|--------------------|------|------------------|------|
|   |                     | #                  | %    | #                | %    |
| <b>GLOBE</b>  |                     |                    |      |                  |      |
| 0.110 microphthalmia  |                     | 3                  | 0.2% | 0                |      |
| 10.000 glaucoma   |                     | 1                  | 0.1% | 0                |      |
| <b>EYELIDS</b>  |                     |                    |      |                  |      |
| 22.000 ectropion, unspecified   |                     | 1                  | 0.1% | 0                |      |
| 25.110 distichiasis   |                     | 0                  |      | 1                | 1.0% |
| <b>NASOLACRIMAL</b>   |                     |                    |      |                  |      |
| 40.910 keratoconjunctivitis sicca                                     |                     | 5                  | 0.4% | 1                | 1.0% |
| <b>CORNEA</b>   |                     |                    |      |                  |      |
| 70.700 corneal dystrophy  |                     | 3                  | 0.2% | 2                | 1.9% |
| 70.730 corneal endothelial degeneration                               |                     | 13                 | 1.1% | 0                |      |
| <b>UVEA</b>   |                     |                    |      |                  |      |
| 93.140 corneal endothelial pigment without PPM                        |                     | 4                  | 0.3% | 0                |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 80                 | 6.5% | 0                |      |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 52                 | 4.2% | 0                |      |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 81                 | 6.6% | 2                | 1.9% |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 8                  | 0.7% | 0                |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 6                  | 0.5% | 3                | 2.9% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 14                 | 1.1% | 2                | 1.9% |
| <b>LENS</b>   |                     |                    |      |                  |      |
| 100.200 cataract, unspecified   |                     | 2                  | 0.2% | 0                |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 51                 | 4.2% | 4                | 3.8% |
| 100.301 punctate cataract, anterior cortex                            |                     | 11                 | 0.9% | 0                |      |
| 100.302 punctate cataract, posterior cortex                           |                     | 1                  | 0.1% | 0                |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 1                  | 0.1% | 0                |      |
| 100.307 punctate cataract, capsular                                   |                     | 4                  | 0.3% | 0                |      |
| 100.311 incipient cataract, anterior cortex                           |                     | 15                 | 1.2% | 0                |      |
| 100.312 incipient cataract, posterior cortex                          |                     | 5                  | 0.4% | 0                |      |
| 100.313 incipient cataract, equatorial cortex                         |                     | 1                  | 0.1% | 0                |      |
| 100.314 incipient cataract, anterior sutures                          |                     | 1                  | 0.1% | 0                |      |
| 100.317 incipient cataract, capsular                                  |                     | 12                 | 1.0% | 0                |      |
| 100.330 generalized/complete cataract                                 |                     | 4                  | 0.3% | 0                |      |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 57                 | 4.6% | 0                |      |
| 100.375 <i>subluxation/luxation, unspecified</i>                      |                     | 51                 | 4.2% | 0                |      |
| <b>VITREOUS</b>   |                     |                    |      |                  |      |
| 110.120 persistent hyaloid artery/remnant                             |                     | 1                  | 0.1% | 0                |      |
| 110.200 vitreous degeneration-anterior chamber                        |                     | 0                  |      | 1                | 1.0% |
| 110.320 vitreal degeneration  |                     | 24                 | 2.0% | 1                | 1.0% |
| <b>RETINA</b>   |                     |                    |      |                  |      |
| 120.170 retinal dysplasia, folds                                      |                     | 3                  | 0.2% | 0                |      |
| 120.180 retinal dysplasia, geographic                                 |                     | 1                  | 0.1% | 0                |      |
| 120.310 generalized progressive retinal atrophy (PRA)                 |                     | 13                 | 1.1% | 0                |      |
| 120.960 retinopathy   |                     | 0                  |      | 2                | 1.9% |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>OPTIC NERVE</b>  |           |           |
| 130.110 micropapilla                                      | 12 1.0%   | 0         |
| 130.120 optic nerve hypoplasia                            | 3 0.2%    | 0         |
| 130.150 optic disc coloboma                               | 1 0.1%    | 0         |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 9 0.7%    | 0         |
| 900.100 other, not inherited                              | 33 2.7%   | 0         |
| 900.110 other. suspect not inherited/significance unknown | 22 1.8%   | 2 1.9%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 909 74.0% | 88 83.8%  |

## MINIATURE PINSCHER

|    | DISORDER   | INHERITANCE                | REFERENCE | BREEDING ADVICE                           |
|----|--|----------------------------|-----------|---|
| A. | Corneal dystrophy<br>- epithelial/stromal  | Not defined                | 1         | Breeder option                            |
| B. | Persistent pupillary<br>membranes<br>- iris to iris<br>- lens pigment foci/no<br>strands | Not defined<br>Not defined | 1<br>1    | Breeder option<br>Passes with no notation |
| C. | Cataract   | Not defined                | 1         | NO  |
| D. | Vitreous degeneration  | Not defined                | 1         | Breeder option                            |

### Description and Comments

**A. Corneal dystrophy - epithelial/stromal**

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

**B. Persistent pupillary membranes (PPMs)**

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

**C. Cataract**

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

**D. Vitreous degeneration**

A liquefaction of the vitreous gel which may predispose to retinal detachment.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Miniature Pinscher. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT MINIATURE PINSCHER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 0.110 microphthalmia  |                     | 3         | 0.4% | 0         |      |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 20.140 ectopic cilia  |                     | 1         | 0.1% | 0         |      |
| 21.000 entropion, unspecified   |                     | 3         | 0.4% | 0         |      |
| 22.000 ectropion, unspecified   |                     | 1         | 0.1% | 0         |      |
| 25.110 distichiasis   |                     | 5         | 0.7% | 1         | 0.3% |
| <b>NASOLACRIMAL</b>   |                     |           |      |           |      |
| 40.910 keratoconjunctivitis sicca                                     |                     | 0         |      | 1         | 0.3% |
| <b>NICTITANS</b>  |                     |           |      |           |      |
| 52.110 prolapsed gland of the third eyelid                            |                     | 2         | 0.3% | 0         |      |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.210 corneal pannus   |                     | 2         | 0.3% | 0         |      |
| 70.220 pigmentary keratitis   |                     | 2         | 0.3% | 4         | 1.3% |
| 70.700 corneal dystrophy  |                     | 41        | 5.5% | 16        | 5.1% |
| 70.730 corneal endothelial degeneration                               |                     | 2         | 0.3% | 0         |      |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.140 corneal endothelial pigment without PPM                        |                     | 1         | 0.1% | 0         |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 25        | 3.4% | 5         | 1.6% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 1         | 0.1% | 0         |      |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 0         |      | 1         | 0.3% |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 1         | 0.1% | 0         |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 5         | 0.7% | 9         | 2.9% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1         | 0.1% | 3         | 1.0% |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 29        | 3.9% | 6         | 1.9% |
| 100.301 punctate cataract, anterior cortex                            |                     | 6         | 0.8% | 2         | 0.6% |
| 100.302 punctate cataract, posterior cortex                           |                     | 5         | 0.7% | 0         |      |
| 100.303 punctate cataract, equatorial cortex                          |                     | 1         | 0.1% | 1         | 0.3% |
| 100.304 punctate cataract, anterior sutures                           |                     | 1         | 0.1% | 0         |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 3         | 0.4% | 0         |      |
| 100.306 punctate cataract, nucleus                                    |                     | 0         |      | 1         | 0.3% |
| 100.307 punctate cataract, capsular                                   |                     | 1         | 0.1% | 1         | 0.3% |
| 100.311 incipient cataract, anterior cortex                           |                     | 16        | 2.1% | 8         | 2.6% |
| 100.312 incipient cataract, posterior cortex                          |                     | 10        | 1.3% | 1         | 0.3% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 3         | 0.4% | 0         |      |
| 100.314 incipient cataract, anterior sutures                          |                     | 0         |      | 1         | 0.3% |
| 100.315 incipient cataract, posterior sutures                         |                     | 1         | 0.1% | 0         |      |
| 100.317 incipient cataract, capsular                                  |                     | 1         | 0.1% | 0         |      |
| 100.321 incomplete cataract, anterior cortex                          |                     | 1         | 0.1% | 3         | 1.0% |
| 100.322 incomplete cataract, posterior cortex                         |                     | 1         | 0.1% | 2         | 0.6% |
| 100.323 incomplete cataract, equatorial cortex                        |                     | 0         |      | 2         | 0.6% |
| 100.330 generalized/complete cataract                                 |                     | 7         | 0.9% | 0         |      |
| 100.340 resorbing/hypermature cataract                                |                     | 0         |      | 1         | 0.3% |



| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.345 significant cataracts (summary)                   | 57 7.6%          | 23 7.4%          |
| 100.375 subluxation/luxation, unspecified                 | 3 0.4%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 4 0.5%           | 1 0.3%           |
| 110.135 PHPV/PTVL   | 2 0.3%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 2 0.3%           | 1 0.3%           |
| 110.320 vitreal degeneration                              | 41 5.5%          | 2 0.6%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.120 coloboma   | 1 0.1%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 2 0.3%           | 1 0.3%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 12 1.6%          | 0                |
| 120.910 retinal detachment without dialysis               | 3 0.4%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 0                | 3 1.0%           |
| 130.120 optic nerve hypoplasia                            | 9 1.2%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 12 1.6%          | 0                |
| 900.100 other, not inherited                              | 26 3.5%          | 1 0.3%           |
| 900.110 other. suspect not inherited/significance unknown | 9 1.2%           | 23 7.4%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 562 75.3%        | 229 73.6%        |

# MINIATURE SCHNAUZER

|    | DISORDER   | INHERITANCE                  | REFERENCE | BREEDING ADVICE                           |
|----|--|------------------------------|-----------|---|
| A. | Microphthalmia with congenital cataract  | Autosomal recessive          | 1-4       | NO  |
| B. | Distichiasis   | Not defined                  | 1         | Breeder option                            |
| C. | Corneal dystrophy - epithelial/stromal   | Not defined                  | 1         | Breeder option                            |
| D. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined   | 1<br>1    | Breeder option<br>Passes with no notation |
| E. | Cataract   | Autosomal recessive          | 1, 5-8    | NO  |
| F. | Vitreous degeneration  | Not defined                  | 1         | Breeder option                            |
| G. | Retinal dysplasia with Persistent hyperplastic primary vitreous (PHPV)             | Autosomal recessive          | 11        | NO  |
| H. | Retinal atrophy-generalized  | Autosomal recessive          | 1, 9, 10  | NO  |
| I. | Ceroid lipofuscinosis  | Presumed autosomal recessive | 12, 13    | NO  |

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## Description and Comments

### A. Microphthalmia with congenital cataract

Congenital nuclear and posterior cortical lens opacities that progress slowly. In some cases, these cataracts appear similar to the congenital cataracts described in "E" below. An associated abnormality in this defect is microphthalmia that is often mild and is accompanied by a 1-3 mm reduction in the axial length of the globe as determined by ultrasonography. The cataracts often do not become mature and cause blindness until the dogs reach 3-5 years of age. Congenital cataracts and microphthalmia are inherited as an autosomal recessive disorder.

B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

Congenital cataracts in the Miniature Schnauzer are bilateral and appear prior to 6 weeks of age. At this time they may already involve the entire lens. Others will first appear as posterior subcapsular opacities and usually progress to complete cataracts. These congenital cataracts are inherited as an autosomal recessive trait. Later-onset cataracts may represent a genetically distinct entity. There are other types of cataract in the breed which are also likely hereditary.

Note: It is not certain whether A and F are genetically distinct, or different manifestations of the same entity, as eyes affected with cataracts are often smaller than normal.

F. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

G. Retinal dysplasia with persistent hyperplastic primary vitreous (PHPV)

In the Miniature Schnauzer PHPV is associated with retinal dysplasia in some dogs. In this association it may be unilateral or bilateral and most often manifests as small white

posterior lens capsule plaques accompanied by white primary vitreous mass extending to the optic disc. Patent hyaloid arteries and posterior lens capsule vessels may also be present.

#### H. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most forms of PRA are inherited as recessive traits.

A form of PRA in the Miniature Schnauzer was previously characterized and called photoreceptor dysplasia (now called Type A PRA). The dysplasia results from the abnormal development of visual cells followed by their degeneration. The disorder appears to affect the generation of an electrical signal within the retinal photoreceptor cells. Although fundus abnormalities usually are not present until 2-3 years of age, abnormalities of the electroretinogram can be demonstrated by 8-10 weeks of age. Clinical signs include mildly impaired night vision and variable rate of progression.

Initial studies suggested a mutation in phosphodiesterase was responsible, but this was disproven. This disease is extremely rare. The causative gene for Type A PRA has not been published although a DNA test is available. Another more common autosomal recessive form of PRA appears to be present in the Miniature Schnauzer, but the causative gene has not yet been determined; it also affects dogs ~2-4 years of age. Lastly, cases of late-onset PRA in the breed are recognized clinically but the inheritance pattern is unknown. (G. Aguirre personal communication 2016).

#### I. Ceroid lipofuscinosis

An inherited disease of man and animals characterized by the accumulation of lipopigment in various tissues of the body including the eye. It results in progressive neurologic disease including blindness. (Also called Batten's disease). This disease is very rare.

## References

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# OCULAR DISORDERS REPORT

## MINIATURE SCHNAUZER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>28,722 |      | 2016-2020<br>5,672 |      |
|---------------------|--|---------------------|------|--------------------|------|
|                     |  | #                   | %    | #                  | %    |
| <b>GLOBE</b>        |  |                     |      |                    |      |
| 0.110               | microphthalmia   | 23                  | 0.1% | 1                  | 0.0% |
| <b>EYELIDS</b>      |  |                     |      |                    |      |
| 21.000              | entropion, unspecified   | 5                   | 0.0% | 9                  | 0.2% |
| 25.110              | distichiasis   | 602                 | 2.1% | 99                 | 1.7% |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 1                   | 0.0% | 2                  | 0.0% |
| 40.910              | keratoconjunctivitis sicca                                     | 6                   | 0.0% | 2                  | 0.0% |
| <b>NICTITANS</b>    |  |                     |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 1                   | 0.0% | 0                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 4                   | 0.0% | 0                  |      |
| <b>CORNEA</b>       |  |                     |      |                    |      |
| 70.210              | corneal pannus   | 2                   | 0.0% | 1                  | 0.0% |
| 70.220              | pigmentary keratitis   | 7                   | 0.0% | 1                  | 0.0% |
| 70.700              | corneal dystrophy  | 149                 | 0.5% | 18                 | 0.3% |
| 70.730              | corneal endothelial degeneration                               | 17                  | 0.1% | 1                  | 0.0% |
| <b>UVEA</b>         |  |                     |      |                    |      |
| 90.250              | pigmentary uveitis   | 2                   | 0.0% | 0                  |      |
| 93.110              | iris hypoplasia  | 0                   |      | 2                  | 0.0% |
| 93.140              | corneal endothelial pigment without PPM                        | 10                  | 0.0% | 0                  |      |
| 93.150              | iris coloboma  | 0                   |      | 1                  | 0.0% |
| 93.710              | persistent pupillary membranes, iris to iris                   | 473                 | 1.6% | 87                 | 1.5% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 49                  | 0.2% | 3                  | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 80                  | 0.3% | 5                  | 0.1% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 12                  | 0.0% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 75                  | 0.3% | 69                 | 1.2% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 13                  | 0.0% | 1                  | 0.0% |
| 93.999              | uveal cysts  | 1                   | 0.0% | 1                  | 0.0% |
| <b>LENS</b>         |  |                     |      |                    |      |
| 100.200             | cataract, unspecified  | 61                  | 0.2% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 593                 | 2.1% | 116                | 2.0% |
| 100.301             | punctate cataract, anterior cortex                             | 94                  | 0.3% | 13                 | 0.2% |
| 100.302             | punctate cataract, posterior cortex                            | 47                  | 0.2% | 6                  | 0.1% |
| 100.303             | punctate cataract, equatorial cortex                           | 31                  | 0.1% | 6                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 15                  | 0.1% | 0                  |      |
| 100.305             | punctate cataract, posterior sutures                           | 58                  | 0.2% | 15                 | 0.3% |
| 100.306             | punctate cataract, nucleus                                     | 15                  | 0.1% | 5                  | 0.1% |
| 100.307             | punctate cataract, capsular                                    | 29                  | 0.1% | 9                  | 0.2% |
| 100.311             | incipient cataract, anterior cortex                            | 103                 | 0.4% | 15                 | 0.3% |
| 100.312             | incipient cataract, posterior cortex                           | 139                 | 0.5% | 22                 | 0.4% |
| 100.313             | incipient cataract, equatorial cortex                          | 61                  | 0.2% | 12                 | 0.2% |
| 100.314             | incipient cataract, anterior sutures                           | 8                   | 0.0% | 1                  | 0.0% |
| 100.315             | incipient cataract, posterior sutures                          | 37                  | 0.1% | 1                  | 0.0% |
| 100.316             | incipient cataract, nucleus                                    | 26                  | 0.1% | 13                 | 0.2% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.317 incipient cataract, capsular                      | 26               | 0.1%  | 5                | 0.1%  |
| 100.321 incomplete cataract, anterior cortex              | 9                | 0.0%  | 10               | 0.2%  |
| 100.322 incomplete cataract, posterior cortex             | 13               | 0.0%  | 11               | 0.2%  |
| 100.323 incomplete cataract, equatorial cortex            | 1                | 0.0%  | 1                | 0.0%  |
| 100.325 incomplete cataract, posterior sutures            | 1                | 0.0%  | 3                | 0.1%  |
| 100.326 incomplete cataract, nucleus                      | 15               | 0.1%  | 14               | 0.2%  |
| 100.327 incomplete cataract, capsular                     | 2                | 0.0%  | 0                |       |
| 100.328 y-suture tip opacities                            | 12               | 0.0%  | 8                | 0.1%  |
| 100.330 generalized/complete cataract                     | 152              | 0.5%  | 4                | 0.1%  |
| 100.340 resorbing/hypermature cataract                    | 1                | 0.0%  | 1                | 0.0%  |
| 100.345 <i>significant cataracts (summary)</i>            | 956              | 3.3%  | 175              | 3.1%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 7                | 0.0%  | 0                |       |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 36               | 0.1%  | 14               | 0.2%  |
| 110.135 PHPV/PTVL   | 23               | 0.1%  | 1                | 0.0%  |
| 110.200 vitreous degeneration-anterior chamber            | 8                | 0.0%  | 10               | 0.2%  |
| 110.320 vitreal degeneration                              | 167              | 0.6%  | 19               | 0.3%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 4                | 0.0%  | 1                | 0.0%  |
| 97.120 coloboma   | 1                | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 66               | 0.2%  | 4                | 0.1%  |
| 120.180 retinal dysplasia, geographic                     | 48               | 0.2%  | 1                | 0.0%  |
| 120.190 retinal dysplasia, detached                       | 32               | 0.1%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 150              | 0.5%  | 3                | 0.1%  |
| 120.400 retinal hemorrhage                                | 6                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 14               | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 1                | 0.0%  | 1                | 0.0%  |
| 120.960 retinopathy                                       | 3                | 0.0%  | 3                | 0.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 43               | 0.1%  | 16               | 0.3%  |
| 130.120 optic nerve hypoplasia                            | 15               | 0.1%  | 2                | 0.0%  |
| 130.150 optic disc coloboma                               | 1                | 0.0%  | 1                | 0.0%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 158              | 0.6%  | 0                |       |
| 900.100 other, not inherited                              | 340              | 1.2%  | 5                | 0.1%  |
| 900.110 other. suspect not inherited/significance unknown | 161              | 0.6%  | 117              | 2.1%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 26,152           | 91.1% | 5,030            | 88.7% |

# MUDI

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |
| B. | Cataract   | Not defined        | 1                | NO                     |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

Any opacity of the lens and/or its capsule, regardless of size or location within the lens. Cataracts are assumed to be hereditary unless associated with known trauma, ocular inflammation, specific metabolic diseases, or nutritional deficiencies.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Mudi breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.



# OCULAR DISORDERS REPORT MUDI

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015<br>58 |       | 2016-2020<br>172 |       |
|-----------------|--|-----------------|-------|------------------|-------|
|                 |  | #               | %     | #                | %     |
| <b>EYELIDS</b>  |  |                 |       |                  |       |
| 25.110          | distichiasis   | 2               | 3.4%  | 0                |       |
| <b>CORNEA</b>   |  |                 |       |                  |       |
| 70.220          | pigmentary keratitis   | 0               |       | 1                | 0.6%  |
| 70.700          | corneal dystrophy  | 0               |       | 1                | 0.6%  |
| <b>UVEA</b>     |  |                 |       |                  |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 5               | 8.6%  | 19               | 11.0% |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 0               |       | 1                | 0.6%  |
| <b>LENS</b>     |  |                 |       |                  |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 2               | 3.4%  | 4                | 2.3%  |
| 100.301         | punctate cataract, anterior cortex                           | 0               |       | 1                | 0.6%  |
| 100.305         | punctate cataract, posterior sutures                         | 0               |       | 4                | 2.3%  |
| 100.316         | incipient cataract, nucleus                                  | 1               | 1.7%  | 1                | 0.6%  |
| 100.328         | y-suture tip opacities                                       | 0               |       | 4                | 2.3%  |
| 100.345         | significant cataracts (summary)                              | 1               | 1.7%  | 10               | 5.8%  |
| <b>OTHER</b>    |  |                 |       |                  |       |
| 900.000         | other, unspecified   | 1               | 1.7%  | 0                |       |
| 900.110         | other. suspect not inherited/significance unknown            | 1               | 1.7%  | 16               | 9.3%  |
| <b>NORMAL</b>   |  |                 |       |                  |       |
| 0.000           | normal globe   | 50              | 86.2% | 129              | 75.0% |

# NEAPOLITAN MASTIFF

|    | DISORDER  | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|-----------|-------------|-----------|-----------------|
| A. | Entropion | Not defined | 1         | Breeder option  |
| B. | Ectropion | Not defined | 1         | Breeder option  |

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## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Ectropion

A conformational defect resulting in eversion of the eyelid(s), which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Neapolitan Mastiff breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT NEAPOLITAN MASTIFF

| Diagnostic Name     | TOTAL DOGS EXAMINED                                  | 1991-2015 |       | 2016-2020 |       |
|---------------------|--|-----------|-------|-----------|-------|
|                     |  | #         | %     | #         | %     |
| <b>EYELIDS</b>      |  |           |       |           |       |
| 20.160              | macropalpebral fissure                               | 14        | 21.2% | 0         |       |
| 21.000              | entropion, unspecified                               | 13        | 19.7% | 13        | 36.1% |
| 22.000              | ectropion, unspecified                               | 22        | 33.3% | 20        | 55.6% |
| 25.110              | distichiasis   | 7         | 10.6% | 4         | 11.1% |
| <b>NASOLACRIMAL</b> |  |           |       |           |       |
| 40.910              | keratoconjunctivitis sicca                           | 1         | 1.5%  | 0         |       |
| <b>NICTITANS</b>    |  |           |       |           |       |
| 51.100              | third eyelid cartilage anomaly                       | 1         | 1.5%  | 1         | 2.8%  |
| 52.110              | prolapsed gland of the third eyelid                  | 4         | 6.1%  | 2         | 5.6%  |
| <b>CORNEA</b>       |  |           |       |           |       |
| 70.220              | pigmentary keratitis                                 | 2         | 3.0%  | 1         | 2.8%  |
| 70.700              | corneal dystrophy                                    | 1         | 1.5%  | 0         |       |
| 70.730              | corneal endothelial degeneration                     | 0         |       | 1         | 2.8%  |
| <b>UVEA</b>         |  |           |       |           |       |
| 93.730              | persistent pupillary membranes, iris to cornea       | 1         | 1.5%  | 0         |       |
| <b>LENS</b>         |  |           |       |           |       |
| 100.210             | cataract. suspect not inherited/significance unknown | 1         | 1.5%  | 3         | 8.3%  |
| 100.306             | punctate cataract, nucleus                           | 1         | 1.5%  | 1         | 2.8%  |
| 100.313             | incipient cataract, equatorial cortex                | 1         | 1.5%  | 0         |       |
| 100.316             | incipient cataract, nucleus                          | 1         | 1.5%  | 0         |       |
| 100.330             | generalized/complete cataract                        | 3         | 4.5%  | 0         |       |
| 100.345             | significant cataracts (summary)                      | 6         | 9.1%  | 1         | 2.8%  |
| <b>RETINA</b>       |  |           |       |           |       |
| 120.170             | retinal dysplasia, folds                             | 2         | 3.0%  | 0         |       |
| 120.960             | retinopathy  | 1         | 1.5%  | 0         |       |
| <b>OTHER</b>        |  |           |       |           |       |
| 900.000             | other, unspecified                                   | 1         | 1.5%  | 0         |       |
| 900.100             | other, not inherited                                 | 0         |       | 1         | 2.8%  |
| 900.110             | other. suspect not inherited/significance unknown    | 4         | 6.1%  | 6         | 16.7% |
| <b>NORMAL</b>       |  |           |       |           |       |
| 0.000               | normal globe   | 22        | 33.3% | 10        | 27.8% |

## **OCULAR DISORDERS REPORT NEDERLANDSE KOOIKERHONDJE**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the NEDERLANDSE KOOIKERHONDJE breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT NEDERLANDSE KOOIKERHONDJE

| Diagnostic Name | TOTAL DOGS EXAMINED                                  | 1991-2015<br>78 |       | 2016-2020<br>142 |       |
|-----------------|--|-----------------|-------|------------------|-------|
|                 |  | #               | %     | #                | %     |
| <b>UVEA</b>     |  |                 |       |                  |       |
| 93.710          | persistent pupillary membranes, iris to iris         | 1               | 1.3%  | 3                | 2.1%  |
| 93.730          | persistent pupillary membranes, iris to cornea       | 1               | 1.3%  | 0                |       |
| <b>LENS</b>     |  |                 |       |                  |       |
| 100.210         | cataract. suspect not inherited/significance unknown | 6               | 7.7%  | 5                | 3.5%  |
| <b>VITREOUS</b> |  |                 |       |                  |       |
| 110.120         | persistent hyaloid artery/remnant                    | 1               | 1.3%  | 2                | 1.4%  |
| 110.320         | vitreal degeneration                                 | 2               | 2.6%  | 3                | 2.1%  |
| <b>RETINA</b>   |  |                 |       |                  |       |
| 120.960         | retinopathy  | 0               |       | 1                | 0.7%  |
| <b>OTHER</b>    |  |                 |       |                  |       |
| 900.000         | other, unspecified                                   | 2               | 2.6%  | 0                |       |
| 900.110         | other. suspect not inherited/significance unknown    | 5               | 6.4%  | 8                | 5.6%  |
| <b>NORMAL</b>   |  |                 |       |                  |       |
| 0.000           | normal globe   | 65              | 83.3% | 122              | 85.9% |

# **OCULAR DISORDERS REPORT NEW ZEALAND HUNTAWAY**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the NEW ZEALAND HUNTAWAY breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT NEW ZEALAND HUNTAWAY

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |        | 2016-2020 |   |
|---|---------------------|-----------|--------|-----------|---|
|   |                     | #         | %      | #         | % |
| <b>UVEA</b>   |                     |           |        |           |   |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 1         | 50.0%  | 0         |   |
| <b>NORMAL</b>   |                     |           |        |           |   |
| 0.000 normal globe  |                     | 2         | 100.0% | 0         |   |

# NEWFOUNDLAND

|    | <b>DISORDER</b>                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|----------------------------------|--------------------|------------------|------------------------|
| A. | Glaucoma                         | Not defined        | 2                | NO                     |
| B. | Entropion                        | Not defined        | 1                | Breeder option         |
| C. | Ectropion                        | Not defined        | 1                | Breeder option         |
| D. | Uveal cysts                      | Not defined        | 1                | Breeder option         |
| E. | Cataract                         | Not defined        | 1                | NO                     |
| F. | Retinal atrophy<br>- generalized | Not defined        | 3                | NO                     |

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## Description and Comments

### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure which, when sustained even for a brief period of time, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening examination.

Some Newfoundlands have an abnormality of the iridocorneal angle termed goniodysgenesis. This abnormality is not visible during routine ophthalmic examination using a slitlamp biomicroscope and an indirect ophthalmoscope. There appears to be an association between goniodysgenesis and glaucoma, but the mechanism by which the angle defect results in glaucoma has not been determined. The inheritance of goniodysgenesis in the Newfoundland is not known. Until the inheritance is determined, control should be directed towards removing dogs from breeding that have glaucoma and have goniodysgenesis, as well as those dogs that produce progeny afflicted with glaucoma.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### C. Ectropion

A conformational defect resulting in eversion of the eyelids, which may cause ocular irritation



due to exposure. It is likely that ectropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

D. Uveal cysts

A pigmented, fluid-filled epithelial-lined structure arising from the posterior iris or ciliary body epithelium. Cysts may remain attached to the pupil margin, iris, or ciliary body, or may detach and be free-floating within the anterior chamber. They may rupture and adhere to the cornea or anterior lens capsule. Uveal cysts may occur in any breed. Uveal cysts are commonly benign, although they may be associated with other pathologic conditions in various breeds.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

F. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as Progressive Retinal Atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Strom AR, Hassig M, Iburg TM, et al. Epidemiology of canine glaucoma presented to University of Zurich from 1995 to 2009. Part 1: Congenital and primary glaucoma (4 and 123 cases). *Vet Ophthalmol*. 2011 Mar;14:121-126.
3. Dekomien G and Epplen JT. Evaluation of the canine RPE65 gene in affected dogs with generalized progressive retinal atrophy. *Mol Vis*. 2003 Nov 11;9:601-605.

# OCULAR DISORDERS REPORT NEWFOUNDLAND

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>3,074 |      | 2016-2020<br>481 |      |
|---------------------|--|--------------------|------|------------------|------|
|                     |  | #                  | %    | #                | %    |
| <b>GLOBE</b>        |  |                    |      |                  |      |
| 0.110               | microphthalmia   | 5                  | 0.2% | 1                | 0.2% |
| 10.000              | glaucoma   | 1                  | 0.0% | 0                |      |
| <b>EYELIDS</b>      |  |                    |      |                  |      |
| 20.160              | macropalpebral fissure                                       | 128                | 4.2% | 0                |      |
| 21.000              | entropion, unspecified                                       | 204                | 6.6% | 33               | 6.9% |
| 22.000              | ectropion, unspecified                                       | 221                | 7.2% | 21               | 4.4% |
| 25.110              | distichiasis   | 21                 | 0.7% | 1                | 0.2% |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |      |
| 40.910              | keratoconjunctivitis sicca                                   | 1                  | 0.0% | 0                |      |
| <b>NICTITANS</b>    |  |                    |      |                  |      |
| 51.100              | third eyelid cartilage anomaly                               | 14                 | 0.5% | 3                | 0.6% |
| 52.110              | prolapsed gland of the third eyelid                          | 9                  | 0.3% | 1                | 0.2% |
| <b>CORNEA</b>       |  |                    |      |                  |      |
| 70.210              | corneal pannus   | 1                  | 0.0% | 0                |      |
| 70.220              | pigmentary keratitis   | 2                  | 0.1% | 0                |      |
| 70.700              | corneal dystrophy  | 1                  | 0.0% | 1                | 0.2% |
| 70.730              | corneal endothelial degeneration                             | 0                  |      | 1                | 0.2% |
| <b>UVEA</b>         |  |                    |      |                  |      |
| 93.140              | corneal endothelial pigment without PPM                      | 1                  | 0.0% | 0                |      |
| 93.710              | persistent pupillary membranes, iris to iris                 | 21                 | 0.7% | 2                | 0.4% |
| 93.720              | persistent pupillary membranes, iris to lens                 | 5                  | 0.2% | 0                |      |
| 93.730              | persistent pupillary membranes, iris to cornea               | 5                  | 0.2% | 1                | 0.2% |
| 93.740              | persistent pupillary membranes, iris sheets                  | 1                  | 0.0% | 0                |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands | 2                  | 0.1% | 0                |      |
| 93.810              | uveal melanoma   | 1                  | 0.0% | 0                |      |
| 93.999              | uveal cysts  | 52                 | 1.7% | 9                | 1.9% |
| <b>LENS</b>         |  |                    |      |                  |      |
| 100.200             | cataract, unspecified  | 11                 | 0.4% | 0                |      |
| 100.210             | cataract. suspect not inherited/significance unknown         | 101                | 3.3% | 13               | 2.7% |
| 100.301             | punctate cataract, anterior cortex                           | 6                  | 0.2% | 2                | 0.4% |
| 100.302             | punctate cataract, posterior cortex                          | 13                 | 0.4% | 1                | 0.2% |
| 100.303             | punctate cataract, equatorial cortex                         | 5                  | 0.2% | 3                | 0.6% |
| 100.305             | punctate cataract, posterior sutures                         | 6                  | 0.2% | 1                | 0.2% |
| 100.306             | punctate cataract, nucleus                                   | 3                  | 0.1% | 0                |      |
| 100.307             | punctate cataract, capsular                                  | 3                  | 0.1% | 3                | 0.6% |
| 100.311             | incipient cataract, anterior cortex                          | 17                 | 0.6% | 1                | 0.2% |
| 100.312             | incipient cataract, posterior cortex                         | 86                 | 2.8% | 14               | 2.9% |
| 100.313             | incipient cataract, equatorial cortex                        | 18                 | 0.6% | 4                | 0.8% |
| 100.314             | incipient cataract, anterior sutures                         | 3                  | 0.1% | 0                |      |
| 100.315             | incipient cataract, posterior sutures                        | 12                 | 0.4% | 2                | 0.4% |
| 100.316             | incipient cataract, nucleus                                  | 12                 | 0.4% | 1                | 0.2% |
| 100.317             | incipient cataract, capsular                                 | 8                  | 0.3% | 1                | 0.2% |
| 100.321             | incomplete cataract, anterior cortex                         | 0                  |      | 2                | 0.4% |
| 100.322             | incomplete cataract, posterior cortex                        | 5                  | 0.2% | 2                | 0.4% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.323 incomplete cataract, equatorial cortex            | 1 0.0%           | 0                |
| 100.325 incomplete cataract, posterior sutures            | 0                | 1 0.2%           |
| 100.326 incomplete cataract, nucleus                      | 0                | 1 0.2%           |
| 100.328 y-suture tip opacities                            | 1 0.0%           | 2 0.4%           |
| 100.330 generalized/complete cataract                     | 38 1.2%          | 0                |
| 100.345 <i>significant cataracts (summary)</i>            | 248 8.1%         | 41 8.5%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 1 0.0%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 4 0.1%           | 1 0.2%           |
| 110.135 PHPV/PTVL   | 4 0.1%           | 0                |
| 110.320 vitreal degeneration                              | 5 0.2%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 26 0.8%          | 3 0.6%           |
| 120.180 retinal dysplasia, geographic                     | 2 0.1%           | 0                |
| 120.190 retinal dysplasia, detached                       | 1 0.0%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 1 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| 120.960 retinopathy                                       | 1 0.0%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.120 optic nerve hypoplasia                            | 7 0.2%           | 0                |
| 130.150 optic disc coloboma                               | 1 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 29 0.9%          | 0                |
| 900.100 other, not inherited                              | 72 2.3%          | 3 0.6%           |
| 900.110 other. suspect not inherited/significance unknown | 44 1.4%          | 15 3.1%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 2,327 75.7%      | 359 74.6%        |

# NORFOLK TERRIER

|    | DISORDER                                  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE         | GENETIC TESTS<br>AVAILABLE              |
|----|---|------------------------|-----------|----------------------------|---|
| A. | Corneal dystrophy<br>- epithelial/stromal | Not defined            | 1         | Breeder option             |   |
| B. | Persistent pupillary<br>membranes         |                        |           |                            |   |
|    | - iris to iris                            | Not defined            | 1         | Breeder option             |   |
|    | - lens pigment foci/no<br>strands         | Not defined            | 1         | Passes with no<br>notation |   |
|    | - endothelial<br>opacity/no strands       | Not defined            | 1         | NO                         |   |
| C. | Cataract                                  | Not defined            | 1         | NO                         |   |
| D. | Lens luxation                             | Autosomal<br>recessive | 2, 3      | NO                         | Mutation in the<br><i>ADAMTS17</i> gene |
| E. | Optic nerve<br>hypoplasia                 | Not defined            | 1         | NO                         |   |

## Description and Comments

### A. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely

(diffuse) or in a localized region.

D. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

E. Optic nerve hypoplasia

A congenital anomaly, which results in a small optic disk diameter and vision loss.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011; 14: 378-384.
3. Komaromy A. Genetics of canine primary glaucomas. *Vet Clin Small Anim.* 2015; 45: 1159-1182.

# OCULAR DISORDERS REPORT NORFOLK TERRIER

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|------------------|--|-----------|-------|-----------|-------|
|                  |  | #         | %     | #         | %     |
| <b>EYELIDS</b>   |  |           |       |           |       |
| 20.160           | macropalpebral fissure   | 1         | 0.1%  | 0         |       |
| 25.110           | distichiasis   | 6         | 0.5%  | 0         |       |
| <b>NICTITANS</b> |  |           |       |           |       |
| 52.110           | prolapsed gland of the third eyelid                            | 2         | 0.2%  | 0         |       |
| <b>CORNEA</b>    |  |           |       |           |       |
| 70.700           | corneal dystrophy  | 12        | 0.9%  | 7         | 1.8%  |
| 70.730           | corneal endothelial degeneration                               | 1         | 0.1%  | 2         | 0.5%  |
| <b>UVEA</b>      |  |           |       |           |       |
| 93.140           | corneal endothelial pigment without PPM                        | 1         | 0.1%  | 0         |       |
| 93.710           | persistent pupillary membranes, iris to iris                   | 269       | 20.5% | 99        | 25.8% |
| 93.720           | persistent pupillary membranes, iris to lens                   | 1         | 0.1%  | 1         | 0.3%  |
| 93.730           | persistent pupillary membranes, iris to cornea                 | 4         | 0.3%  | 2         | 0.5%  |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands   | 8         | 0.6%  | 7         | 1.8%  |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands | 2         | 0.2%  | 4         | 1.0%  |
| <b>LENS</b>      |  |           |       |           |       |
| 100.200          | cataract, unspecified  | 1         | 0.1%  | 0         |       |
| 100.210          | cataract. suspect not inherited/significance unknown           | 44        | 3.4%  | 2         | 0.5%  |
| 100.301          | punctate cataract, anterior cortex                             | 5         | 0.4%  | 1         | 0.3%  |
| 100.302          | punctate cataract, posterior cortex                            | 4         | 0.3%  | 0         |       |
| 100.305          | punctate cataract, posterior sutures                           | 8         | 0.6%  | 0         |       |
| 100.306          | punctate cataract, nucleus                                     | 1         | 0.1%  | 0         |       |
| 100.307          | punctate cataract, capsular                                    | 2         | 0.2%  | 0         |       |
| 100.311          | incipient cataract, anterior cortex                            | 7         | 0.5%  | 5         | 1.3%  |
| 100.312          | incipient cataract, posterior cortex                           | 16        | 1.2%  | 3         | 0.8%  |
| 100.313          | incipient cataract, equatorial cortex                          | 5         | 0.4%  | 0         |       |
| 100.315          | incipient cataract, posterior sutures                          | 2         | 0.2%  | 1         | 0.3%  |
| 100.317          | incipient cataract, capsular                                   | 4         | 0.3%  | 1         | 0.3%  |
| 100.321          | incomplete cataract, anterior cortex                           | 0         |       | 1         | 0.3%  |
| 100.322          | incomplete cataract, posterior cortex                          | 2         | 0.2%  | 0         |       |
| 100.328          | y-suture tip opacities   | 0         |       | 1         | 0.3%  |
| 100.330          | generalized/complete cataract                                  | 4         | 0.3%  | 0         |       |
| 100.345          | <i>significant cataracts (summary)</i>                         | 61        | 4.6%  | 13        | 3.4%  |
| <b>VITREOUS</b>  |  |           |       |           |       |
| 110.120          | persistent hyaloid artery/remnant                              | 6         | 0.5%  | 2         | 0.5%  |
| 110.135          | PHPV/PTVL  | 1         | 0.1%  | 0         |       |
| 110.320          | vitreal degeneration   | 8         | 0.6%  | 0         |       |
| <b>FUNDUS</b>    |  |           |       |           |       |
| 97.120           | coloboma   | 1         | 0.1%  | 0         |       |
| <b>RETINA</b>    |  |           |       |           |       |
| 120.170          | retinal dysplasia, folds                                       | 7         | 0.5%  | 0         |       |
| 120.180          | retinal dysplasia, geographic                                  | 2         | 0.2%  | 0         |       |
| 120.310          | generalized progressive retinal atrophy (PRA)                  | 10        | 0.8%  | 1         | 0.3%  |

| <b>RETINA CONTINUED</b>                                   | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 120.910 retinal detachment without dialysis               | 1 0.1%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 10 0.8%          | 4 1.0%           |
| 130.120 optic nerve hypoplasia                            | 18 1.4%          | 8 2.1%           |
| 130.150 optic disc coloboma                               | 19 1.4%          | 1 0.3%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 14 1.1%          | 0                |
| 900.100 other, not inherited                              | 38 2.9%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 22 1.7%          | 14 3.7%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 947 72.1%        | 240 62.7%        |

# NORBOTTENSPETS

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|------------------------------------|------------------------|-----------|--------------------|-------------------------------------|
| A. | Cataract                           | Not defined            | 1         | NO                 |                                     |
| B. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 1         | NO                 | Mutation of the<br><i>prcd</i> gene |

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## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### B. Retinal atrophy – *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Norbottenspets is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Norbottenspets. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.



# OCULAR DISORDERS REPORT NORRBOTTENSPETS

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015<br>99 |       | 2016-2020<br>22 |       |
|-----------------|--|-----------------|-------|-----------------|-------|
|                 |  | #               | %     | #               | %     |
| <b>EYELIDS</b>  |  |                 |       |                 |       |
| 25.110          | distichiasis   | 1               | 1.0%  | 1               | 4.5%  |
| <b>CORNEA</b>   |  |                 |       |                 |       |
| 70.700          | corneal dystrophy  | 1               | 1.0%  | 0               |       |
| <b>UVEA</b>     |  |                 |       |                 |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 6               | 6.1%  | 2               | 9.1%  |
| 93.720          | persistent pupillary membranes, iris to lens                 | 1               | 1.0%  | 0               |       |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 0               |       | 2               | 9.1%  |
| <b>LENS</b>     |  |                 |       |                 |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 5               | 5.1%  | 2               | 9.1%  |
| 100.302         | punctate cataract, posterior cortex                          | 2               | 2.0%  | 0               |       |
| 100.305         | punctate cataract, posterior sutures                         | 1               | 1.0%  | 0               |       |
| 100.306         | punctate cataract, nucleus                                   | 1               | 1.0%  | 0               |       |
| 100.311         | incipient cataract, anterior cortex                          | 7               | 7.1%  | 0               |       |
| 100.312         | incipient cataract, posterior cortex                         | 9               | 9.1%  | 0               |       |
| 100.315         | incipient cataract, posterior sutures                        | 1               | 1.0%  | 0               |       |
| 100.316         | incipient cataract, nucleus                                  | 3               | 3.0%  | 0               |       |
| 100.330         | generalized/complete cataract                                | 1               | 1.0%  | 0               |       |
| 100.345         | <i>significant cataracts (summary)</i>                       | 25              | 25.3% | 0               |       |
| <b>RETINA</b>   |  |                 |       |                 |       |
| 120.170         | retinal dysplasia, folds                                     | 1               | 1.0%  | 1               | 4.5%  |
| 120.310         | generalized progressive retinal atrophy (PRA)                | 2               | 2.0%  | 0               |       |
| <b>OTHER</b>    |  |                 |       |                 |       |
| 900.100         | other, not inherited   | 3               | 3.0%  | 0               |       |
| 900.110         | other. suspect not inherited/significance unknown            | 0               |       | 1               | 4.5%  |
| <b>NORMAL</b>   |  |                 |       |                 |       |
| 0.000           | normal globe   | 75              | 75.8% | 17              | 77.3% |

## **OCULAR DISORDERS REPORT NORTH AMERICAN SHEPHERD**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the NORTH AMERICAN SHEPHERD breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT NORTH AMERICAN SHEPHERD

| Diagnostic Name                                | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |   |
|--|---------------------|-----------|-------|-----------|---|
|  |                     | #         | %     | #         | % |
| <b>VITREOUS</b>                                |                     |           |       |           |   |
| 110.200 vitreous degeneration-anterior chamber |                     | 1         | 16.7% | 0         |   |
| <b>NORMAL</b>                                  |                     |           |       |           |   |
| 0.000 normal globe                             |                     | 5         | 83.3% | 0         |   |

# NORTHERN INUIT

|    | DISORDER  | INHERITANCE         | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE         |
|----|---|---------------------|-----------|--------------------|------------------------------------|
| A. | Retinal dysplasia - folds/geographic/detached (with skeletal defects) | Autosomal recessive | 1         | NO                 | Mutation in the <i>COL9A3</i> gene |

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## Description and Comments

- A. Retinal dysplasia - folds or detachment with skeletal defects

This condition is also known as oculo-skeletal dysplasia (OSD) or dwarfism with retinal dysplasia type 1 (DRD1) also occurs in the Labrador Retriever. A similar condition, DRD2, occurs in the Samoyed. The condition is autosomal recessive and homozygous affected dogs have shortened forelimbs ("downhill" conformation) with valgus deformity. They have severe ocular defects including cataract, retinal folds/multifocal retinal dysplasia, vitreal degeneration and retinal detachment. The ocular abnormalities result in blindness in most dogs. Heterozygous dogs can have either a normal ocular exam or have multiple retinal folds, vitreal membranes, or vitreal degeneration suggesting a semi-dominant mechanism with respect to the eyes. It is important to note that generally the retinal folds present in heterozygous dogs tend to cluster around the major superior blood vessels of the central tapetal region. The condition is caused by a 1 base pair insertion of *COL9A3*. A DNA test is available.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Northern Inuit. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT NORTHERN INUIT

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015 |        | 2016-2020 |       |
|--|---------------------|-----------|--------|-----------|-------|
|  |                     | #         | %      | #         | %     |
| <b>LENS</b>  |                     |           |        |           |       |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 0         |        | 1         | 8.3%  |
| <b>NORMAL</b>  |                     |           |        |           |       |
| 0.000 normal globe   |                     | 1         | 100.0% | 11        | 91.7% |

# NORWEGIAN BUHUND

|    | DISORDER                  | INHERITANCE                       | REFERENCE | BREEDING ADVICE |
|----|---------------------------|-----------------------------------|-----------|-----------------|
| A. | Cataract                  | Not defined                       | 1, 3      | NO              |
| B. | Cataract<br>- pulverulent | Presumed<br>autosomal<br>dominant | 2, 3      | Breeder option  |
| C. | Y-suture top opacity      | Not defined                       | 1         | Breeder option  |

## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

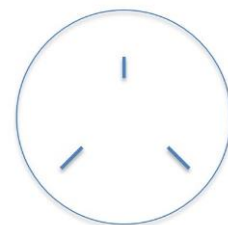
### B. Cataract - pulverulent

With the pulverulent cataract in the Norwegian Buhund, initial lens changes may be visible as early as 6.5 weeks of age as small dots parallel to the suture lines behind the nucleus. By the age of 4 to 5.5 years, the opacities progress to involve the fetal nucleus which then resembles a ball of candy floss. The adult nucleus and the cortex remain clear. An autosomal dominant mode of inheritance with a high degree of penetrance has been suggested.

Rates of progression of these cataracts can vary, and have been noted to develop in older animals (over the age of 7) that were previously documented to be free from this condition.

### C. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are



considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.

These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Bjerkas E and Haaland MB. Pulverulent nuclear cataract in the Norwegian Buhund. *J Small Anim Pract.* 1995;36:471-474.
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# OCULAR DISORDERS REPORT NORWEGIAN BUHUND

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>637 |       | 2016-2020<br>280 |       |
|---|---------------------|------------------|-------|------------------|-------|
|   |                     | #                | %     | #                | %     |
| <b>GLOBE</b>  |                     |                  |       |                  |       |
| 10.000 glaucoma   |                     | 0                |       | 1                | 0.4%  |
| <b>EYELIDS</b>  |                     |                  |       |                  |       |
| 25.110 distichiasis   |                     | 2                | 0.3%  | 0                |       |
| <b>CORNEA</b>   |                     |                  |       |                  |       |
| 70.220 pigmentary keratitis   |                     | 0                |       | 1                | 0.4%  |
| 70.700 corneal dystrophy  |                     | 5                | 0.8%  | 2                | 0.7%  |
| <b>UVEA</b>   |                     |                  |       |                  |       |
| 93.110 iris hypoplasia  |                     | 0                |       | 1                | 0.4%  |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 2                | 0.3%  | 0                |       |
| 93.740 persistent pupillary membranes, iris sheets                  |                     | 1                | 0.2%  | 0                |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 0                |       | 1                | 0.4%  |
| 93.999 uveal cysts  |                     | 0                |       | 1                | 0.4%  |
| <b>LENS</b>   |                     |                  |       |                  |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 76               | 11.9% | 33               | 11.8% |
| 100.301 punctate cataract, anterior cortex                          |                     | 6                | 0.9%  | 0                |       |
| 100.302 punctate cataract, posterior cortex                         |                     | 7                | 1.1%  | 4                | 1.4%  |
| 100.303 punctate cataract, equatorial cortex                        |                     | 1                | 0.2%  | 0                |       |
| 100.305 punctate cataract, posterior sutures                        |                     | 4                | 0.6%  | 7                | 2.5%  |
| 100.306 punctate cataract, nucleus                                  |                     | 9                | 1.4%  | 4                | 1.4%  |
| 100.307 punctate cataract, capsular                                 |                     | 1                | 0.2%  | 1                | 0.4%  |
| 100.311 incipient cataract, anterior cortex                         |                     | 3                | 0.5%  | 3                | 1.1%  |
| 100.312 incipient cataract, posterior cortex                        |                     | 17               | 2.7%  | 7                | 2.5%  |
| 100.313 incipient cataract, equatorial cortex                       |                     | 0                |       | 2                | 0.7%  |
| 100.315 incipient cataract, posterior sutures                       |                     | 10               | 1.6%  | 3                | 1.1%  |
| 100.316 incipient cataract, nucleus                                 |                     | 14               | 2.2%  | 4                | 1.4%  |
| 100.321 incomplete cataract, anterior cortex                        |                     | 1                | 0.2%  | 0                |       |
| 100.322 incomplete cataract, posterior cortex                       |                     | 1                | 0.2%  | 0                |       |
| 100.323 incomplete cataract, equatorial cortex                      |                     | 0                |       | 1                | 0.4%  |
| 100.325 incomplete cataract, posterior sutures                      |                     | 1                | 0.2%  | 0                |       |
| 100.328 y-suture tip opacities                                      |                     | 0                |       | 6                | 2.1%  |
| 100.330 generalized/complete cataract                               |                     | 6                | 0.9%  | 0                |       |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 81               | 12.7% | 42               | 15.0% |
| <b>VITREOUS</b>   |                     |                  |       |                  |       |
| 110.320 vitreal degeneration  |                     | 0                |       | 1                | 0.4%  |
| <b>RETINA</b>   |                     |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                                    |                     | 8                | 1.3%  | 3                | 1.1%  |
| 120.310 generalized progressive retinal atrophy (PRA)               |                     | 3                | 0.5%  | 0                |       |
| 120.960 retinopathy   |                     | 3                | 0.5%  | 5                | 1.8%  |
| <b>OTHER</b>  |                     |                  |       |                  |       |
| 900.000 other, unspecified  |                     | 14               | 2.2%  | 0                |       |
| 900.100 other, not inherited  |                     | 17               | 2.7%  | 6                | 2.1%  |
| 900.110 other. suspect not inherited/significance unknown           |                     | 13               | 2.0%  | 22               | 7.9%  |



|                                     | 1991-2015 | 2016-2020 |
|-------------------------------------|-----------|-----------|
| <b>NORMAL</b><br>0.000 normal globe | 484 76.0% | 181 64.6% |

# NORWEGIAN ELKHOUND

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE            |
|----|---|------------------------|-----------|--------------------|---------------------------------------|
| A. | Glaucoma  | Not defined            | 1-6       | NO                 |                                       |
| B. | Distichiasis  | Not defined            | 4         | Breeder option     |                                       |
| C. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined            | 1         | Breeder option     |                                       |
| D. | Cataract  | Not defined            | 1         | NO                 |                                       |
| E. | Retinal atrophy<br>( <i>prcd</i> )                  | Autosomal<br>recessive | 7         | NO                 | Mutation of the<br><i>prcd</i> gene   |
| F. | Retinal atrophy<br>- generalized                    |                        |           |                    |                                       |
|    | 1. Rod dysplasia ( <i>rd</i> )                      | Autosomal<br>recessive | 8-11      | NO                 |                                       |
|    | 2. Early retinal<br>degeneration ( <i>erd</i> )     | Autosomal<br>recessive | 12-16     | NO                 | Mutation of the<br><i>STK38L</i> gene |
| G. | Retinal dysplasia<br>- folds                        | Not defined            | 1         | Breeder option     |                                       |

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## Description and Comments

### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of the IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine screening exam for certification.

In the Norwegian Elkhound, glaucoma appears to be familial. In most cases the drainage angle is reported to be open. A mutation has been found in *ADAMTS10* in some Norwegian Elkhounds with glaucoma, but a genetic test is not yet available.

B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that PRA in the Norwegian Elkhound is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

F. Retinal atrophy - generalized

1. **Rod dysplasia (*rd*):** Inappropriate development of the visual cells resulting in vision impairment in dim light by 6 months and total blindness at 3-5 years. Ophthalmoscopic signs may be evident after 5 months of age, with signs of retinal vascular thinning after 2 years. An ERG can provide a diagnosis as early as 6 weeks of age. In the Norwegian Elkhound, this is an autosomal recessive trait.

2. **Early retinal degeneration (*erd*):** Another form of PRA reported in the Norwegian Elkhound. Animals are night blind at 6 weeks and blind by 1 year of age. Clinical signs are

evident by 6 months. On histopathologic examination there is an abnormal structural development of the photoreceptors followed by rapid rod/cone degeneration. The mutation is found in the *STK38L* gene and is inherited as an autosomal recessive trait. While a DNA test is available, no Norwegian Elkhounds are thought to exist with this mutation anymore.

G. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

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16. Goldstein O, Kukekova AV, Aguirre GD, et al. Exonic SINE insertion in STK38L causes canine early retinal degeneration (erd). *Genomics.* 2010;96:362-368.

# OCULAR DISORDERS REPORT NORWEGIAN ELKHOUND

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>2,540 |      | 2016-2020<br>248 |      |
|---------------------|--|--------------------|------|------------------|------|
|                     |  | #                  | %    | #                | %    |
| <b>GLOBE</b>        |  |                    |      |                  |      |
| 0.110               | microphthalmia   | 4                  | 0.2% | 0                |      |
| 10.000              | glaucoma   | 2                  | 0.1% | 0                |      |
| <b>EYELIDS</b>      |  |                    |      |                  |      |
| 20.160              | macropalpebral fissure                                       | 16                 | 0.6% | 0                |      |
| 21.000              | entropion, unspecified                                       | 5                  | 0.2% | 0                |      |
| 22.000              | ectropion, unspecified                                       | 14                 | 0.6% | 0                |      |
| 25.110              | distichiasis   | 44                 | 1.7% | 2                | 0.8% |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |      |
| 32.110              | imperforate lower nasolacrimal punctum                       | 1                  | 0.0% | 0                |      |
| <b>NICTITANS</b>    |  |                    |      |                  |      |
| 51.100              | third eyelid cartilage anomaly                               | 1                  | 0.0% | 0                |      |
| 52.110              | prolapsed gland of the third eyelid                          | 1                  | 0.0% | 0                |      |
| <b>CORNEA</b>       |  |                    |      |                  |      |
| 70.210              | corneal pannus   | 2                  | 0.1% | 0                |      |
| 70.700              | corneal dystrophy  | 9                  | 0.4% | 2                | 0.8% |
| <b>UVEA</b>         |  |                    |      |                  |      |
| 93.710              | persistent pupillary membranes, iris to iris                 | 34                 | 1.3% | 3                | 1.2% |
| 93.720              | persistent pupillary membranes, iris to lens                 | 10                 | 0.4% | 1                | 0.4% |
| 93.730              | persistent pupillary membranes, iris to cornea               | 6                  | 0.2% | 1                | 0.4% |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands | 3                  | 0.1% | 4                | 1.6% |
| 93.999              | uveal cysts  | 7                  | 0.3% | 0                |      |
| <b>LENS</b>         |  |                    |      |                  |      |
| 100.200             | cataract, unspecified  | 23                 | 0.9% | 0                |      |
| 100.210             | cataract. suspect not inherited/significance unknown         | 107                | 4.2% | 17               | 6.9% |
| 100.301             | punctate cataract, anterior cortex                           | 8                  | 0.3% | 1                | 0.4% |
| 100.302             | punctate cataract, posterior cortex                          | 8                  | 0.3% | 2                | 0.8% |
| 100.303             | punctate cataract, equatorial cortex                         | 4                  | 0.2% | 0                |      |
| 100.304             | punctate cataract, anterior sutures                          | 1                  | 0.0% | 0                |      |
| 100.305             | punctate cataract, posterior sutures                         | 10                 | 0.4% | 1                | 0.4% |
| 100.306             | punctate cataract, nucleus                                   | 3                  | 0.1% | 1                | 0.4% |
| 100.307             | punctate cataract, capsular                                  | 2                  | 0.1% | 1                | 0.4% |
| 100.311             | incipient cataract, anterior cortex                          | 11                 | 0.4% | 1                | 0.4% |
| 100.312             | incipient cataract, posterior cortex                         | 37                 | 1.5% | 2                | 0.8% |
| 100.313             | incipient cataract, equatorial cortex                        | 21                 | 0.8% | 4                | 1.6% |
| 100.314             | incipient cataract, anterior sutures                         | 3                  | 0.1% | 0                |      |
| 100.315             | incipient cataract, posterior sutures                        | 7                  | 0.3% | 1                | 0.4% |
| 100.316             | incipient cataract, nucleus                                  | 8                  | 0.3% | 1                | 0.4% |
| 100.317             | incipient cataract, capsular                                 | 9                  | 0.4% | 0                |      |
| 100.321             | incomplete cataract, anterior cortex                         | 1                  | 0.0% | 1                | 0.4% |
| 100.323             | incomplete cataract, equatorial cortex                       | 0                  |      | 1                | 0.4% |
| 100.326             | incomplete cataract, nucleus                                 | 0                  |      | 1                | 0.4% |
| 100.327             | incomplete cataract, capsular                                | 0                  |      | 1                | 0.4% |
| 100.328             | y-suture tip opacities                                       | 1                  | 0.0% | 3                | 1.2% |
| 100.330             | generalized/complete cataract                                | 7                  | 0.3% | 0                |      |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.345 significant cataracts (summary)                   | 164 6.5%         | 22 8.9%          |
| 100.375 subluxation/luxation, unspecified                 | 4 0.2%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 7 0.3%           | 0                |
| 110.135 PHPV/PTVL   | 2 0.1%           | 0                |
| 110.320 vitreal degeneration                              | 7 0.3%           | 1 0.4%           |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 44 1.7%          | 6 2.4%           |
| 120.180 retinal dysplasia, geographic                     | 2 0.1%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 10 0.4%          | 0                |
| 120.400 retinal hemorrhage                                | 3 0.1%           | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 0                | 1 0.4%           |
| 130.120 optic nerve hypoplasia                            | 2 0.1%           | 1 0.4%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 22 0.9%          | 0                |
| 900.100 other, not inherited                              | 32 1.3%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 16 0.6%          | 23 9.3%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 2,186 86.1%      | 180 72.6%        |

# **OCULAR DISORDERS REPORT NORWEGIAN LUNDEHUND**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the NORWEGIAN LUNDEHUND breed. Therefore, there are no conditions listed with breeding advice.



# OCULAR DISORDERS REPORT NORWEGIAN LUNDEHUND

| Diagnostic Name | TOTAL DOGS EXAMINED                                  | 1991-2015<br>48 |       | 2016-2020<br>2 |        |
|-----------------|--|-----------------|-------|----------------|--------|
|                 |  | #               | %     | #              | %      |
| <b>UVEA</b>     |  |                 |       |                |        |
| 93.710          | persistent pupillary membranes, iris to iris         | 13              | 27.1% | 0              |        |
| 93.720          | persistent pupillary membranes, iris to lens         | 1               | 2.1%  | 0              |        |
| <b>LENS</b>     |  |                 |       |                |        |
| 100.210         | cataract. suspect not inherited/significance unknown | 8               | 16.7% | 0              |        |
| 100.301         | punctate cataract, anterior cortex                   | 1               | 2.1%  | 0              |        |
| 100.302         | punctate cataract, posterior cortex                  | 2               | 4.2%  | 0              |        |
| 100.311         | incipient cataract, anterior cortex                  | 2               | 4.2%  | 0              |        |
| 100.313         | incipient cataract, equatorial cortex                | 1               | 2.1%  | 0              |        |
| 100.315         | incipient cataract, posterior sutures                | 2               | 4.2%  | 0              |        |
| 100.330         | generalized/complete cataract                        | 3               | 6.2%  | 0              |        |
| 100.345         | <i>significant cataracts (summary)</i>               | 11              | 22.9% | 0              |        |
| <b>VITREOUS</b> |  |                 |       |                |        |
| 110.320         | vitreal degeneration                                 | 2               | 4.2%  | 0              |        |
| <b>OTHER</b>    |  |                 |       |                |        |
| 900.000         | other, unspecified                                   | 1               | 2.1%  | 0              |        |
| <b>NORMAL</b>   |  |                 |       |                |        |
| 0.000           | normal globe   | 29              | 60.4% | 2              | 100.0% |

# NORWICH TERRIER

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|--|---------------------|-----------|-----------------|--------------------------------------|
| A. | Distichiasis                                     | Not defined         | 1         | Breeder option  |                                      |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option  |                                      |
| C. | Cataract   | Not defined         | 2         | NO              |                                      |
| D. | Lens luxation                                    | Autosomal recessive | 2, 3      | NO              | Mutation in the <i>ADAMTS17</i> gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### D. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to

be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Farias FH, Johnson GS, Taylor JF, et al. An *ADAMTS17* splice donor site mutation in dogs with primary lens luxation. *Invest Ophthalmol Vis Sci.* 2010;51:4716-4721.
3. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011;14:378-384.

# OCULAR DISORDERS REPORT NORWICH TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>3,080 |      | 2016-2020<br>611 |      |
|---------------------|--|--------------------|------|------------------|------|
|                     |  | #                  | %    | #                | %    |
| <b>EYELIDS</b>      |  |                    |      |                  |      |
| 20.160              | macropalpebral fissure   | 1                  | 0.0% | 0                |      |
| 22.000              | ectropion, unspecified   | 1                  | 0.0% | 0                |      |
| 25.110              | distichiasis   | 18                 | 0.6% | 12               | 2.0% |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 1                  | 0.0% | 1                | 0.2% |
| <b>NICTITANS</b>    |  |                    |      |                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 4                  | 0.1% | 0                |      |
| <b>CORNEA</b>       |  |                    |      |                  |      |
| 70.700              | corneal dystrophy  | 18                 | 0.6% | 5                | 0.8% |
| 70.730              | corneal endothelial degeneration                               | 4                  | 0.1% | 0                |      |
| <b>UVEA</b>         |  |                    |      |                  |      |
| 93.150              | iris coloboma  | 1                  | 0.0% | 0                |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 177                | 5.7% | 20               | 3.3% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 4                  | 0.1% | 0                |      |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 8                  | 0.3% | 0                |      |
| 93.740              | persistent pupillary membranes, iris sheets                    | 1                  | 0.0% | 0                |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 4                  | 0.1% | 3                | 0.5% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 5                  | 0.2% | 0                |      |
| 93.999              | uveal cysts  | 1                  | 0.0% | 0                |      |
| <b>LENS</b>         |  |                    |      |                  |      |
| 100.200             | cataract, unspecified  | 5                  | 0.2% | 0                |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 73                 | 2.4% | 11               | 1.8% |
| 100.301             | punctate cataract, anterior cortex                             | 10                 | 0.3% | 0                |      |
| 100.302             | punctate cataract, posterior cortex                            | 9                  | 0.3% | 2                | 0.3% |
| 100.303             | punctate cataract, equatorial cortex                           | 2                  | 0.1% | 1                | 0.2% |
| 100.305             | punctate cataract, posterior sutures                           | 6                  | 0.2% | 1                | 0.2% |
| 100.306             | punctate cataract, nucleus                                     | 3                  | 0.1% | 0                |      |
| 100.307             | punctate cataract, capsular                                    | 1                  | 0.0% | 2                | 0.3% |
| 100.311             | incipient cataract, anterior cortex                            | 15                 | 0.5% | 4                | 0.7% |
| 100.312             | incipient cataract, posterior cortex                           | 17                 | 0.6% | 2                | 0.3% |
| 100.313             | incipient cataract, equatorial cortex                          | 13                 | 0.4% | 2                | 0.3% |
| 100.314             | incipient cataract, anterior sutures                           | 1                  | 0.0% | 0                |      |
| 100.315             | incipient cataract, posterior sutures                          | 6                  | 0.2% | 0                |      |
| 100.316             | incipient cataract, nucleus                                    | 11                 | 0.4% | 4                | 0.7% |
| 100.317             | incipient cataract, capsular                                   | 1                  | 0.0% | 3                | 0.5% |
| 100.321             | incomplete cataract, anterior cortex                           | 1                  | 0.0% | 0                |      |
| 100.322             | incomplete cataract, posterior cortex                          | 1                  | 0.0% | 1                | 0.2% |
| 100.323             | incomplete cataract, equatorial cortex                         | 0                  |      | 1                | 0.2% |
| 100.328             | y-suture tip opacities   | 0                  |      | 2                | 0.3% |
| 100.330             | generalized/complete cataract                                  | 12                 | 0.4% | 0                |      |
| 100.345             | <i>significant cataracts (summary)</i>                         | 114                | 3.7% | 25               | 4.1% |
| 100.375             | <i>subluxation/luxation, unspecified</i>                       | 1                  | 0.0% | 0                |      |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>VITREOUS</b>   |             |           |
| 110.120 persistent hyaloid artery/remnant                 | 3 0.1%      | 0         |
| 110.135 PHPV/PTVL   | 1 0.0%      | 0         |
| 110.320 vitreal degeneration                              | 11 0.4%     | 0         |
| <b>FUNDUS</b>   |             |           |
| 97.120 coloboma   | 2 0.1%      | 0         |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 6 0.2%      | 0         |
| 120.180 retinal dysplasia, geographic                     | 4 0.1%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 14 0.5%     | 0         |
| 120.960 retinopathy                                       | 4 0.1%      | 3 0.5%    |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 1 0.0%      | 0         |
| 130.120 optic nerve hypoplasia                            | 8 0.3%      | 0         |
| 130.150 optic disc coloboma                               | 3 0.1%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 28 0.9%     | 0         |
| 900.100 other, not inherited                              | 51 1.7%     | 1 0.2%    |
| 900.110 other. suspect not inherited/significance unknown | 24 0.8%     | 11 1.8%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 2,745 89.1% | 530 86.7% |

# NOVA SCOTIA DUCK TOLLING RETRIEVER

|    | DISORDER                                     | INHERITANCE         | REFERENCE | BREEDING ADVICE         | GENETIC TESTS AVAILABLE           |
|----|--|---------------------|-----------|-------------------------|-----------------------------------|
| A. | Distichiasis                                 | Not defined         | 1         | Breeder option          |                                   |
| B. | Corneal dystrophy<br>- epithelial/stromal    | Not defined         | 1         | Breeder option          |                                   |
| C. | Persistent pupillary membranes               |                     |           |                         |                                   |
|    | - iris to iris                               | Not defined         | 1         | Breeder option          |                                   |
|    | - iris to lens                               | Not defined         | 1         | NO                      |                                   |
|    | - lens pigment foci/no strands               | Not defined         | 1         | Passes with no notation |                                   |
| D. | Cataract                                     | Not defined         | 1         | NO                      |                                   |
| E. | Y-suture tip opacity                         | Not defined         | 1         | Breeder option          |                                   |
| F. | Retinal atrophy<br>( <i>prcd</i> )           | Autosomal recessive | 1, 2      | NO                      | Mutation of the <i>prcd</i> gene  |
| G. | Choroidal hypoplasia<br>(Collie eye anomaly) | Autosomal recessive | 1, 3, 4   | NO                      | Mutation of the <i>NHEJ1</i> gene |
|    | - staphyloma/coloboma                        |                     |           |                         |                                   |
|    | - retinal detachment                         |                     |           |                         |                                   |
|    | - retinal hemorrhage                         |                     |           |                         |                                   |
|    | - optic nerve coloboma                       |                     |           |                         |                                   |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

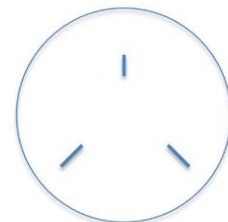
In the Nova Scotia Duck Tolling Retriever, many of the PPMs identified on routine screening examinations bridge from the iris to the lens where they are associated with focal cataract. This may result in vision impairment.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

F. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Nova Scotia Duck Tolling Retriever is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

G. Choroidal hypoplasia (Collie eye anomaly)

- staphyloma/coloboma
- retinal detachment
- retinal hemorrhage
- optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie eye anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006 Nov;88:551-563. PMID: 16938425
3. Parker HG, Kukekova AV, Akey DT, et al. Breed relationships facilitate fine-mapping studies: a 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. *Genome Res*. 2007 Nov;17:1562-1571.
4. Lowe JK, Kukekova AV, Kirkness EF, et al. Linkage mapping of the primary disease locus for Collie eye anomaly. *Genomics*. 2003;82:86-95.



# OCULAR DISORDERS REPORT NOVA SCOTIA DUCK TOLLING RET.

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>5,440 |       | 2016-2020<br>1,258 |       |
|---------------------|--|--------------------|-------|--------------------|-------|
|                     |  | #                  | %     | #                  | %     |
| <b>GLOBE</b>        |  |                    |       |                    |       |
| 0.110               | microphthalmia   | 1                  | 0.0%  | 2                  | 0.2%  |
| 10.000              | glaucoma   | 1                  | 0.0%  | 0                  |       |
| <b>EYELIDS</b>      |  |                    |       |                    |       |
| 20.140              | ectopic cilia  | 1                  | 0.0%  | 0                  |       |
| 25.110              | distichiasis   | 670                | 12.3% | 149                | 11.8% |
| <b>NASOLACRIMAL</b> |  |                    |       |                    |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 6                  | 0.1%  | 7                  | 0.6%  |
| 40.910              | keratoconjunctivitis sicca                                     | 1                  | 0.0%  | 0                  |       |
| <b>NICTITANS</b>    |  |                    |       |                    |       |
| 51.100              | third eyelid cartilage anomaly                                 | 5                  | 0.1%  | 0                  |       |
| 52.110              | prolapsed gland of the third eyelid                            | 5                  | 0.1%  | 0                  |       |
| <b>CORNEA</b>       |  |                    |       |                    |       |
| 70.700              | corneal dystrophy  | 141                | 2.6%  | 36                 | 2.9%  |
| 70.730              | corneal endothelial degeneration                               | 4                  | 0.1%  | 0                  |       |
| <b>UVEA</b>         |  |                    |       |                    |       |
| 93.140              | corneal endothelial pigment without PPM                        | 1                  | 0.0%  | 0                  |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 118                | 2.2%  | 35                 | 2.8%  |
| 93.720              | persistent pupillary membranes, iris to lens                   | 53                 | 1.0%  | 1                  | 0.1%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 2                  | 0.0%  | 1                  | 0.1%  |
| 93.740              | persistent pupillary membranes, iris sheets                    | 8                  | 0.1%  | 0                  |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 117                | 2.2%  | 82                 | 6.5%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 2                  | 0.0%  | 0                  |       |
| 93.999              | uveal cysts  | 21                 | 0.4%  | 3                  | 0.2%  |
| <b>LENS</b>         |  |                    |       |                    |       |
| 100.200             | cataract, unspecified  | 18                 | 0.3%  | 0                  |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 309                | 5.7%  | 83                 | 6.6%  |
| 100.301             | punctate cataract, anterior cortex                             | 19                 | 0.3%  | 4                  | 0.3%  |
| 100.302             | punctate cataract, posterior cortex                            | 25                 | 0.5%  | 2                  | 0.2%  |
| 100.303             | punctate cataract, equatorial cortex                           | 10                 | 0.2%  | 1                  | 0.1%  |
| 100.305             | punctate cataract, posterior sutures                           | 5                  | 0.1%  | 6                  | 0.5%  |
| 100.306             | punctate cataract, nucleus                                     | 6                  | 0.1%  | 4                  | 0.3%  |
| 100.307             | punctate cataract, capsular                                    | 9                  | 0.2%  | 5                  | 0.4%  |
| 100.311             | incipient cataract, anterior cortex                            | 17                 | 0.3%  | 2                  | 0.2%  |
| 100.312             | incipient cataract, posterior cortex                           | 34                 | 0.6%  | 2                  | 0.2%  |
| 100.313             | incipient cataract, equatorial cortex                          | 16                 | 0.3%  | 1                  | 0.1%  |
| 100.314             | incipient cataract, anterior sutures                           | 1                  | 0.0%  | 1                  | 0.1%  |
| 100.315             | incipient cataract, posterior sutures                          | 3                  | 0.1%  | 1                  | 0.1%  |
| 100.316             | incipient cataract, nucleus                                    | 9                  | 0.2%  | 0                  |       |
| 100.317             | incipient cataract, capsular                                   | 7                  | 0.1%  | 0                  |       |
| 100.321             | incomplete cataract, anterior cortex                           | 3                  | 0.1%  | 0                  |       |
| 100.322             | incomplete cataract, posterior cortex                          | 1                  | 0.0%  | 1                  | 0.1%  |
| 100.328             | y-suture tip opacities   | 2                  | 0.0%  | 11                 | 0.9%  |
| 100.330             | generalized/complete cataract                                  | 7                  | 0.1%  | 0                  |       |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.345 <i>significant cataracts (summary)</i>            | 192 3.5%         | 41 3.3%          |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 9 0.2%           | 15 1.2%          |
| 110.135 PHPV/PTVL   | 7 0.1%           | 1 0.1%           |
| 110.200 vitreous degeneration-anterior chamber            | 0                | 1 0.1%           |
| 110.320 vitreal degeneration                              | 13 0.2%          | 1 0.1%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 2 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 47 0.9%          | 5 0.4%           |
| 120.180 retinal dysplasia, geographic                     | 13 0.2%          | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 97 1.8%          | 1 0.1%           |
| 120.920 retinal detachment with dialysis                  | 1 0.0%           | 0                |
| 120.960 retinopathy                                       | 1 0.0%           | 1 0.1%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 12 0.2%          | 1 0.1%           |
| 130.120 optic nerve hypoplasia                            | 13 0.2%          | 1 0.1%           |
| 130.150 optic disc coloboma                               | 3 0.1%           | 1 0.1%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 98 1.8%          | 0                |
| 900.100 other, not inherited                              | 279 5.1%         | 1 0.1%           |
| 900.110 other. suspect not inherited/significance unknown | 89 1.6%          | 79 6.3%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 4,105 75.5%      | 812 64.5%        |

# OLD ENGLISH SHEEPDOG

|    | DISORDER                                      | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|---|-------------|-----------|-----------------|
| A. | Microphthalmia with multiple ocular anomalies | Not defined | 1, 2      | NO              |
| B. | Distichiasis                                  | Not defined | 1         | Breeder option  |
| C. | Corneal dystrophy - epithelial/stromal        | Not defined | 1         | Breeder option  |
| D. | Persistent pupillary membranes - iris to iris | Not defined | 1         | Breeder option  |
| E. | Cataract                                      | Not defined | 1, 3      | NO              |
| F. | Retinal dysplasia - folds                     | Not defined | 1         | Breeder option  |

## Description and Comments

### A. Microphthalmia with multiple congenital ocular defects

Microphthalmia is a developmental anomaly in which the eyeball is abnormally small. This is often associated with other ocular malformations, including defects of the cornea, anterior chamber, lens and/or retina.

Microphthalmia with cataract and retinal abnormalities including retinal detachment, has been reported in litters of Old English Sheepdogs. Lesions were non-progressive. However, blindness did result in some dogs. The mode of inheritance is unknown, but affected dogs should not be bred.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal

layers; usually inherited and bilateral.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region. In one study of 66 interrelated Old English Sheepdogs, an autosomal recessive mode of inheritance was suggested. Retinal detachment was an associated finding in 5/43 affected dogs in this study. The location of the opacity within the lens and the age of onset was highly variable.

F. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Barrie K. Posterior lenticonus, microphthalmia, cataracts and retinal folds in Old English Sheepdogs. *J Am Anim Hosp Assoc.* 1979;15:715.
3. Koch SA. Cataracts in interrelated Old English Sheepdogs. *J Am Vet Med Assoc.* 1972 Feb 1;160:299-301.

# OCULAR DISORDERS REPORT OLD ENGLISH SHEEPDOG

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>4,945 |      | 2016-2020<br>933 |       |
|---------------------|--|--------------------|------|------------------|-------|
|                     |  | #                  | %    | #                | %     |
| <b>GLOBE</b>        |  |                    |      |                  |       |
| 0.110               | microphthalmia   | 10                 | 0.2% | 0                |       |
| 10.000              | glaucoma   | 4                  | 0.1% | 0                |       |
| <b>EYELIDS</b>      |  |                    |      |                  |       |
| 20.140              | ectopic cilia  | 1                  | 0.0% | 0                |       |
| 20.160              | macropalpebral fissure   | 1                  | 0.0% | 0                |       |
| 21.000              | entropion, unspecified   | 12                 | 0.2% | 1                | 0.1%  |
| 22.000              | ectropion, unspecified   | 2                  | 0.0% | 0                |       |
| 25.110              | distichiasis   | 84                 | 1.7% | 16               | 1.7%  |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 2                  | 0.0% | 1                | 0.1%  |
| <b>NICTITANS</b>    |  |                    |      |                  |       |
| 51.100              | third eyelid cartilage anomaly                                 | 1                  | 0.0% | 0                |       |
| 52.110              | prolapsed gland of the third eyelid                            | 1                  | 0.0% | 0                |       |
| <b>CORNEA</b>       |  |                    |      |                  |       |
| 70.700              | corneal dystrophy  | 18                 | 0.4% | 3                | 0.3%  |
| <b>UVEA</b>         |  |                    |      |                  |       |
| 93.140              | corneal endothelial pigment without PPM                        | 1                  | 0.0% | 0                |       |
| 93.150              | iris coloboma  | 1                  | 0.0% | 0                |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 420                | 8.5% | 148              | 15.9% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 7                  | 0.1% | 3                | 0.3%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 9                  | 0.2% | 1                | 0.1%  |
| 93.740              | persistent pupillary membranes, iris sheets                    | 10                 | 0.2% | 0                |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 2                  | 0.0% | 3                | 0.3%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 3                  | 0.1% | 0                |       |
| 93.810              | uveal melanoma   | 0                  |      | 1                | 0.1%  |
| 93.999              | uveal cysts  | 0                  |      | 2                | 0.2%  |
| <b>LENS</b>         |  |                    |      |                  |       |
| 100.200             | cataract, unspecified  | 35                 | 0.7% | 0                |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 260                | 5.3% | 61               | 6.5%  |
| 100.301             | punctate cataract, anterior cortex                             | 33                 | 0.7% | 11               | 1.2%  |
| 100.302             | punctate cataract, posterior cortex                            | 9                  | 0.2% | 1                | 0.1%  |
| 100.303             | punctate cataract, equatorial cortex                           | 6                  | 0.1% | 3                | 0.3%  |
| 100.304             | punctate cataract, anterior sutures                            | 5                  | 0.1% | 1                | 0.1%  |
| 100.305             | punctate cataract, posterior sutures                           | 5                  | 0.1% | 2                | 0.2%  |
| 100.306             | punctate cataract, nucleus                                     | 12                 | 0.2% | 4                | 0.4%  |
| 100.307             | punctate cataract, capsular                                    | 6                  | 0.1% | 4                | 0.4%  |
| 100.311             | incipient cataract, anterior cortex                            | 44                 | 0.9% | 9                | 1.0%  |
| 100.312             | incipient cataract, posterior cortex                           | 43                 | 0.9% | 8                | 0.9%  |
| 100.313             | incipient cataract, equatorial cortex                          | 17                 | 0.3% | 1                | 0.1%  |
| 100.314             | incipient cataract, anterior sutures                           | 11                 | 0.2% | 1                | 0.1%  |
| 100.315             | incipient cataract, posterior sutures                          | 13                 | 0.3% | 0                |       |
| 100.316             | incipient cataract, nucleus                                    | 30                 | 0.6% | 3                | 0.3%  |
| 100.317             | incipient cataract, capsular                                   | 5                  | 0.1% | 1                | 0.1%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.321 incomplete cataract, anterior cortex              | 2 0.0%           | 1 0.1%           |
| 100.322 incomplete cataract, posterior cortex             | 2 0.0%           | 2 0.2%           |
| 100.326 incomplete cataract, nucleus                      | 1 0.0%           | 1 0.1%           |
| 100.327 incomplete cataract, capsular                     | 0                | 1 0.1%           |
| 100.328 y-suture tip opacities                            | 1 0.0%           | 3 0.3%           |
| 100.330 generalized/complete cataract                     | 61 1.2%          | 1 0.1%           |
| 100.340 resorbing/hypermature cataract                    | 2 0.0%           | 0                |
| 100.345 <i>significant cataracts (summary)</i>            | 343 6.9%         | 58 6.2%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 6 0.1%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 16 0.3%          | 3 0.3%           |
| 110.135 PHPV/PTVL   | 3 0.1%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 3 0.1%           | 0                |
| 110.320 vitreal degeneration                              | 26 0.5%          | 0                |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 3 0.1%           | 1 0.1%           |
| 97.120 coloboma   | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 87 1.8%          | 8 0.9%           |
| 120.180 retinal dysplasia, geographic                     | 8 0.2%           | 0                |
| 120.190 retinal dysplasia, detached                       | 2 0.0%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 13 0.3%          | 0                |
| 120.400 retinal hemorrhage                                | 1 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 9 0.2%           | 0                |
| 120.960 retinopathy                                       | 0                | 5 0.5%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 14 0.3%          | 9 1.0%           |
| 130.120 optic nerve hypoplasia                            | 15 0.3%          | 0                |
| 130.150 optic disc coloboma                               | 4 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 35 0.7%          | 0                |
| 900.100 other, not inherited                              | 76 1.5%          | 2 0.2%           |
| 900.110 other. suspect not inherited/significance unknown | 44 0.9%          | 28 3.0%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 3,969 80.3%      | 641 68.7%        |

# **OCULAR DISORDERS REPORT OLDE ENGLISH BULLDOGGE**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the OLDE ENGLISH BULLDOGGE breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT OLDE ENGLISH BULLDOGGE

| Diagnostic Name | TOTAL DOGS EXAMINED                                  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 21.000          | entropion, unspecified                               | 0         |       | 2         | 11.8% |
| 25.110          | distichiasis   | 5         | 45.5% | 3         | 17.6% |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.110          | iris hypoplasia                                      | 0         |       | 1         | 5.9%  |
| 93.710          | persistent pupillary membranes, iris to iris         | 1         | 9.1%  | 0         |       |
| 93.720          | persistent pupillary membranes, iris to lens         | 1         | 9.1%  | 0         |       |
| 93.999          | uveal cysts  | 0         |       | 1         | 5.9%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown | 0         |       | 1         | 5.9%  |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.170         | retinal dysplasia, folds                             | 0         |       | 1         | 5.9%  |
| 120.180         | retinal dysplasia, geographic                        | 0         |       | 1         | 5.9%  |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.110         | other. suspect not inherited/significance unknown    | 0         |       | 2         | 11.8% |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 5         | 45.5% | 8         | 47.1% |



# **OCULAR DISORDERS REPORT OTTERHOUND**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the OTTERHOUND breed. Therefore, there are no conditions listed with breeding advice.

# OCULAR DISORDERS REPORT OTTERHOUND

| Diagnostic Name                                     | TOTAL DOGS EXAMINED | 1991-2015 |        | 2016-2020 |       |
|---|---------------------|-----------|--------|-----------|-------|
|   |                     | #         | %      | #         | %     |
| <b>UVEA</b>   |                     |           |        |           |       |
| 93.710 persistent pupillary membranes, iris to iris |                     | 1         | 16.7%  | 0         |       |
| 93.999 uveal cysts                                  |                     | 0         |        | 1         | 50.0% |
| <b>NORMAL</b>                                       |                     |           |        |           |       |
| 0.000 normal globe                                  |                     | 6         | 100.0% | 1         | 50.0% |

# PAPILLON

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE           |
|----|---|------------------------|-----------|--------------------|--------------------------------------|
| A. | Distichiasis  | Not defined            | 1         | Breeder option     |                                      |
| B. | Corneal dystrophy<br>- epithelial/stromal           | Not defined            | 1         | Breeder option     |                                      |
| C. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined            | 1         | Breeder option     |                                      |
| D. | Cataract  | Not defined            | 1         | NO                 |                                      |
| E. | Vitreous degeneration                               | Not defined            | 1         | Breeder option     |                                      |
| F. | Retinal atrophy<br>( <i>CNGB1</i> )                 | Autosomal<br>recessive | 1, 2-5    | NO                 | Mutation in the<br><i>CNGB1</i> gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur

#### D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

Nuclear and posterior cortical cataracts have been reported in the Papillon.

#### E. Vitreous degeneration

A liquefaction of the vitreous gel, which may predispose to retinal detachment resulting in blindness.

#### F. Retinal atrophy - *CNGB1*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. In one study of 707 dogs in Sweden, an autosomal recessive mode of inheritance was suggested. Clinical onset is reported at 5-6 years of age. In approximately 70% of cases of PRA in the Papillon, a *CNGB1* mutation is present, leading to an abnormal *CNGB1* protein in the rod outer segments. The mode of transmission is autosomal recessive. A genetic test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Haakanson N, Narfstrom K. Progressive retinal atrophy in Papillon dogs in Sweden: A clinical survey. *Prog Vet Comp Ophthalmol*. 1995;5:83.
3. Narfstrom K, Ekesten B. Electroretinographic evaluation of Papillons with and without hereditary retinal degeneration. *Am J Vet Res*. 1998;59:221-226.
4. Ahonen SJ, Arumilli M, Lohi H. A *CNGB1* frameshift mutation in Papillon and Phalene dogs with progressive retinal atrophy. *PLoS One*. 2013;8:e72122.
5. Winkler PA, Ekenstedt KJ, Occelli LM, et al. A large animal model for *CNGB1* autosomal recessive retinitis pigmentosa. *PLoS One*. 2013;8:e72229.

# OCULAR DISORDERS REPORT PAPILLON

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>10,550 |      | 2016-2020<br>1,392 |      |
|---------------------|--|---------------------|------|--------------------|------|
|                     |  | #                   | %    | #                  | %    |
| <b>GLOBE</b>        |  |                     |      |                    |      |
| 0.110               | microphthalmia   | 9                   | 0.1% | 1                  | 0.1% |
| 10.000              | glaucoma   | 1                   | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                     |      |                    |      |
| 21.000              | entropion, unspecified   | 16                  | 0.2% | 5                  | 0.4% |
| 25.110              | distichiasis   | 145                 | 1.4% | 27                 | 1.9% |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 3                   | 0.0% | 5                  | 0.4% |
| 40.910              | keratoconjunctivitis sicca                                     | 1                   | 0.0% | 0                  |      |
| <b>NICTITANS</b>    |  |                     |      |                    |      |
| 52.110              | prolapsed gland of the third eyelid                            | 3                   | 0.0% | 0                  |      |
| <b>CORNEA</b>       |  |                     |      |                    |      |
| 70.210              | corneal pannus   | 5                   | 0.0% | 1                  | 0.1% |
| 70.220              | pigmentary keratitis   | 2                   | 0.0% | 0                  |      |
| 70.700              | corneal dystrophy  | 99                  | 0.9% | 34                 | 2.4% |
| 70.730              | corneal endothelial degeneration                               | 4                   | 0.0% | 0                  |      |
| <b>UVEA</b>         |  |                     |      |                    |      |
| 93.110              | iris hypoplasia  | 2                   | 0.0% | 1                  | 0.1% |
| 93.140              | corneal endothelial pigment without PPM                        | 1                   | 0.0% | 0                  |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 312                 | 3.0% | 57                 | 4.1% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 7                   | 0.1% | 1                  | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 8                   | 0.1% | 1                  | 0.1% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 6                   | 0.1% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 13                  | 0.1% | 5                  | 0.4% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 5                   | 0.0% | 3                  | 0.2% |
| 93.810              | uveal melanoma   | 0                   |      | 3                  | 0.2% |
| 93.999              | uveal cysts  | 4                   | 0.0% | 1                  | 0.1% |
| <b>LENS</b>         |  |                     |      |                    |      |
| 100.200             | cataract, unspecified  | 19                  | 0.2% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 341                 | 3.2% | 56                 | 4.0% |
| 100.301             | punctate cataract, anterior cortex                             | 53                  | 0.5% | 4                  | 0.3% |
| 100.302             | punctate cataract, posterior cortex                            | 17                  | 0.2% | 2                  | 0.1% |
| 100.303             | punctate cataract, equatorial cortex                           | 11                  | 0.1% | 1                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 4                   | 0.0% | 2                  | 0.1% |
| 100.305             | punctate cataract, posterior sutures                           | 7                   | 0.1% | 4                  | 0.3% |
| 100.306             | punctate cataract, nucleus                                     | 15                  | 0.1% | 3                  | 0.2% |
| 100.307             | punctate cataract, capsular                                    | 7                   | 0.1% | 5                  | 0.4% |
| 100.311             | incipient cataract, anterior cortex                            | 80                  | 0.8% | 8                  | 0.6% |
| 100.312             | incipient cataract, posterior cortex                           | 51                  | 0.5% | 6                  | 0.4% |
| 100.313             | incipient cataract, equatorial cortex                          | 30                  | 0.3% | 2                  | 0.1% |
| 100.314             | incipient cataract, anterior sutures                           | 6                   | 0.1% | 0                  |      |
| 100.315             | incipient cataract, posterior sutures                          | 10                  | 0.1% | 0                  |      |
| 100.316             | incipient cataract, nucleus                                    | 21                  | 0.2% | 2                  | 0.1% |
| 100.317             | incipient cataract, capsular                                   | 10                  | 0.1% | 2                  | 0.1% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.321 incomplete cataract, anterior cortex              | 2 0.0%           | 1 0.1%           |
| 100.322 incomplete cataract, posterior cortex             | 3 0.0%           | 2 0.1%           |
| 100.323 incomplete cataract, equatorial cortex            | 1 0.0%           | 0                |
| 100.326 incomplete cataract, nucleus                      | 3 0.0%           | 0                |
| 100.328 y-suture tip opacities                            | 0                | 4 0.3%           |
| 100.330 generalized/complete cataract                     | 45 0.4%          | 3 0.2%           |
| 100.340 resorbing/hypermature cataract                    | 0                | 1 0.1%           |
| 100.345 <i>significant cataracts (summary)</i>            | 395 3.7%         | 52 3.7%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 5 0.0%           | 1 0.1%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 38 0.4%          | 6 0.4%           |
| 110.135 PHPV/PTVL   | 14 0.1%          | 1 0.1%           |
| 110.200 vitreous degeneration-anterior chamber            | 4 0.0%           | 11 0.8%          |
| 110.320 vitreal degeneration                              | 303 2.9%         | 23 1.7%          |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 0                | 1 0.1%           |
| 97.120 coloboma   | 2 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 66 0.6%          | 5 0.4%           |
| 120.180 retinal dysplasia, geographic                     | 11 0.1%          | 3 0.2%           |
| 120.190 retinal dysplasia, detached                       | 3 0.0%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 110 1.0%         | 7 0.5%           |
| 120.400 retinal hemorrhage                                | 1 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 8 0.1%           | 0                |
| 120.920 retinal detachment with dialysis                  | 1 0.0%           | 1 0.1%           |
| 120.960 retinopathy                                       | 1 0.0%           | 3 0.2%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 8 0.1%           | 0                |
| 130.120 optic nerve hypoplasia                            | 10 0.1%          | 2 0.1%           |
| 130.150 optic disc coloboma                               | 3 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 77 0.7%          | 0                |
| 900.100 other, not inherited                              | 202 1.9%         | 3 0.2%           |
| 900.110 other. suspect not inherited/significance unknown | 75 0.7%          | 54 3.9%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 9,138 86.6%      | 1,081 77.7%      |

# PARSON RUSSELL TERRIER

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|--|---------------------|-----------|-----------------|--------------------------------------|
| A. | Distichiasis                                     | Not defined         | 1         | Breeder option  |                                      |
| B. | Persistent pupillary membranes<br>- iris to iris | Not define          | 1         | Breeder options |                                      |
| C. | Cataract   | Not defined         | 1, 2      | NO              |                                      |
| D. | Lens luxation                                    | Autosomal recessive | 3, 4      | NO              | Mutation in the <i>ADAMTS17</i> gene |
| E. | Vitreous degeneration                            | Not defined         | 1         | Breeder option  |                                      |
| F. | Retinal atrophy<br>- generalized                 | Not defined         | 1         | NO              |                                      |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation may result in blinding retinal detachment and/or elevated intraocular pressure (glaucoma) causing vision impairment, pain, and blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

E. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment resulting in blindness.

F. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as Progressive Retinal Atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Oberbauer AM, Hollingsworth SR, Belanger JM, et al. Inheritance of cataracts and primary lens luxation in Jack Russell Terriers. *Am J Vet Res.* 2008;69:222-227.
3. Farias FH, Johnson GS, Taylor JF, et al. An *ADAMTS17* splice donor site mutation in dogs with primary lens luxation. *Invest Ophthalmol Vis Sci.* 2010;51:4716-4721.
4. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011;14:378-384.



# OCULAR DISORDERS REPORT

## PARSON RUSSELL TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |       |
|---|---------------------|-----------|------|-----------|-------|
|   |                     | #         | %    | #         | %     |
| <b>EYELIDS</b>  |                     |           |      |           |       |
| 25.110 distichiasis   |                     | 64        | 2.5% | 8         | 2.7%  |
| <b>NICTITANS</b>  |                     |           |      |           |       |
| 52.110 prolapsed gland of the third eyelid                            |                     | 0         |      | 1         | 0.3%  |
| <b>CORNEA</b>   |                     |           |      |           |       |
| 70.700 corneal dystrophy  |                     | 14        | 0.5% | 0         |       |
| 70.730 corneal endothelial degeneration                               |                     | 2         | 0.1% | 0         |       |
| <b>UVEA</b>   |                     |           |      |           |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 162       | 6.2% | 34        | 11.7% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 1         | 0.0% | 0         |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 3         | 0.1% | 0         |       |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 1         | 0.0% | 0         |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 5         | 0.2% | 3         | 1.0%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 3         | 0.1% | 2         | 0.7%  |
| 93.999 uveal cysts  |                     | 2         | 0.1% | 0         |       |
| <b>LENS</b>   |                     |           |      |           |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 82        | 3.1% | 12        | 4.1%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 7         | 0.3% | 3         | 1.0%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 8         | 0.3% | 0         |       |
| 100.303 punctate cataract, equatorial cortex                          |                     | 4         | 0.2% | 0         |       |
| 100.304 punctate cataract, anterior sutures                           |                     | 0         |      | 1         | 0.3%  |
| 100.305 punctate cataract, posterior sutures                          |                     | 4         | 0.2% | 2         | 0.7%  |
| 100.306 punctate cataract, nucleus                                    |                     | 2         | 0.1% | 0         |       |
| 100.307 punctate cataract, capsular                                   |                     | 2         | 0.1% | 1         | 0.3%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 15        | 0.6% | 2         | 0.7%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 39        | 1.5% | 2         | 0.7%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 6         | 0.2% | 1         | 0.3%  |
| 100.314 incipient cataract, anterior sutures                          |                     | 1         | 0.0% | 0         |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 13        | 0.5% | 2         | 0.7%  |
| 100.316 incipient cataract, nucleus                                   |                     | 1         | 0.0% | 0         |       |
| 100.317 incipient cataract, capsular                                  |                     | 9         | 0.3% | 0         |       |
| 100.321 incomplete cataract, anterior cortex                          |                     | 0         |      | 1         | 0.3%  |
| 100.322 incomplete cataract, posterior cortex                         |                     | 2         | 0.1% | 1         | 0.3%  |
| 100.328 y-suture tip opacities  |                     | 0         |      | 2         | 0.7%  |
| 100.330 generalized/complete cataract                                 |                     | 11        | 0.4% | 0         |       |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 124       | 4.7% | 18        | 6.2%  |
| 100.375 <i>subluxation/luxation, unspecified</i>                      |                     | 1         | 0.0% | 0         |       |
| <b>VITREOUS</b>   |                     |           |      |           |       |
| 110.120 persistent hyaloid artery/remnant                             |                     | 4         | 0.2% | 2         | 0.7%  |
| 110.135 PHPV/PTVL   |                     | 1         | 0.0% | 0         |       |
| 110.320 vitreal degeneration  |                     | 45        | 1.7% | 1         | 0.3%  |
| <b>FUNDUS</b>   |                     |           |      |           |       |
| 97.120 coloboma   |                     | 1         | 0.0% | 0         |       |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 8 0.3%      | 2 0.7%    |
| 120.180 retinal dysplasia, geographic                     | 2 0.1%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 25 1.0%     | 0         |
| 120.910 retinal detachment without dialysis               | 1 0.0%      | 0         |
| 120.920 retinal detachment with dialysis                  | 0           | 1 0.3%    |
| 120.960 retinopathy                                       | 1 0.0%      | 0         |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 2 0.1%      | 0         |
| 130.120 optic nerve hypoplasia                            | 2 0.1%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 39 1.5%     | 0         |
| 900.100 other, not inherited                              | 97 3.7%     | 0         |
| 900.110 other. suspect not inherited/significance unknown | 24 0.9%     | 20 6.9%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 2,263 86.7% | 208 71.5% |

# PATTERDALE TERRIER

|    | DISORDER      | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|---------------|------------------------|-----------|--------------------|---|
| A. | Lens luxation | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>ADAMTS17</i> gene |

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## Description and Comments

### A. Lens Luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Patterdale Terrier. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011; 14: 378-384.

# OCULAR DISORDERS REPORT PATTERDALE TERRIER

| Diagnostic Name | TOTAL DOGS EXAMINED           | 1991-2015 |       | 2016-2020 |        |
|-----------------|-------------------------------|-----------|-------|-----------|--------|
|                 |                               | #         | %     | #         | %      |
| <b>EYELIDS</b>  |                               |           |       |           |        |
| 25.110          | distichiasis                  | 1         | 6.7%  | 0         |        |
| <b>RETINA</b>   |                               |           |       |           |        |
| 120.170         | retinal dysplasia, folds      | 1         | 6.7%  | 0         |        |
| 120.180         | retinal dysplasia, geographic | 1         | 6.7%  | 0         |        |
| <b>NORMAL</b>   |                               |           |       |           |        |
| 0.000           | normal globe                  | 13        | 86.7% | 2         | 100.0% |

# PEKINGESE

|    | DISORDER                      | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|-------------------------------|-------------|-----------|-----------------|
| A. | Distichiasis                  | Not defined | 1-3       | Breeder option  |
| B. | Entropion                     | Not defined | 1         | Breeder option  |
| C. | Exposure keratopathy syndrome | Not defined | 1         | Breeder option  |
| D. | Cataract                      | Not defined | 1         | NO              |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### C. Exposure keratopathy syndrome

A corneal disease involving all or part of the cornea, resulting from inadequate blinking. This results from a combination of anatomic features including shallow orbits, exophthalmos, macroblepharon and lagophthalmos. Macroblepharon is defined as an exceptionally large palpebral fissure, macroblepharon in conjunction with laxity of the lateral canthal structures may lead to lower lid ectropion and upper lid entropion. Either of these conditions may lead to severe ocular irritation.

### D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Barnett KC. Comparative aspects of canine hereditary eye disease. *Adv Vet Sci Comp Med.* 1976;20:39-67.
3. Gelatt KN. Pediatric ophthalmology in small animal practice. *Vet Clin North Am.* 1973;3:321.
4. Priester W. Canine progressive retinal atrophy: Occurrence by age, breed, and sex. *American Journal of Veterinary Research.* 1974;35:571-574.

# OCULAR DISORDERS REPORT PEKINGESE

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 0.110 microphthalmia  |                     | 1         | 0.5%  | 0         |       |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 20.140 ectopic cilia  |                     | 2         | 1.0%  | 0         |       |
| 20.160 macropalpebral fissure                                       |                     | 12        | 6.1%  | 0         |       |
| 21.000 entropion, unspecified                                       |                     | 11        | 5.6%  | 32        | 31.4% |
| 22.000 ectropion, unspecified                                       |                     | 2         | 1.0%  | 0         |       |
| 25.110 distichiasis   |                     | 21        | 10.6% | 6         | 5.9%  |
| <b>NASOLACRIMAL</b>   |                     |           |       |           |       |
| 40.910 keratoconjunctivitis sicca                                   |                     | 1         | 0.5%  | 1         | 1.0%  |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.210 corneal pannus   |                     | 7         | 3.5%  | 0         |       |
| 70.220 pigmentary keratitis   |                     | 29        | 14.6% | 18        | 17.6% |
| <b>UVEA</b>   |                     |           |       |           |       |
| 90.250 pigmentary uveitis   |                     | 0         |       | 1         | 1.0%  |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 0         |       | 1         | 1.0%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 1         | 0.5%  | 0         |       |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.200 cataract, unspecified                                       |                     | 3         | 1.5%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 3         | 1.5%  | 2         | 2.0%  |
| 100.301 punctate cataract, anterior cortex                          |                     | 3         | 1.5%  | 1         | 1.0%  |
| 100.302 punctate cataract, posterior cortex                         |                     | 2         | 1.0%  | 1         | 1.0%  |
| 100.303 punctate cataract, equatorial cortex                        |                     | 0         |       | 1         | 1.0%  |
| 100.305 punctate cataract, posterior sutures                        |                     | 1         | 0.5%  | 0         |       |
| 100.311 incipient cataract, anterior cortex                         |                     | 5         | 2.5%  | 0         |       |
| 100.312 incipient cataract, posterior cortex                        |                     | 3         | 1.5%  | 0         |       |
| 100.313 incipient cataract, equatorial cortex                       |                     | 4         | 2.0%  | 0         |       |
| 100.315 incipient cataract, posterior sutures                       |                     | 3         | 1.5%  | 0         |       |
| 100.316 incipient cataract, nucleus                                 |                     | 1         | 0.5%  | 0         |       |
| 100.330 generalized/complete cataract                               |                     | 2         | 1.0%  | 0         |       |
| 100.340 resorbing/hypermature cataract                              |                     | 0         |       | 1         | 1.0%  |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 27        | 13.6% | 4         | 3.9%  |
| 100.375 <i>subluxation/luxation, unspecified</i>                    |                     | 2         | 1.0%  | 0         |       |
| <b>RETINA</b>   |                     |           |       |           |       |
| 120.170 retinal dysplasia, folds                                    |                     | 1         | 0.5%  | 0         |       |
| 120.190 retinal dysplasia, detached                                 |                     | 1         | 0.5%  | 0         |       |
| 120.310 generalized progressive retinal atrophy (PRA)               |                     | 3         | 1.5%  | 0         |       |
| <b>OPTIC NERVE</b>  |                     |           |       |           |       |
| 130.110 micropapilla  |                     | 0         |       | 1         | 1.0%  |
| 130.120 optic nerve hypoplasia                                      |                     | 1         | 0.5%  | 0         |       |
| <b>OTHER</b>  |                     |           |       |           |       |
| 900.000 other, unspecified  |                     | 6         | 3.0%  | 0         |       |
| 900.100 other, not inherited  |                     | 11        | 5.6%  | 0         |       |

| <b>OTHER CONTINUED</b>                                    | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 900.110 other. suspect not inherited/significance unknown | 7 3.5%           | 7 6.9%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 110 55.6%        | 52 51.0%         |



# PEMBROKE WELSH CORGI

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--------------------------------|-------------|-----------|-----------------|
| A. | Distichiasis                   | Not defined | 1         | Breeder option  |
| B. | Persistent pupillary membranes |             |           |                 |
|    | - iris to iris                 | Not defined | 1         | Breeder option  |
|    | - iris to cornea               | Not defined | 1         | NO              |
| C. | Cataract                       | Not defined | 1         | NO              |
| D. | Retinal dysplasia              | Not defined | 1, 2      | Breeder option  |
|    | - folds                        |             |           |                 |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Persistent pupillary membranes are a significant problem in this breed with frequent documentation of strands bridging from the iris to the cornea noted on routine screening eye examinations. These may be associated with corneal opacity which may result in vision impairment, thus the recommendation against breeding Pembroke Welsh Corgis with PPM.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

There are no specific references providing detailed descriptions of hereditary ocular conditions of the Pembroke Welsh Corgi. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT PEMBROKE WELSH CORGI

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>19,021 |       | 2016-2020<br>3,809 |       |
|---------------------|--|---------------------|-------|--------------------|-------|
|                     |  | #                   | %     | #                  | %     |
| <b>GLOBE</b>        |  |                     |       |                    |       |
| 0.110               | microphthalmia   | 19                  | 0.1%  | 2                  | 0.1%  |
| 10.000              | glaucoma   | 1                   | 0.0%  | 0                  |       |
| <b>EYELIDS</b>      |  |                     |       |                    |       |
| 20.140              | ectopic cilia  | 3                   | 0.0%  | 0                  |       |
| 22.000              | ectropion, unspecified   | 1                   | 0.0%  | 0                  |       |
| 25.110              | distichiasis   | 331                 | 1.7%  | 51                 | 1.3%  |
| <b>NASOLACRIMAL</b> |  |                     |       |                    |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 3                   | 0.0%  | 6                  | 0.2%  |
| 40.910              | keratoconjunctivitis sicca                                     | 6                   | 0.0%  | 1                  | 0.0%  |
| <b>NICTITANS</b>    |  |                     |       |                    |       |
| 51.100              | third eyelid cartilage anomaly                                 | 1                   | 0.0%  | 0                  |       |
| 52.110              | prolapsed gland of the third eyelid                            | 2                   | 0.0%  | 0                  |       |
| <b>CORNEA</b>       |  |                     |       |                    |       |
| 70.210              | corneal pannus   | 3                   | 0.0%  | 0                  |       |
| 70.220              | pigmentary keratitis   | 1                   | 0.0%  | 1                  | 0.0%  |
| 70.700              | corneal dystrophy  | 64                  | 0.3%  | 10                 | 0.3%  |
| 70.730              | corneal endothelial degeneration                               | 67                  | 0.4%  | 4                  | 0.1%  |
| <b>UVEA</b>         |  |                     |       |                    |       |
| 93.110              | iris hypoplasia  | 3                   | 0.0%  | 3                  | 0.1%  |
| 93.140              | corneal endothelial pigment without PPM                        | 8                   | 0.0%  | 0                  |       |
| 93.150              | iris coloboma  | 5                   | 0.0%  | 0                  |       |
| 93.180              | iris sphincter dysplasia                                       | 1                   | 0.0%  | 0                  |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 3,418               | 18.0% | 817                | 21.4% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 63                  | 0.3%  | 12                 | 0.3%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 397                 | 2.1%  | 38                 | 1.0%  |
| 93.740              | persistent pupillary membranes, iris sheets                    | 15                  | 0.1%  | 0                  |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 2                   | 0.0%  | 1                  | 0.0%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 49                  | 0.3%  | 30                 | 0.8%  |
| 93.999              | uveal cysts  | 11                  | 0.1%  | 0                  |       |
| <b>LENS</b>         |  |                     |       |                    |       |
| 100.200             | cataract, unspecified  | 79                  | 0.4%  | 0                  |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 433                 | 2.3%  | 79                 | 2.1%  |
| 100.301             | punctate cataract, anterior cortex                             | 61                  | 0.3%  | 13                 | 0.3%  |
| 100.302             | punctate cataract, posterior cortex                            | 51                  | 0.3%  | 13                 | 0.3%  |
| 100.303             | punctate cataract, equatorial cortex                           | 25                  | 0.1%  | 4                  | 0.1%  |
| 100.304             | punctate cataract, anterior sutures                            | 3                   | 0.0%  | 0                  |       |
| 100.305             | punctate cataract, posterior sutures                           | 20                  | 0.1%  | 4                  | 0.1%  |
| 100.306             | punctate cataract, nucleus                                     | 52                  | 0.3%  | 12                 | 0.3%  |
| 100.307             | punctate cataract, capsular                                    | 22                  | 0.1%  | 8                  | 0.2%  |
| 100.311             | incipient cataract, anterior cortex                            | 94                  | 0.5%  | 21                 | 0.6%  |
| 100.312             | incipient cataract, posterior cortex                           | 173                 | 0.9%  | 24                 | 0.6%  |
| 100.313             | incipient cataract, equatorial cortex                          | 63                  | 0.3%  | 7                  | 0.2%  |
| 100.314             | incipient cataract, anterior sutures                           | 7                   | 0.0%  | 0                  |       |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.315 incipient cataract, posterior sutures             | 19               | 0.1%  | 3                | 0.1%  |
| 100.316 incipient cataract, nucleus                       | 186              | 1.0%  | 24               | 0.6%  |
| 100.317 incipient cataract, capsular                      | 22               | 0.1%  | 6                | 0.2%  |
| 100.321 incomplete cataract, anterior cortex              | 5                | 0.0%  | 5                | 0.1%  |
| 100.322 incomplete cataract, posterior cortex             | 4                | 0.0%  | 11               | 0.3%  |
| 100.323 incomplete cataract, equatorial cortex            | 2                | 0.0%  | 4                | 0.1%  |
| 100.325 incomplete cataract, posterior sutures            | 1                | 0.0%  | 0                |       |
| 100.326 incomplete cataract, nucleus                      | 12               | 0.1%  | 12               | 0.3%  |
| 100.327 incomplete cataract, capsular                     | 2                | 0.0%  | 1                | 0.0%  |
| 100.328 y-suture tip opacities                            | 2                | 0.0%  | 6                | 0.2%  |
| 100.330 generalized/complete cataract                     | 76               | 0.4%  | 4                | 0.1%  |
| 100.340 resorbing/hypermature cataract                    | 0                |       | 1                | 0.0%  |
| 100.345 <i>significant cataracts (summary)</i>            | 981              | 5.2%  | 183              | 4.8%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 6                | 0.0%  | 3                | 0.1%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 66               | 0.3%  | 22               | 0.6%  |
| 110.135 PHPV/PTVL   | 20               | 0.1%  | 4                | 0.1%  |
| 110.200 vitreous degeneration-anterior chamber            | 5                | 0.0%  | 0                |       |
| 110.320 vitreal degeneration                              | 85               | 0.4%  | 12               | 0.3%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 4                | 0.0%  | 1                | 0.0%  |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 1,150            | 6.0%  | 161              | 4.2%  |
| 120.180 retinal dysplasia, geographic                     | 170              | 0.9%  | 14               | 0.4%  |
| 120.190 retinal dysplasia, detached                       | 3                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 35               | 0.2%  | 1                | 0.0%  |
| 120.400 retinal hemorrhage                                | 7                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 3                | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 2                | 0.0%  | 4                | 0.1%  |
| 120.960 retinopathy                                       | 5                | 0.0%  | 5                | 0.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 6                | 0.0%  | 0                |       |
| 130.120 optic nerve hypoplasia                            | 9                | 0.0%  | 0                |       |
| 130.150 optic disc coloboma                               | 2                | 0.0%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 125              | 0.7%  | 0                |       |
| 900.100 other, not inherited                              | 313              | 1.6%  | 5                | 0.1%  |
| 900.110 other. suspect not inherited/significance unknown | 185              | 1.0%  | 118              | 3.1%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 13,655           | 71.8% | 2,489            | 65.3% |

# PERRO DE PRESA CANARIO

|    | DISORDER                                | INHERITANCE            | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|---|------------------------|-----------|-----------------|--------------------------------------|
| A. | Multifocal retinopathy<br>- <i>cmr1</i> | Autosomal<br>recessive | 1         | Breeder option  | Mutation in the<br><i>BEST1</i> gene |

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## Description and Comments

### A. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (*cmr1*) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene (*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff. A DNA test is available.

## References

1. Zangerl B, Wickstrom K, Slavik J, et al. Assessment of canine *BEST1* variations identifies new mutations and establishes an independent bestrophinopathy model (*cmr3*). *Mol Vis.* 2010;16:2791-2804.

# OCULAR DISORDERS REPORT PERRO DE PRESA CANARIO

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|--|---------------------|-----------|-------|-----------|-------|
|  |                     | #         | %     | #         | %     |
| <b>GLOBE</b>   |                     |           |       |           |       |
| 10.000 glaucoma  |                     | 0         |       | 1         | 20.0% |
| <b>LENS</b>  |                     |           |       |           |       |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 2         | 25.0% | 0         |       |
| 100.302 punctate cataract, posterior cortex                  |                     | 0         |       | 1         | 20.0% |
| 100.345 <i>significant cataracts (summary)</i>               |                     | 0         |       | 1         | 20.0% |
| <b>OTHER</b>   |                     |           |       |           |       |
| 900.100 other, not inherited                                 |                     | 0         |       | 1         | 20.0% |
| <b>NORMAL</b>  |                     |           |       |           |       |
| 0.000 normal globe   |                     | 6         | 75.0% | 4         | 80.0% |

# PERUVIAN INCA ORCHID

|    | <b>DISORDER</b> | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|-----------------|--------------------|------------------|------------------------|
| A. | Cataract        | Not defined        | 1                | NO                     |

---

## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Peruvian Inca Orchid breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT PERUVIAN INCA ORCHID

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015<br>21 |        | 2016-2020<br>51 |       |
|--|---------------------|-----------------|--------|-----------------|-------|
|  |                     | #               | %      | #               | %     |
| <b>GLOBE</b>   |                     |                 |        |                 |       |
| 0.110 microphthalmia   |                     | 1               | 4.8%   | 1               | 2.0%  |
| <b>UVEA</b>  |                     |                 |        |                 |       |
| 93.710 persistent pupillary membranes, iris to iris          |                     | 1               | 4.8%   | 1               | 2.0%  |
| <b>LENS</b>  |                     |                 |        |                 |       |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 1               | 4.8%   | 0               |       |
| 100.305 punctate cataract, posterior sutures                 |                     | 0               |        | 1               | 2.0%  |
| 100.306 punctate cataract, nucleus                           |                     | 0               |        | 1               | 2.0%  |
| 100.311 incipient cataract, anterior cortex                  |                     | 0               |        | 4               | 7.8%  |
| 100.312 incipient cataract, posterior cortex                 |                     | 0               |        | 1               | 2.0%  |
| 100.315 incipient cataract, posterior sutures                |                     | 0               |        | 1               | 2.0%  |
| 100.316 incipient cataract, nucleus                          |                     | 0               |        | 1               | 2.0%  |
| 100.321 incomplete cataract, anterior cortex                 |                     | 0               |        | 1               | 2.0%  |
| 100.322 incomplete cataract, posterior cortex                |                     | 0               |        | 1               | 2.0%  |
| 100.326 incomplete cataract, nucleus                         |                     | 0               |        | 1               | 2.0%  |
| 100.345 <i>significant cataracts (summary)</i>               |                     | 0               |        | 12              | 23.5% |
| <b>VITREOUS</b>  |                     |                 |        |                 |       |
| 110.120 persistent hyaloid artery/remnant                    |                     | 0               |        | 1               | 2.0%  |
| <b>RETINA</b>  |                     |                 |        |                 |       |
| 120.180 retinal dysplasia, geographic                        |                     | 0               |        | 2               | 3.9%  |
| 120.310 generalized progressive retinal atrophy (PRA)        |                     | 0               |        | 3               | 5.9%  |
| 120.960 retinopathy  |                     | 0               |        | 2               | 3.9%  |
| <b>OTHER</b>   |                     |                 |        |                 |       |
| 900.000 other, unspecified                                   |                     | 1               | 4.8%   | 0               |       |
| 900.100 other, not inherited                                 |                     | 0               |        | 1               | 2.0%  |
| 900.110 other. suspect not inherited/significance unknown    |                     | 0               |        | 1               | 2.0%  |
| <b>NORMAL</b>  |                     |                 |        |                 |       |
| 0.000 normal globe   |                     | 21              | 100.0% | 39              | 76.5% |



## PETIT BASSET GRIFFON VENDEEN

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE         | GENETIC TESTS<br>AVAILABLE              |
|----|------------------------------------|------------------------|-----------|----------------------------|---|
| A. | Glaucoma – POAG                    | Autosomal<br>recessive | 2, 3      | NO                         | Mutation in the<br><i>ADAMTS17</i> gene |
| B. | Corneal dystrophy<br>- endothelial | Not defined            | 1         | Breeder option             |   |
| C. | Persistent pupillary<br>membranes  |                        |           |                            |   |
|    | - iris to iris                     | Not defined            | 1         | Breeder option             |   |
|    | - lens pigment foci/<br>no strands | Not defined            | 1         | Passes with no<br>notation |   |
|    | - all other forms                  | Not defined            | 1         | NO                         |   |
| D. | Cataract                           | Not defined            | 1         | NO                         |   |
| E. | Retinal dysplasia<br>- folds       | Not defined            | 1         | Breeder option             |   |

### Description and Comments

#### A. Glaucoma

An elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine breed eye screening exam.

Primary Open Angle Glaucoma (POAG) in the Petit Basset Griffon Vendéen is caused by an inversion with a breakpoint disrupting the *ADAMTS17* gene. Pectinate ligament abnormalities are not present on gonioscopy and the iridocorneal angle remains open. The initial clinical features are noted around 3-4 years and include a small rise in intraocular pressure accompanied by lens subluxation. Retinal degeneration and optic nerve cupping noted in late stages when globe enlargement and vision disruption has occurred. A DNA test is available.

#### B. Corneal dystrophy - endothelial

Corneal endothelial dystrophy is an abnormal loss of the inner lining of the cornea that causes progressive fluid retention (edema). With time the edema results in keratitis and decreased vision. This usually does not occur until the animal is older.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee, 1999 and/or Data from OFA All-Breeds Report, 1991-1998.
2. Forman OP, Pettitt L, Komaromy AM, et al. A Novel Genome-Wide Association Study Approach Using Genotyping by Exome Sequencing Leads to the Identification of a Primary Open Angle Glaucoma Association Inversion Disrupting ADAMTS17; PLoS one, 2015: 10(12):e0143546.
3. Bedford, PGC (2017), Open-angle glaucoma in the Petit Basset Griffon Vendéen. Vet Ophthalmol, 20: 98-102. doi.10.1111/vop.12369.

# OCULAR DISORDERS REPORT

## PETIT BASSET GRIFFON VENDEEN

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>2,398 |       | 2016-2020<br>220 |       |
|---|---------------------|--------------------|-------|------------------|-------|
|   |                     | #                  | %     | #                | %     |
| <b>GLOBE</b>  |                     |                    |       |                  |       |
| 10.000 glaucoma   |                     | 3                  | 0.1%  | 1                | 0.5%  |
| <b>EYELIDS</b>  |                     |                    |       |                  |       |
| 21.000 entropion, unspecified   |                     | 3                  | 0.1%  | 0                |       |
| 25.110 distichiasis   |                     | 11                 | 0.5%  | 0                |       |
| <b>NICTITANS</b>  |                     |                    |       |                  |       |
| 52.110 prolapsed gland of the third eyelid                            |                     | 1                  | 0.0%  | 0                |       |
| <b>CORNEA</b>   |                     |                    |       |                  |       |
| 70.220 pigmentary keratitis   |                     | 1                  | 0.0%  | 0                |       |
| 70.700 corneal dystrophy  |                     | 17                 | 0.7%  | 0                |       |
| 70.730 corneal endothelial degeneration                               |                     | 26                 | 1.1%  | 0                |       |
| <b>UVEA</b>   |                     |                    |       |                  |       |
| 93.140 corneal endothelial pigment without PPM                        |                     | 2                  | 0.1%  | 0                |       |
| 93.150 iris coloboma  |                     | 1                  | 0.0%  | 0                |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 459                | 19.1% | 47               | 21.4% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 34                 | 1.4%  | 2                | 0.9%  |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 210                | 8.8%  | 14               | 6.4%  |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 15                 | 0.6%  | 0                |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 11                 | 0.5%  | 8                | 3.6%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 58                 | 2.4%  | 15               | 6.8%  |
| 93.999 uveal cysts  |                     | 4                  | 0.2%  | 0                |       |
| <b>LENS</b>   |                     |                    |       |                  |       |
| 100.200 cataract, unspecified   |                     | 2                  | 0.1%  | 0                |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 110                | 4.6%  | 5                | 2.3%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 24                 | 1.0%  | 5                | 2.3%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 5                  | 0.2%  | 0                |       |
| 100.303 punctate cataract, equatorial cortex                          |                     | 3                  | 0.1%  | 1                | 0.5%  |
| 100.304 punctate cataract, anterior sutures                           |                     | 4                  | 0.2%  | 0                |       |
| 100.305 punctate cataract, posterior sutures                          |                     | 5                  | 0.2%  | 2                | 0.9%  |
| 100.306 punctate cataract, nucleus                                    |                     | 2                  | 0.1%  | 0                |       |
| 100.307 punctate cataract, capsular                                   |                     | 12                 | 0.5%  | 5                | 2.3%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 23                 | 1.0%  | 3                | 1.4%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 7                  | 0.3%  | 0                |       |
| 100.313 incipient cataract, equatorial cortex                         |                     | 5                  | 0.2%  | 0                |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 6                  | 0.3%  | 0                |       |
| 100.316 incipient cataract, nucleus                                   |                     | 3                  | 0.1%  | 0                |       |
| 100.317 incipient cataract, capsular                                  |                     | 12                 | 0.5%  | 1                | 0.5%  |
| 100.326 incomplete cataract, nucleus                                  |                     | 0                  |       | 1                | 0.5%  |
| 100.328 y-suture tip opacities  |                     | 1                  | 0.0%  | 2                | 0.9%  |
| 100.330 generalized/complete cataract                                 |                     | 12                 | 0.5%  | 0                |       |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 126                | 5.3%  | 20               | 9.1%  |
| 100.375 <i>subluxation/luxation, unspecified</i>                      |                     | 8                  | 0.3%  | 2                | 0.9%  |

|   | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 12 0.5%          | 1 0.5%           |
| 110.320 vitreal degeneration                              | 13 0.5%          | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 108 4.5%         | 6 2.7%           |
| 120.180 retinal dysplasia, geographic                     | 11 0.5%          | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 3 0.1%           | 0                |
| 120.400 retinal hemorrhage                                | 2 0.1%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 3 0.1%           | 1 0.5%           |
| 130.150 optic disc coloboma                               | 1 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 38 1.6%          | 0                |
| 900.100 other, not inherited                              | 76 3.2%          | 5 2.3%           |
| 900.110 other. suspect not inherited/significance unknown | 46 1.9%          | 4 1.8%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,538 64.1%      | 135 61.4%        |

# PHARAOH HOUND

|    | DISORDER   | INHERITANCE                | REFERENCE | BREEDING ADVICE                           |
|----|--|----------------------------|-----------|---|
| A. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined | 1<br>1    | Breeder option<br>Passes with no notation |
| B. | Cataract   | Not defined                | 1         | NO  |

---

## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Pharaoh Hound breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT PHARAOH HOUND

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015 |      | 2016-2020 |      |
|------------------|--|-----------|------|-----------|------|
|                  |  | #         | %    | #         | %    |
| <b>EYELIDS</b>   |  |           |      |           |      |
| 25.110           | distichiasis   | 7         | 1.9% | 0         |      |
| <b>NICTITANS</b> |  |           |      |           |      |
| 52.110           | prolapsed gland of the third eyelid                            | 1         | 0.3% | 0         |      |
| <b>CORNEA</b>    |  |           |      |           |      |
| 70.700           | corneal dystrophy  | 3         | 0.8% | 1         | 0.7% |
| <b>UVEA</b>      |  |           |      |           |      |
| 93.140           | corneal endothelial pigment without PPM                        | 1         | 0.3% | 0         |      |
| 93.710           | persistent pupillary membranes, iris to iris                   | 28        | 7.6% | 12        | 8.5% |
| 93.720           | persistent pupillary membranes, iris to lens                   | 1         | 0.3% | 0         |      |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands   | 11        | 3.0% | 3         | 2.1% |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands | 1         | 0.3% | 0         |      |
| 93.999           | uveal cysts  | 1         | 0.3% | 0         |      |
| <b>LENS</b>      |  |           |      |           |      |
| 100.200          | cataract, unspecified  | 1         | 0.3% | 0         |      |
| 100.210          | cataract. suspect not inherited/significance unknown           | 21        | 5.7% | 9         | 6.3% |
| 100.301          | punctate cataract, anterior cortex                             | 2         | 0.5% | 1         | 0.7% |
| 100.302          | punctate cataract, posterior cortex                            | 1         | 0.3% | 0         |      |
| 100.305          | punctate cataract, posterior sutures                           | 2         | 0.5% | 0         |      |
| 100.306          | punctate cataract, nucleus                                     | 1         | 0.3% | 0         |      |
| 100.307          | punctate cataract, capsular                                    | 1         | 0.3% | 0         |      |
| 100.311          | incipient cataract, anterior cortex                            | 1         | 0.3% | 0         |      |
| 100.312          | incipient cataract, posterior cortex                           | 2         | 0.5% | 1         | 0.7% |
| 100.313          | incipient cataract, equatorial cortex                          | 2         | 0.5% | 0         |      |
| 100.315          | incipient cataract, posterior sutures                          | 3         | 0.8% | 1         | 0.7% |
| 100.316          | incipient cataract, nucleus                                    | 1         | 0.3% | 0         |      |
| 100.328          | y-suture tip opacities   | 1         | 0.3% | 0         |      |
| 100.330          | generalized/complete cataract                                  | 1         | 0.3% | 0         |      |
| 100.345          | <i>significant cataracts (summary)</i>                         | 19        | 5.1% | 3         | 2.1% |
| <b>VITREOUS</b>  |  |           |      |           |      |
| 110.320          | vitreal degeneration   | 0         |      | 1         | 0.7% |
| <b>RETINA</b>    |  |           |      |           |      |
| 120.170          | retinal dysplasia, folds                                       | 3         | 0.8% | 1         | 0.7% |
| 120.180          | retinal dysplasia, geographic                                  | 2         | 0.5% | 0         |      |
| 120.310          | generalized progressive retinal atrophy (PRA)                  | 3         | 0.8% | 0         |      |
| 120.960          | retinopathy  | 0         |      | 3         | 2.1% |
| <b>OTHER</b>     |  |           |      |           |      |
| 900.000          | other, unspecified   | 4         | 1.1% | 0         |      |
| 900.100          | other, not inherited   | 7         | 1.9% | 0         |      |
| 900.110          | other. suspect not inherited/significance unknown              | 3         | 0.8% | 2         | 1.4% |

|                                     | 1991-2015 | 2016-2020 |
|-------------------------------------|-----------|-----------|
| <b>NORMAL</b><br>0.000 normal globe | 303 81.9% | 107 75.4% |

# PLOTT

|    | DISORDER                        | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE          |
|----|---------------------------------|---------------------|-----------|-----------------|----------------------------------|
| A. | Retinal atrophy ( <i>prcd</i> ) | Autosomal recessive | 1, 2      | NO              | Mutation in the <i>prcd</i> gene |

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## Description and Comments

### A. Retinal atrophy – *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Plott is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Plott. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Zangerl, B., et al. (2006). "Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans." Genomics **88**(5): 551-563. PMID: 16938425



# OCULAR DISORDERS REPORT PLOTT

| Diagnostic Name                     | TOTAL DOGS EXAMINED | 1991-2015 |   | 2016-2020 |        |
|-------------------------------------|---------------------|-----------|---|-----------|--------|
|                                     |                     | #         | % | #         | %      |
| <b>NORMAL</b><br>0.000 normal globe |                     | 0         |   | 9         | 100.0% |

# POINTER

|    | DISORDER                                  | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|---|-------------|-----------|-----------------|
| A. | Corneal dystrophy<br>- epithelial/stromal | Not defined | 1         | Breeder option  |
| B. | Cataract                                  | Not defined | 1         | NO              |

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## Description and Comments

### A. Corneal Dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Pointer breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT POINTER

| Diagnostic Name    | TOTAL DOGS EXAMINED  | 1991-2015<br>686 |      | 2016-2020<br>146 |      |
|--------------------|--|------------------|------|------------------|------|
|                    |  | #                | %    | #                | %    |
| <b>EYELIDS</b>     |  |                  |      |                  |      |
| 21.000             | entropion, unspecified   | 5                | 0.7% | 0                |      |
| 22.000             | ectropion, unspecified   | 1                | 0.1% | 0                |      |
| 25.110             | distichiasis   | 4                | 0.6% | 0                |      |
| <b>NICTITANS</b>   |  |                  |      |                  |      |
| 52.110             | prolapsed gland of the third eyelid                            | 1                | 0.1% | 0                |      |
| <b>CORNEA</b>      |  |                  |      |                  |      |
| 70.700             | corneal dystrophy  | 6                | 0.9% | 6                | 4.1% |
| <b>UVEA</b>        |  |                  |      |                  |      |
| 93.710             | persistent pupillary membranes, iris to iris                   | 11               | 1.6% | 1                | 0.7% |
| 93.720             | persistent pupillary membranes, iris to lens                   | 1                | 0.1% | 0                |      |
| 93.730             | persistent pupillary membranes, iris to cornea                 | 1                | 0.1% | 0                |      |
| 93.760             | persistent pupillary membranes, endothelial opacity/no strands | 0                |      | 1                | 0.7% |
| <b>LENS</b>        |  |                  |      |                  |      |
| 100.210            | cataract. suspect not inherited/significance unknown           | 18               | 2.6% | 3                | 2.1% |
| 100.302            | punctate cataract, posterior cortex                            | 1                | 0.1% | 0                |      |
| 100.303            | punctate cataract, equatorial cortex                           | 1                | 0.1% | 0                |      |
| 100.305            | punctate cataract, posterior sutures                           | 0                |      | 1                | 0.7% |
| 100.306            | punctate cataract, nucleus                                     | 1                | 0.1% | 1                | 0.7% |
| 100.312            | incipient cataract, posterior cortex                           | 3                | 0.4% | 0                |      |
| 100.313            | incipient cataract, equatorial cortex                          | 1                | 0.1% | 0                |      |
| 100.315            | incipient cataract, posterior sutures                          | 1                | 0.1% | 0                |      |
| 100.317            | incipient cataract, capsular                                   | 0                |      | 1                | 0.7% |
| 100.321            | incomplete cataract, anterior cortex                           | 0                |      | 1                | 0.7% |
| 100.322            | incomplete cataract, posterior cortex                          | 0                |      | 2                | 1.4% |
| 100.326            | incomplete cataract, nucleus                                   | 0                |      | 2                | 1.4% |
| 100.345            | <i>significant cataracts (summary)</i>                         | 8                | 1.2% | 8                | 5.5% |
| <b>VITREOUS</b>    |  |                  |      |                  |      |
| 110.120            | persistent hyaloid artery/remnant                              | 1                | 0.1% | 0                |      |
| <b>RETINA</b>      |  |                  |      |                  |      |
| 120.170            | retinal dysplasia, folds                                       | 7                | 1.0% | 0                |      |
| 120.180            | retinal dysplasia, geographic                                  | 3                | 0.4% | 1                | 0.7% |
| 120.310            | generalized progressive retinal atrophy (PRA)                  | 2                | 0.3% | 0                |      |
| <b>OPTIC NERVE</b> |  |                  |      |                  |      |
| 130.110            | micropapilla   | 4                | 0.6% | 1                | 0.7% |
| 130.120            | optic nerve hypoplasia   | 1                | 0.1% | 0                |      |
| <b>OTHER</b>       |  |                  |      |                  |      |
| 900.000            | other, unspecified   | 7                | 1.0% | 0                |      |
| 900.100            | other, not inherited   | 6                | 0.9% | 0                |      |
| 900.110            | other. suspect not inherited/significance unknown              | 9                | 1.3% | 10               | 6.8% |

|                                     | 1991-2015 | 2016-2020 |
|-------------------------------------|-----------|-----------|
| <b>NORMAL</b><br>0.000 normal globe | 620 90.4% | 122 83.6% |

# POLISH LOWLAND SHEEPDOG

## (Polski Owczarek Nizinny)

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE                                   |
|----|--|---------------------|-----------|-----------------|---|
| A. | Corneal dystrophy<br>- epithelial/stromal                      | Not defined         | 1         | Breeder option  |   |
| B. | Persistent pupillary membranes<br>- iris to iris               | Not defined         | 1         | Breeder option  |   |
| C. | Cataract   | Not defined         | 1         | NO              |   |
| D. | Retinal atrophy<br>- rod-cone dysplasia type 1 ( <i>rcd4</i> ) | Autosomal recessive | 2         | NO              | Mutation in the <i>C2orf71</i> or <i>C17H2orf71</i> genes |
| E. | Ceroid lipofuscinosis  | Not defined         | 3         | NO              |   |

### Description and Comments

**A. Corneal Dystrophy - epithelial/stromal**

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

**B. Persistent pupillary membranes (PPMs)**

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

**C. Cataract**

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Rod-cone dysplasia, type 4 (*rcd4*)

A form of PRA identified in the Gordon and Irish Setter breeds. Clinical night blindness is observed on average as late as 10 years of age and progresses to total blindness. This form of PRA has been referred to as late-onset PRA (LOPRA). The disorder is caused by a mutation present in the *C2orf71* gene. A DNA test is available.

A form of PRA, similar to that found in Gordon and Irish setters, has also been found in the the Polish Lowland Sheepdog. This form of PRA has been referred to as late-onset, slowly progressive PRA (LOPRA). Slight vascular attenuation, first seen between 4.5 -6 years of age precedes tapetal hyperreflectivity. All fundic changes were bilaterally symmetric and progressed slowly eventually causing clinical blindness, bilateral complete vascular attenuation, and tapetal hyperreflectivity by 12 years of age, on average. Almost all affected dogs were homozygous for the *rcd4* mutation in *C17H2orf71* gene. A DNA test is available.

E. Ceroid lipofuscinosis

A systemic metabolic disorder that affects the retina and retinal pigment epithelium with accumulation of lipopigments resulting in retinal degeneration.

**Historical Note:**

Central progressive retinal atrophy was previously a condition listed for this breed. However as the condition is no longer identified in the breed, the condition has been removed. Central progressive retinal atrophy was a progressive retinal degeneration in which photoreceptor death occurred secondary to disease of the underlying pigment epithelium. Progression was slow and some animals never lost vision. CPRA occurred in England, but was uncommon elsewhere.

**References**

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Downs LM, Bell JS, Freeman J, et al. Late-onset progressive retinal atrophy in the Gordon and Irish Setter breeds is associated with a frameshift mutation in *C2orf71*. *Anim Genet.* 2012;44:169-177.
3. Narfstrom K, Wrigstad A, Ekesten B, et al. Neuronal ceroid lipofuscinosis: clinical and morphologic findings in nine affected Polish Owczarek Nizinny (PON) dogs. *Vet Ophthalmol.* 2007;10:111-120.

# OCULAR DISORDERS REPORT POLISH LOWLAND SHEEPDOG

| Diagnostic Name     | TOTAL DOGS EXAMINED                                  | 1991-2015 |       | 2016-2020 |       |
|---------------------|--|-----------|-------|-----------|-------|
|                     |  | #         | %     | #         | %     |
| <b>EYELIDS</b>      |  |           |       |           |       |
| 25.110              | distichiasis   | 17        | 1.6%  | 4         | 2.2%  |
| <b>NASOLACRIMAL</b> |  |           |       |           |       |
| 32.110              | imperforate lower nasolacrimal punctum               | 1         | 0.1%  | 1         | 0.6%  |
| <b>CORNEA</b>       |  |           |       |           |       |
| 70.700              | corneal dystrophy                                    | 30        | 2.8%  | 6         | 3.4%  |
| 70.730              | corneal endothelial degeneration                     | 1         | 0.1%  | 0         |       |
| <b>UVEA</b>         |  |           |       |           |       |
| 93.710              | persistent pupillary membranes, iris to iris         | 70        | 6.6%  | 20        | 11.2% |
| 93.999              | uveal cysts  | 2         | 0.2%  | 0         |       |
| <b>LENS</b>         |  |           |       |           |       |
| 100.210             | cataract. suspect not inherited/significance unknown | 46        | 4.3%  | 7         | 3.9%  |
| 100.301             | punctate cataract, anterior cortex                   | 5         | 0.5%  | 7         | 3.9%  |
| 100.302             | punctate cataract, posterior cortex                  | 8         | 0.7%  | 2         | 1.1%  |
| 100.303             | punctate cataract, equatorial cortex                 | 1         | 0.1%  | 0         |       |
| 100.305             | punctate cataract, posterior sutures                 | 1         | 0.1%  | 0         |       |
| 100.306             | punctate cataract, nucleus                           | 0         |       | 1         | 0.6%  |
| 100.307             | punctate cataract, capsular                          | 1         | 0.1%  | 1         | 0.6%  |
| 100.311             | incipient cataract, anterior cortex                  | 4         | 0.4%  | 1         | 0.6%  |
| 100.312             | incipient cataract, posterior cortex                 | 3         | 0.3%  | 0         |       |
| 100.313             | incipient cataract, equatorial cortex                | 1         | 0.1%  | 2         | 1.1%  |
| 100.315             | incipient cataract, posterior sutures                | 3         | 0.3%  | 0         |       |
| 100.316             | incipient cataract, nucleus                          | 1         | 0.1%  | 1         | 0.6%  |
| 100.317             | incipient cataract, capsular                         | 2         | 0.2%  | 0         |       |
| 100.321             | incomplete cataract, anterior cortex                 | 0         |       | 2         | 1.1%  |
| 100.330             | generalized/complete cataract                        | 1         | 0.1%  | 0         |       |
| 100.345             | <i>significant cataracts (summary)</i>               | 31        | 2.9%  | 17        | 9.5%  |
| <b>VITREOUS</b>     |  |           |       |           |       |
| 110.120             | persistent hyaloid artery/remnant                    | 1         | 0.1%  | 0         |       |
| 110.320             | vitreal degeneration                                 | 2         | 0.2%  | 0         |       |
| <b>RETINA</b>       |  |           |       |           |       |
| 120.170             | retinal dysplasia, folds                             | 10        | 0.9%  | 0         |       |
| 120.310             | generalized progressive retinal atrophy (PRA)        | 16        | 1.5%  | 5         | 2.8%  |
| 120.960             | retinopathy  | 1         | 0.1%  | 0         |       |
| <b>OTHER</b>        |  |           |       |           |       |
| 900.000             | other, unspecified                                   | 5         | 0.5%  | 0         |       |
| 900.100             | other, not inherited                                 | 24        | 2.2%  | 1         | 0.6%  |
| 900.110             | other. suspect not inherited/significance unknown    | 0         |       | 7         | 3.9%  |
| <b>NORMAL</b>       |  |           |       |           |       |
| 0.000               | normal globe   | 905       | 84.8% | 121       | 67.6% |

# POMERANIAN

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|---|------------------------|-----------|-----------------|--------------------------------------|
| A. | Distichiasis  | Not defined            | 1         | Breeder option  |                                      |
| B. | Entropion   | Not defined            | 1         | Breeder option  |                                      |
| C. | Persistent pupillary membranes<br>- iris to iris                  | Not defined            | 1         | Breeder option  |                                      |
| D. | Cataract  | Not defined            | 1         | NO              |                                      |
| E. | Retinal atrophy<br>- rod-cone dysplasia<br>type 3 ( <i>rca3</i> ) | Autosomal<br>recessive | 1         | NO              | Mutation in the<br><i>PDE6A</i> gene |
| F. | Retinal atrophy ( <i>prca</i> )                                   | Autosomal<br>recessive | 1         | NO              | Mutation in the<br><i>prca</i> gene  |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. Selection should be directed against entropion and toward head conformation that minimizes or eliminates the likelihood of the defect.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or from sheets of tissue in the anterior chamber. The last three forms pose the greatest



threat to vision and when severe, vision impairment or blindness may occur.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Retinal atrophy - rod-cone dysplasia type 3 (*rcd3*)

PRA is an autosomal recessive trait caused by a one base pair deletion in the gene encoding the alpha subunit of cyclic GMP phosphodiesterase (*rcd3*). PRA begins early in life with clinical signs of night blindness and a lack of rod ERG responses is seen at 6-8 weeks of age. Dogs are completely blind by 2-3 years of age when ophthalmoscopic signs are first visible. The mutation is found in the *PDE6A* gene. A DNA test is available.

F. Retinal atrophy – *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Pomeranian is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Pomeranian breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT POMERANIAN

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>1,092 |      | 2016-2020<br>738 |      |
|---|---------------------|--------------------|------|------------------|------|
|   |                     | #                  | %    | #                | %    |
| <b>GLOBE</b>  |                     |                    |      |                  |      |
| 0.110 microphthalmia  |                     | 2                  | 0.2% | 4                | 0.5% |
| <b>EYELIDS</b>  |                     |                    |      |                  |      |
| 20.140 ectopic cilia  |                     | 1                  | 0.1% | 0                |      |
| 21.000 entropion, unspecified   |                     | 3                  | 0.3% | 23               | 3.1% |
| 22.000 ectropion, unspecified   |                     | 1                  | 0.1% | 0                |      |
| 25.110 distichiasis   |                     | 52                 | 4.8% | 21               | 2.8% |
| <b>NASOLACRIMAL</b>   |                     |                    |      |                  |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 1                  | 0.1% | 1                | 0.1% |
| 40.910 keratoconjunctivitis sicca                                     |                     | 1                  | 0.1% | 0                |      |
| <b>CORNEA</b>   |                     |                    |      |                  |      |
| 70.210 corneal pannus   |                     | 1                  | 0.1% | 0                |      |
| 70.220 pigmentary keratitis   |                     | 2                  | 0.2% | 0                |      |
| 70.700 corneal dystrophy  |                     | 3                  | 0.3% | 1                | 0.1% |
| 70.730 corneal endothelial degeneration                               |                     | 2                  | 0.2% | 0                |      |
| <b>UVEA</b>   |                     |                    |      |                  |      |
| 93.110 iris hypoplasia  |                     | 0                  |      | 1                | 0.1% |
| 93.150 iris coloboma  |                     | 0                  |      | 2                | 0.3% |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 62                 | 5.7% | 71               | 9.6% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 3                  | 0.3% | 1                | 0.1% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 4                  | 0.4% | 1                | 0.1% |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 5                  | 0.5% | 6                | 0.8% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1                  | 0.1% | 3                | 0.4% |
| 93.810 uveal melanoma   |                     | 1                  | 0.1% | 0                |      |
| 97.150 chorioretinal coloboma, congenital                             |                     | 0                  |      | 1                | 0.1% |
| <b>LENS</b>   |                     |                    |      |                  |      |
| 100.200 cataract, unspecified   |                     | 1                  | 0.1% | 0                |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 28                 | 2.6% | 8                | 1.1% |
| 100.301 punctate cataract, anterior cortex                            |                     | 2                  | 0.2% | 2                | 0.3% |
| 100.302 punctate cataract, posterior cortex                           |                     | 2                  | 0.2% | 2                | 0.3% |
| 100.303 punctate cataract, equatorial cortex                          |                     | 1                  | 0.1% | 0                |      |
| 100.304 punctate cataract, anterior sutures                           |                     | 1                  | 0.1% | 0                |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 3                  | 0.3% | 0                |      |
| 100.306 punctate cataract, nucleus                                    |                     | 1                  | 0.1% | 0                |      |
| 100.307 punctate cataract, capsular                                   |                     | 1                  | 0.1% | 1                | 0.1% |
| 100.311 incipient cataract, anterior cortex                           |                     | 9                  | 0.8% | 3                | 0.4% |
| 100.312 incipient cataract, posterior cortex                          |                     | 6                  | 0.5% | 2                | 0.3% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 3                  | 0.3% | 2                | 0.3% |
| 100.315 incipient cataract, posterior sutures                         |                     | 0                  |      | 1                | 0.1% |
| 100.316 incipient cataract, nucleus                                   |                     | 2                  | 0.2% | 1                | 0.1% |
| 100.317 incipient cataract, capsular                                  |                     | 0                  |      | 3                | 0.4% |
| 100.322 incomplete cataract, posterior cortex                         |                     | 0                  |      | 1                | 0.1% |
| 100.330 generalized/complete cataract                                 |                     | 11                 | 1.0% | 1                | 0.1% |
| 100.340 resorbing/hypermature cataract                                |                     | 1                  | 0.1% | 0                |      |
| 100.345 significant cataracts (summary)                               |                     | 44                 | 4.0% | 19               | 2.6% |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>VITREOUS</b>   |           |           |
| 110.120 persistent hyaloid artery/remnant                 | 3 0.3%    | 1 0.1%    |
| 110.135 PHPV/PTVL   | 1 0.1%    | 0         |
| 110.200 vitreous degeneration-anterior chamber            | 3 0.3%    | 3 0.4%    |
| 110.320 vitreal degeneration                              | 14 1.3%   | 3 0.4%    |
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 2 0.2%    | 5 0.7%    |
| 120.180 retinal dysplasia, geographic                     | 3 0.3%    | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 16 1.5%   | 1 0.1%    |
| 120.400 retinal hemorrhage                                | 1 0.1%    | 0         |
| 120.910 retinal detachment without dialysis               | 2 0.2%    | 0         |
| 120.920 retinal detachment with dialysis                  | 0         | 1 0.1%    |
| 120.960 retinopathy                                       | 0         | 2 0.3%    |
| <b>OPTIC NERVE</b>  |           |           |
| 130.120 optic nerve hypoplasia                            | 2 0.2%    | 0         |
| 130.150 optic disc coloboma                               | 2 0.2%    | 1 0.1%    |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 10 0.9%   | 0         |
| 900.100 other, not inherited                              | 27 2.5%   | 2 0.3%    |
| 900.110 other. suspect not inherited/significance unknown | 11 1.0%   | 21 2.8%   |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 902 82.6% | 563 76.3% |

# POODLE

## (Toy, Miniature, and Standard varieties)

\* All varieties of the Poodle are basically the same genetic makeup, having their size governed by differences in an "insulin-like growth factor." (See Reference 2.)

|    | DISORDER   | INHERITANCE            | REFERENCE | BREEDING ADVICE            | GENETIC TESTS AVAILABLE  |
|----|--|------------------------|-----------|----------------------------|--|
| A. | Distichiasis   | Not defined            | 1         | Breeder option             |  |
| B. | Corneal dystrophy<br>- epithelial/stromal  | Not defined            | 1         | Breeder option             |  |
| C. | Persistent pupillary<br>membranes<br>- iris to iris<br>- lens pigment foci/no<br>strands | Not defined            | 1         | Breeder option             |  |
|    |  | Not defined            | 1         | Passes with no<br>notation |  |
| D. | Cataract   | Not defined            | 1, 2-4    | NO                         |  |
| E. | Y-suture tip opacity   | Not defined            | 1         | Breeder option             |  |
| F. | Vitreous degeneration  | Not defined            | 1         | Breeder option             |  |
| G. | Retinal atrophy<br>( <i>prcd</i> )   | Autosomal<br>recessive | 1, 10-20  | NO                         | Mutation in the<br><i>prcd</i> gene  |
| H. | Retinal atrophy<br>- rod-cone dysplasia<br>type 4 ( <i>rcd4</i> )                        | Autosomal<br>recessive | 1         | NO                         | Mutation in the<br><i>C2orf71</i> gene<br>*only in Standards<br>& Miniatures |
| I. | Cone degeneration<br>(achromatopsia)   | Autosomal<br>recessive | 4, 21     | NO                         | Mutation has not<br>been published<br>*only in Standards                     |
| J. | Micropapilla   | Not defined            | 1         | Breeder option             |  |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

B. Corneal Dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

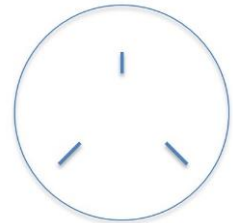
D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

The Poodle cataract can involve the lens cortex and lens nucleus. The rate and degree of progression are variable. A familial form of cataract has been described in the Standard Poodle, beginning with an equatorial opacity initially observed in dogs prior to 2 years of age.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

F. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment and/or glaucoma. Either condition may cause blindness.

G. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that PRA in the Poodle is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available. It is important to note that in all breeds in which a molecular diagnostic test for the disease is available, it is possible to have dogs diagnosed clinically as affected, yet the DNA test results are normal. This suggests that other genetic causes of PRA exist or that the diagnosed affected dog has an acquired disease that mimics the inherited disorder.

H. Rod-cone dysplasia, type 4 (*rcd4*)

A form of PRA identified in the Gordon and Irish Setter breeds. Clinical night blindness is observed on average as late as 10 years of age and progresses to total blindness. This form of PRA has been referred to as late-onset PRA (LOPRA). The disorder is caused by a mutation present in the *C2orf71* gene. A DNA test is now available that will unequivocally identify genetically normal, affected and carrier dogs. The test is accurate only for this mutation and will not identify other forms of PRA.

I. Cone degeneration: Day Blindness/Retinal degeneration:

An autosomal recessive disorder of standard poodles and 'Doodles' (where the mix-bred dogs are backcrossed to standard poodles that carry the genetic defect); the disease also has been referred to as achromatopsia. The salient clinical findings is profound visual difficulty in bright light, day blindness, with subjective normal night vision. In the early stages of the disease, fundus examination is normal with some dogs showing focal hyperreflectivity of the cone-rich fovea like region of the retina; the photopic ERG is not recordable. In some older dogs, there is progression resulting in poor/absent vision under both dim and bright light conditions, markedly abnormal or non-recordable ERG, and a fundus appearance indicative of late stage retinal degeneration and indistinguishable from progressive retinal atrophy.

J. Micropapilla

Micropapilla refers to a small optic disc which is not associated with vision impairment. Optic nerve hypoplasia refers to a congenital defect of the optic nerve which causes blindness and abnormal pupil response in the affected eye. May be difficult to differentiate between micropapilla and optic nerve hypoplasia on a routine (dilated) screening ophthalmoscopic exam.

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# OCULAR DISORDERS REPORT POODLE

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>47,985 |      | 2016-2020<br>10,591 |      |
|---------------------|--|---------------------|------|---------------------|------|
|                     |  | #                   | %    | #                   | %    |
| <b>GLOBE</b>        |  |                     |      |                     |      |
| 0.110               | microphthalmia   | 21                  | 0.0% | 6                   | 0.1% |
| 10.000              | glaucoma   | 6                   | 0.0% | 0                   |      |
| <b>EYELIDS</b>      |  |                     |      |                     |      |
| 20.110              | eyelid dermoid   | 1                   | 0.0% | 0                   |      |
| 20.140              | ectopic cilia  | 34                  | 0.1% | 10                  | 0.1% |
| 20.160              | macropalpebral fissure   | 1                   | 0.0% | 0                   |      |
| 21.000              | entropion, unspecified   | 122                 | 0.3% | 26                  | 0.2% |
| 22.000              | ectropion, unspecified   | 5                   | 0.0% | 2                   | 0.0% |
| 25.110              | distichiasis   | 2,967               | 6.2% | 566                 | 5.3% |
| <b>NASOLACRIMAL</b> |  |                     |      |                     |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 8                   | 0.0% | 11                  | 0.1% |
| 40.910              | keratoconjunctivitis sicca                                     | 10                  | 0.0% | 3                   | 0.0% |
| <b>NICTITANS</b>    |  |                     |      |                     |      |
| 50.210              | pannus of third eyelid   | 1                   | 0.0% | 0                   |      |
| 51.100              | third eyelid cartilage anomaly                                 | 40                  | 0.1% | 5                   | 0.0% |
| 52.110              | prolapsed gland of the third eyelid                            | 18                  | 0.0% | 1                   | 0.0% |
| <b>CORNEA</b>       |  |                     |      |                     |      |
| 70.210              | corneal pannus   | 39                  | 0.1% | 0                   |      |
| 70.220              | pigmentary keratitis   | 27                  | 0.1% | 8                   | 0.1% |
| 70.700              | corneal dystrophy  | 263                 | 0.5% | 57                  | 0.5% |
| 70.730              | corneal endothelial degeneration                               | 12                  | 0.0% | 0                   |      |
| <b>UVEA</b>         |  |                     |      |                     |      |
| 90.250              | pigmentary uveitis   | 2                   | 0.0% | 1                   | 0.0% |
| 93.110              | iris hypoplasia  | 1                   | 0.0% | 3                   | 0.0% |
| 93.140              | corneal endothelial pigment without PPM                        | 5                   | 0.0% | 0                   |      |
| 93.150              | iris coloboma  | 6                   | 0.0% | 0                   |      |
| 93.180              | liris sphincter dysplasia                                      | 0                   |      | 1                   | 0.0% |
| 93.710              | persistent pupillary membranes, iris to iris                   | 1,483               | 3.1% | 590                 | 5.6% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 83                  | 0.2% | 22                  | 0.2% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 33                  | 0.1% | 5                   | 0.0% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 38                  | 0.1% | 1                   | 0.0% |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 167                 | 0.3% | 215                 | 2.0% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 4                   | 0.0% | 6                   | 0.1% |
| 93.810              | uveal melanoma   | 4                   | 0.0% | 0                   |      |
| 93.999              | uveal cysts  | 8                   | 0.0% | 4                   | 0.0% |
| 97.150              | chorioretinal coloboma, congenital                             | 1                   | 0.0% | 1                   | 0.0% |
| <b>LENS</b>         |  |                     |      |                     |      |
| 100.110             | microphakia, congenital  | 0                   |      | 1                   | 0.0% |
| 100.200             | cataract, unspecified  | 384                 | 0.8% | 0                   |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 2,576               | 5.4% | 450                 | 4.2% |
| 100.301             | punctate cataract, anterior cortex                             | 441                 | 0.9% | 69                  | 0.7% |
| 100.302             | punctate cataract, posterior cortex                            | 193                 | 0.4% | 27                  | 0.3% |
| 100.303             | punctate cataract, equatorial cortex                           | 120                 | 0.3% | 21                  | 0.2% |

| <b>LENS CONTINUED</b>                                 | <b>1991-2015</b> |      | <b>2016-2020</b> |      |
|---|------------------|------|------------------|------|
| 100.304 punctate cataract, anterior sutures           | 53               | 0.1% | 12               | 0.1% |
| 100.305 punctate cataract, posterior sutures          | 119              | 0.2% | 43               | 0.4% |
| 100.306 punctate cataract, nucleus                    | 38               | 0.1% | 18               | 0.2% |
| 100.307 punctate cataract, capsular                   | 41               | 0.1% | 42               | 0.4% |
| 100.311 incipient cataract, anterior cortex           | 482              | 1.0% | 57               | 0.5% |
| 100.312 incipient cataract, posterior cortex          | 400              | 0.8% | 49               | 0.5% |
| 100.313 incipient cataract, equatorial cortex         | 258              | 0.5% | 32               | 0.3% |
| 100.314 incipient cataract, anterior sutures          | 37               | 0.1% | 3                | 0.0% |
| 100.315 incipient cataract, posterior sutures         | 89               | 0.2% | 16               | 0.2% |
| 100.316 incipient cataract, nucleus                   | 65               | 0.1% | 17               | 0.2% |
| 100.317 incipient cataract, capsular                  | 39               | 0.1% | 10               | 0.1% |
| 100.320 incomplete cataract, unspecified              | 0                |      | 2                | 0.0% |
| 100.321 incomplete cataract, anterior cortex          | 9                | 0.0% | 20               | 0.2% |
| 100.322 incomplete cataract, posterior cortex         | 14               | 0.0% | 15               | 0.1% |
| 100.323 incomplete cataract, equatorial cortex        | 7                | 0.0% | 10               | 0.1% |
| 100.324 incomplete cataract, anterior sutures         | 0                |      | 1                | 0.0% |
| 100.325 incomplete cataract, posterior sutures        | 0                |      | 2                | 0.0% |
| 100.326 incomplete cataract, nucleus                  | 3                | 0.0% | 5                | 0.0% |
| 100.327 incomplete cataract, capsular                 | 0                |      | 2                | 0.0% |
| 100.328 y-suture tip opacities                        | 18               | 0.0% | 60               | 0.6% |
| 100.330 generalized/complete cataract                 | 429              | 0.9% | 11               | 0.1% |
| 100.340 resorbing/hypermature cataract                | 2                | 0.0% | 4                | 0.0% |
| 100.345 <i>significant cataracts (summary)</i>        | 3,241            | 6.8% | 548              | 5.2% |
| 100.375 <i>subluxation/luxation, unspecified</i>      | 29               | 0.1% | 0                |      |
| <b>VITREOUS</b>                                       |                  |      |                  |      |
| 110.120 persistent hyaloid artery/remnant             | 77               | 0.2% | 40               | 0.4% |
| 110.135 PHPV/PTVL                                     | 24               | 0.1% | 9                | 0.1% |
| 110.200 vitreous degeneration-anterior chamber        | 11               | 0.0% | 10               | 0.1% |
| 110.320 vitreal degeneration                          | 312              | 0.7% | 53               | 0.5% |
| <b>FUNDUS</b>   |                  |      |                  |      |
| 97.110 choroidal hypoplasia                           | 3                | 0.0% | 0                |      |
| 97.120 coloboma                                       | 11               | 0.0% | 0                |      |
| <b>RETINA</b>   |                  |      |                  |      |
| 120.170 retinal dysplasia, folds                      | 127              | 0.3% | 33               | 0.3% |
| 120.180 retinal dysplasia, geographic                 | 21               | 0.0% | 2                | 0.0% |
| 120.190 retinal dysplasia, detached                   | 9                | 0.0% | 1                | 0.0% |
| 120.310 generalized progressive retinal atrophy (PRA) | 582              | 1.2% | 16               | 0.2% |
| 120.400 retinal hemorrhage                            | 3                | 0.0% | 0                |      |
| 120.910 retinal detachment without dialysis           | 27               | 0.1% | 0                |      |
| 120.920 retinal detachment with dialysis              | 1                | 0.0% | 4                | 0.0% |
| 120.960 retinopathy                                   | 12               | 0.0% | 12               | 0.1% |
| <b>OPTIC NERVE</b>                                    |                  |      |                  |      |
| 130.110 micropapilla                                  | 161              | 0.3% | 51               | 0.5% |
| 130.120 optic nerve hypoplasia                        | 210              | 0.4% | 41               | 0.4% |
| 130.150 optic disc coloboma                           | 49               | 0.1% | 6                | 0.1% |
| <b>OTHER</b>  |                  |      |                  |      |
| 900.000 other, unspecified                            | 433              | 0.9% | 0                |      |
| 900.100 other, not inherited                          | 897              | 1.9% | 19               | 0.2% |

| <b>OTHER CONTINUED</b>                                    | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 900.110 other. suspect not inherited/significance unknown | 458 1.0%         | 419 4.0%         |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 38,825 80.9%     | 7,931 74.9%      |

# PORTUGUESE PODENGO PEQUENO

|    | DISORDER  | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|---|---------------------|-----------|-----------------|-----------------------------------|
| A. | Distichiasis  | Not defined         | 1         | Breeder option  |                                   |
| B. | Persistent pupillary membranes<br>- iris to iris            | Not defined         | 1         | Breeder option  |                                   |
| C. | Cataract  | Not defined         | 1         | NO              |                                   |
| D. | Vitreous degeneration                                       | Not defined         | 1         | Breeder option  |                                   |
| E. | Retinal atrophy - rod-cone dysplasia type 3 ( <i>rcd3</i> ) | Autosomal recessive | 1         | NO              | Mutation in the <i>PDE6A</i> gene |
| F. | Retinal atrophy ( <i>prcd</i> )                             | Autosomal recessive | 2         | NO              | Mutation in the <i>prcd</i> gene  |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataracts

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary

membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

E. Retinal atrophy - rod-cone dysplasia type 3 (*rcd3*)

PRA in the Portuguese Podengo Pequeno is an autosomal recessive trait caused by a one base pair deletion in the gene encoding the alpha subunit of cyclic GMP phosphodiesterase (*rcd3*). PRA begins early in life with clinical signs of night blindness and a lack of rod ERG responses is seen at 6-8 weeks of age. Dogs are completely blind by 2-3 years of age when ophthalmoscopic signs are first visible. The mutation is found in the *PDE6A* gene. A DNA test is available.

F. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Other forms of retinal degeneration that are not *prcd* are recognized in the Portuguese Podengo Pequeno. The currently available genetic test will not detect these other forms of PRA.

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2. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006;88:551-563. PMID: 16938425

# OCULAR DISORDERS REPORT PORTUGUESE PODENGO PEQUENO

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 20.140          | ectopic cilia  | 0         |       | 1         | 0.4%  |
| 25.110          | distichiasis   | 8         | 5.1%  | 13        | 5.3%  |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.700          | corneal dystrophy  | 0         |       | 4         | 1.6%  |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 8         | 5.1%  | 15        | 6.1%  |
| 93.730          | persistent pupillary membranes, iris to cornea               | 1         | 0.6%  | 0         |       |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 0         |       | 1         | 0.4%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 6         | 3.8%  | 10        | 4.1%  |
| 100.301         | punctate cataract, anterior cortex                           | 1         | 0.6%  | 0         |       |
| 100.302         | punctate cataract, posterior cortex                          | 0         |       | 1         | 0.4%  |
| 100.303         | punctate cataract, equatorial cortex                         | 0         |       | 2         | 0.8%  |
| 100.306         | punctate cataract, nucleus                                   | 0         |       | 1         | 0.4%  |
| 100.311         | incipient cataract, anterior cortex                          | 1         | 0.6%  | 4         | 1.6%  |
| 100.312         | incipient cataract, posterior cortex                         | 2         | 1.3%  | 1         | 0.4%  |
| 100.313         | incipient cataract, equatorial cortex                        | 0         |       | 1         | 0.4%  |
| 100.315         | incipient cataract, posterior sutures                        | 0         |       | 2         | 0.8%  |
| 100.316         | incipient cataract, nucleus                                  | 0         |       | 3         | 1.2%  |
| 100.317         | incipient cataract, capsular                                 | 1         | 0.6%  | 0         |       |
| 100.325         | incomplete cataract, posterior sutures                       | 0         |       | 1         | 0.4%  |
| 100.330         | generalized/complete cataract                                | 1         | 0.6%  | 0         |       |
| 100.340         | resorbing/hypermature cataract                               | 1         | 0.6%  | 0         |       |
| 100.345         | <i>significant cataracts (summary)</i>                       | 7         | 4.4%  | 16        | 6.6%  |
| 100.375         | <i>subluxation/luxation, unspecified</i>                     | 2         | 1.3%  | 1         | 0.4%  |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.120         | persistent hyaloid artery/remnant                            | 1         | 0.6%  | 1         | 0.4%  |
| 110.200         | vitreal degeneration-anterior chamber                        | 2         | 1.3%  | 4         | 1.6%  |
| 110.320         | vitreal degeneration   | 8         | 5.1%  | 4         | 1.6%  |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.180         | retinal dysplasia, geographic                                | 0         |       | 1         | 0.4%  |
| 120.310         | generalized progressive retinal atrophy (PRA)                | 2         | 1.3%  | 3         | 1.2%  |
| 120.960         | retinopathy  | 2         | 1.3%  | 1         | 0.4%  |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.100         | other, not inherited   | 0         |       | 1         | 0.4%  |
| 900.110         | other. suspect not inherited/significance unknown            | 4         | 2.5%  | 10        | 4.1%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 121       | 76.6% | 178       | 73.0% |

# PORTUGUESE WATER DOG

|    | DISORDER   | INHERITANCE                | REFERENCE | BREEDING<br>ADVICE                        | GENETIC TESTS<br>AVAILABLE       |
|----|--|----------------------------|-----------|---|----------------------------------|
| A. | Microphthalmia with multiple ocular defects  | Autosomal recessive        | 1-3       | NO  | Mutation is not yet published    |
| B. | Distichiasis   | Not defined                | 1         | Breeder option                            |                                  |
| C. | Corneal dystrophy - epithelial/stromal   | Not defined                | 1         | Breeder option                            |                                  |
| D. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined | 1<br>1    | Breeder option<br>Passes with no notation |                                  |
| E. | Cataract   | Not defined                | 1         | NO  |                                  |
| F. | Y-suture tip opacity   | Not defined                | 1         | Breeder option                            |                                  |
| G. | Retinal atrophy ( <i>prcd</i> )  | Autosomal recessive        | 1, 4, 5   | NO  | Mutation in the <i>prcd</i> gene |
| H. | Retinal dysplasia - folds  | Not defined                | 1         | Breeder option                            |                                  |

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## Description and Comments

### A. Microphthalmia with multiple congenital ocular defects

This is a congenital abnormality present bilaterally and characterized by a small globe and associated ocular defects which can affect the cornea, anterior chamber, lens and/or retina. These associated defects may be variable in severity. Several cases have been identified, all of which appeared to have a common ancestry. All affected animals so far identified have been the progeny of dogs that were phenotypically normal, suggesting that the defect is not dominantly inherited.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds.

Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

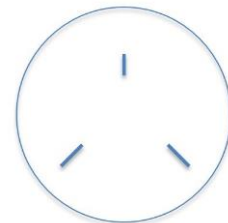
Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

F. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which



should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

G. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Portuguese Water Dog is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

A second, earlier onset form of PRA has also been identified recently in the Portuguese Water Dog. The onset of visual deficits occurs at 2-3 years of age, and, dogs show advanced retinal degeneration at the time visual deficits are recognized. The condition appears inherited as autosomal recessive. A DNA test is available.

H. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Case records (1986-1994), Section of Medical Genetics, School of Veterinary Medicine, University of Pennsylvania.
3. Shaw, G. C., et al. (2019). "Microphthalmia With Multiple Anterior Segment Defects in Portuguese Water Dogs." *Vet Pathol* **56**(2): 269-273. PMID: 30131012
4. Miyadera K, Aguirre G. A new form of early-onset pra in Portuguese Water Dogs - ECVO 2014 Abstract #65. *Vet Ophthalmol*. 2014;17:E25.
5. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006;88:551-563. Epub 2006/08/30. PMID: 16938425

# OCULAR DISORDERS REPORT PORTUGUESE WATER DOG

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>28,970 |      | 2016-2020<br>8,500 |      |
|---------------------|--|---------------------|------|--------------------|------|
|                     |  | #                   | %    | #                  | %    |
| <b>GLOBE</b>        |  |                     |      |                    |      |
| 0.110               | microphthalmia   | 15                  | 0.1% | 18                 | 0.2% |
| 10.000              | glaucoma   | 6                   | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                     |      |                    |      |
| 20.140              | ectopic cilia  | 3                   | 0.0% | 0                  |      |
| 20.160              | macropalpebral fissure   | 1                   | 0.0% | 0                  |      |
| 21.000              | entropion, unspecified   | 53                  | 0.2% | 17                 | 0.2% |
| 22.000              | ectropion, unspecified   | 3                   | 0.0% | 0                  |      |
| 25.110              | distichiasis   | 1,066               | 3.7% | 274                | 3.2% |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 0                   |      | 2                  | 0.0% |
| 40.910              | keratoconjunctivitis sicca                                     | 6                   | 0.0% | 5                  | 0.1% |
| <b>NICTITANS</b>    |  |                     |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 0                   |      | 1                  | 0.0% |
| 52.110              | prolapsed gland of the third eyelid                            | 1                   | 0.0% | 0                  |      |
| <b>CORNEA</b>       |  |                     |      |                    |      |
| 70.210              | corneal pannus   | 4                   | 0.0% | 0                  |      |
| 70.220              | pigmentary keratitis   | 4                   | 0.0% | 3                  | 0.0% |
| 70.700              | corneal dystrophy  | 189                 | 0.7% | 140                | 1.6% |
| 70.730              | corneal endothelial degeneration                               | 4                   | 0.0% | 3                  | 0.0% |
| <b>UVEA</b>         |  |                     |      |                    |      |
| 93.110              | iris hypoplasia  | 2                   | 0.0% | 2                  | 0.0% |
| 93.140              | corneal endothelial pigment without PPM                        | 2                   | 0.0% | 0                  |      |
| 93.150              | iris coloboma  | 1                   | 0.0% | 0                  |      |
| 93.180              | iris sphincter dysplasia                                       | 0                   |      | 1                  | 0.0% |
| 93.710              | persistent pupillary membranes, iris to iris                   | 1,755               | 6.1% | 686                | 8.1% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 39                  | 0.1% | 14                 | 0.2% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 32                  | 0.1% | 4                  | 0.0% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 43                  | 0.1% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 33                  | 0.1% | 53                 | 0.6% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 8                   | 0.0% | 4                  | 0.0% |
| 93.810              | uveal melanoma   | 6                   | 0.0% | 1                  | 0.0% |
| 93.999              | uveal cysts  | 13                  | 0.0% | 2                  | 0.0% |
| <b>LENS</b>         |  |                     |      |                    |      |
| 100.200             | cataract, unspecified  | 69                  | 0.2% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 1,901               | 6.6% | 559                | 6.6% |
| 100.301             | punctate cataract, anterior cortex                             | 146                 | 0.5% | 44                 | 0.5% |
| 100.302             | punctate cataract, posterior cortex                            | 57                  | 0.2% | 20                 | 0.2% |
| 100.303             | punctate cataract, equatorial cortex                           | 54                  | 0.2% | 8                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 22                  | 0.1% | 2                  | 0.0% |
| 100.305             | punctate cataract, posterior sutures                           | 30                  | 0.1% | 14                 | 0.2% |
| 100.306             | punctate cataract, nucleus                                     | 14                  | 0.0% | 6                  | 0.1% |
| 100.307             | punctate cataract, capsular                                    | 21                  | 0.1% | 11                 | 0.1% |
| 100.311             | incipient cataract, anterior cortex                            | 97                  | 0.3% | 23                 | 0.3% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.312 incipient cataract, posterior cortex              | 86               | 0.3%  | 21               | 0.2%  |
| 100.313 incipient cataract, equatorial cortex             | 82               | 0.3%  | 17               | 0.2%  |
| 100.314 incipient cataract, anterior sutures              | 12               | 0.0%  | 1                | 0.0%  |
| 100.315 incipient cataract, posterior sutures             | 15               | 0.1%  | 3                | 0.0%  |
| 100.316 incipient cataract, nucleus                       | 21               | 0.1%  | 3                | 0.0%  |
| 100.317 incipient cataract, capsular                      | 21               | 0.1%  | 8                | 0.1%  |
| 100.321 incomplete cataract, anterior cortex              | 6                | 0.0%  | 9                | 0.1%  |
| 100.322 incomplete cataract, posterior cortex             | 7                | 0.0%  | 8                | 0.1%  |
| 100.323 incomplete cataract, equatorial cortex            | 1                | 0.0%  | 5                | 0.1%  |
| 100.324 incomplete cataract, anterior sutures             | 1                | 0.0%  | 0                |       |
| 100.326 incomplete cataract, nucleus                      | 1                | 0.0%  | 1                | 0.0%  |
| 100.328 y-suture tip opacities                            | 16               | 0.1%  | 31               | 0.4%  |
| 100.330 generalized/complete cataract                     | 70               | 0.2%  | 12               | 0.1%  |
| 100.340 resorbing/hypermature cataract                    | 1                | 0.0%  | 1                | 0.0%  |
| 100.345 <i>significant cataracts (summary)</i>            | 850              | 2.9%  | 248              | 2.9%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 11               | 0.0%  | 3                | 0.0%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 43               | 0.1%  | 20               | 0.2%  |
| 110.135 PHPV/PTVL   | 16               | 0.1%  | 3                | 0.0%  |
| 110.200 vitreous degeneration-anterior chamber            | 2                | 0.0%  | 6                | 0.1%  |
| 110.320 vitreal degeneration                              | 44               | 0.2%  | 7                | 0.1%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 2                | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 224              | 0.8%  | 89               | 1.0%  |
| 120.180 retinal dysplasia, geographic                     | 19               | 0.1%  | 1                | 0.0%  |
| 120.190 retinal dysplasia, detached                       | 2                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 173              | 0.6%  | 5                | 0.1%  |
| 120.400 retinal hemorrhage                                | 8                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 3                | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 2                | 0.0%  | 1                | 0.0%  |
| 120.960 retinopathy                                       | 1                | 0.0%  | 5                | 0.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 14               | 0.0%  | 6                | 0.1%  |
| 130.120 optic nerve hypoplasia                            | 11               | 0.0%  | 0                |       |
| 130.150 optic disc coloboma                               | 6                | 0.0%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 313              | 1.1%  | 0                |       |
| 900.100 other, not inherited                              | 535              | 1.8%  | 5                | 0.1%  |
| 900.110 other. suspect not inherited/significance unknown | 279              | 1.0%  | 307              | 3.6%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 24,389           | 84.2% | 6,371            | 75.0% |

# PUG

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Entropion  | Not defined        | 1                | Breeder option         |
| B. | Distichiasis                                     | Not defined        | 1                | Breeder option         |
| C. | Pigmentary Keratitis/Pigmentary Keratopathy      | Not defined        | 1-3              | Breeder option         |
| D. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |
| E. | Cataract   | Not defined        | 1, 4             | NO                     |
| F. | Vitreous degeneration                            | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. In the Pug, entropion usually involves the medial canthal margin of the lower eyelid(s).

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### C. Exposure/Pigmentary keratitis/Pigmentary keratopathy

A condition characterized by variable degrees of superficial vascularization, fibrosis and/or pigmentation of the cornea. May be associated with excessive exposure/irritation of the globe due to shallow orbits, lower eyelid medial entropion, lagophthalmos and macropalpebral fissure.

The breed standard indicates the Pug should have a "large massive round head with very large, bold and prominent eyes." These characteristics give rise to the ocular exposure and irritative problems common in the breed.

Pigmentary keratopathy is a condition reported in Pugs in which the cornea becomes pigmented, often resulting in vision impairment. Development of pigmentary keratopathy is associated with congenital uveal pathology - iris hypoplasia and the presence of persistent pupillary membranes - but not with other factors such as Schirmer tear test values or medial canthal entropion.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally in the neonatal period. These strands may bridge from iris to iris, iris to cornea, iris to lens, or from sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

F. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Labelle AL, Dresser CB, Hamor RE, et al. Characteristics of, prevalence of, and risk factors for corneal pigmentation (pigmentary keratopathy) in Pugs. *J Am Vet Med Assoc.* 2013;243:667-674.
3. Maini, S., et al. (2019). "Pigmentary keratitis in pugs in the United Kingdom: prevalence and associated features." *BMC Vet Res* **15**(1): 384. PMID: 31666065
4. Gelatt KN, Mackay EO. Prevalence of primary breed-related cataracts in the dog in North America. *Vet Ophthalmol.* 2005;8:101-111.

# OCULAR DISORDERS REPORT PUG

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>2,564 |       | 2016-2020<br>694 |       |
|---|---------------------|--------------------|-------|------------------|-------|
|   |                     | #                  | %     | #                | %     |
| <b>GLOBE</b>  |                     |                    |       |                  |       |
| 0.110 microphthalmia  |                     | 3                  | 0.1%  | 0                |       |
| <b>EYELIDS</b>  |                     |                    |       |                  |       |
| 20.110 eyelid dermoid   |                     | 1                  | 0.0%  | 0                |       |
| 20.140 ectopic cilia  |                     | 14                 | 0.5%  | 1                | 0.1%  |
| 20.160 macropalpebral fissure   |                     | 67                 | 2.6%  | 0                |       |
| 21.000 entropion, unspecified   |                     | 482                | 18.8% | 108              | 15.6% |
| 22.000 ectropion, unspecified   |                     | 11                 | 0.4%  | 0                |       |
| 25.110 distichiasis   |                     | 228                | 8.9%  | 50               | 7.2%  |
| <b>NASOLACRIMAL</b>   |                     |                    |       |                  |       |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 0                  |       | 1                | 0.1%  |
| 40.910 keratoconjunctivitis sicca                                     |                     | 7                  | 0.3%  | 2                | 0.3%  |
| <b>NICTITANS</b>  |                     |                    |       |                  |       |
| 50.210 pannus of third eyelid   |                     | 1                  | 0.0%  | 0                |       |
| <b>CORNEA</b>   |                     |                    |       |                  |       |
| 70.210 corneal pannus   |                     | 80                 | 3.1%  | 0                |       |
| 70.220 pigmentary keratitis   |                     | 787                | 30.7% | 348              | 50.1% |
| 70.700 corneal dystrophy  |                     | 14                 | 0.5%  | 1                | 0.1%  |
| 70.730 corneal endothelial degeneration                               |                     | 4                  | 0.2%  | 0                |       |
| <b>UVEA</b>   |                     |                    |       |                  |       |
| 90.250 pigmentary uveitis   |                     | 0                  |       | 1                | 0.1%  |
| 93.150 iris coloboma  |                     | 2                  | 0.1%  | 1                | 0.1%  |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 264                | 10.3% | 87               | 12.5% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 8                  | 0.3%  | 0                |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 16                 | 0.6%  | 1                | 0.1%  |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 1                  | 0.0%  | 0                |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 0                  |       | 1                | 0.1%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 5                  | 0.2%  | 3                | 0.4%  |
| 93.999 uveal cysts  |                     | 2                  | 0.1%  | 0                |       |
| <b>LENS</b>   |                     |                    |       |                  |       |
| 100.200 cataract, unspecified   |                     | 4                  | 0.2%  | 0                |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 52                 | 2.0%  | 14               | 2.0%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 4                  | 0.2%  | 1                | 0.1%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 4                  | 0.2%  | 2                | 0.3%  |
| 100.303 punctate cataract, equatorial cortex                          |                     | 5                  | 0.2%  | 0                |       |
| 100.304 punctate cataract, anterior sutures                           |                     | 2                  | 0.1%  | 0                |       |
| 100.305 punctate cataract, posterior sutures                          |                     | 6                  | 0.2%  | 0                |       |
| 100.306 punctate cataract, nucleus                                    |                     | 4                  | 0.2%  | 0                |       |
| 100.307 punctate cataract, capsular                                   |                     | 3                  | 0.1%  | 1                | 0.1%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 18                 | 0.7%  | 3                | 0.4%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 16                 | 0.6%  | 4                | 0.6%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 8                  | 0.3%  | 0                |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 8                  | 0.3%  | 1                | 0.1%  |
| 100.316 incipient cataract, nucleus                                   |                     | 4                  | 0.2%  | 0                |       |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.317 incipient cataract, capsular                      | 5 0.2%           | 0                |
| 100.321 incomplete cataract, anterior cortex              | 3 0.1%           | 1 0.1%           |
| 100.322 incomplete cataract, posterior cortex             | 3 0.1%           | 0                |
| 100.324 incomplete cataract, anterior sutures             | 0                | 1 0.1%           |
| 100.325 incomplete cataract, posterior sutures            | 1 0.0%           | 1 0.1%           |
| 100.326 incomplete cataract, nucleus                      | 1 0.0%           | 0                |
| 100.330 generalized/complete cataract                     | 13 0.5%          | 0                |
| 100.345 <i>significant cataracts (summary)</i>            | 112 4.4%         | 15 2.2%          |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 13 0.5%          | 3 0.4%           |
| 110.135 PHPV/PTVL   | 3 0.1%           | 0                |
| 110.200 vitreous degeneration-anterior chamber            | 1 0.0%           | 0                |
| 110.320 vitreal degeneration                              | 28 1.1%          | 2 0.3%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.120 coloboma   | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 19 0.7%          | 2 0.3%           |
| 120.180 retinal dysplasia, geographic                     | 11 0.4%          | 1 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 3 0.1%           | 0                |
| 120.400 retinal hemorrhage                                | 1 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.120 optic nerve hypoplasia                            | 1 0.0%           | 0                |
| 130.150 optic disc coloboma                               | 1 0.0%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 36 1.4%          | 0                |
| 900.100 other, not inherited                              | 162 6.3%         | 7 1.0%           |
| 900.110 other. suspect not inherited/significance unknown | 90 3.5%          | 37 5.3%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,050 41.0%      | 222 32.0%        |

# PULI

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|--|---------------------|-----------|-----------------|--------------------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option  |                                      |
| B. | Cataract   | Not defined         | 1         | NO              |                                      |
| C. | Lens luxation                                    | Autosomal recessive | 2         | NO              | Mutation in the <i>ADAMTS17</i> gene |
| D. | Retinal atrophy ( <i>prcd</i> )                  | Autosomal recessive | 1         | NO              | Mutation in the <i>prcd</i> gene     |
| E. | Retinal dysplasia<br>- folds                     | Not defined         | 1         | Breeder option  |                                      |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation may result in blinding retinal detachment and/or elevated intraocular pressure (glaucoma) causing vision impairment, pain, and blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.



D. Retinal atrophy – *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Puli is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

E. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gould D, Pettitt L, McLaughlin B, et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011;14:378-384.

# OCULAR DISORDERS REPORT

## PULI

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 20.110 eyelid dermoid   |                     | 1         | 0.1%  | 0         |       |
| 20.140 ectopic cilia  |                     | 1         | 0.1%  | 0         |       |
| 20.160 macropalpebral fissure   |                     | 1         | 0.1%  | 0         |       |
| 21.000 entropion, unspecified   |                     | 8         | 0.7%  | 0         |       |
| 25.110 distichiasis   |                     | 7         | 0.6%  | 0         |       |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.220 pigmentary keratitis   |                     | 5         | 0.5%  | 0         |       |
| 70.700 corneal dystrophy  |                     | 18        | 1.7%  | 0         |       |
| 70.730 corneal endothelial degeneration                               |                     | 1         | 0.1%  | 0         |       |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 244       | 22.5% | 35        | 20.8% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 14        | 1.3%  | 0         |       |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 8         | 0.7%  | 0         |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 5         | 0.5%  | 3         | 1.8%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1         | 0.1%  | 0         |       |
| 93.999 uveal cysts  |                     | 1         | 0.1%  | 0         |       |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.200 cataract, unspecified   |                     | 3         | 0.3%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 63        | 5.8%  | 6         | 3.6%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 5         | 0.5%  | 4         | 2.4%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 2         | 0.2%  | 2         | 1.2%  |
| 100.305 punctate cataract, posterior sutures                          |                     | 6         | 0.6%  | 2         | 1.2%  |
| 100.306 punctate cataract, nucleus                                    |                     | 3         | 0.3%  | 0         |       |
| 100.307 punctate cataract, capsular                                   |                     | 2         | 0.2%  | 0         |       |
| 100.311 incipient cataract, anterior cortex                           |                     | 8         | 0.7%  | 4         | 2.4%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 4         | 0.4%  | 2         | 1.2%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 7         | 0.6%  | 0         |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 1         | 0.1%  | 0         |       |
| 100.316 incipient cataract, nucleus                                   |                     | 3         | 0.3%  | 0         |       |
| 100.317 incipient cataract, capsular                                  |                     | 1         | 0.1%  | 0         |       |
| 100.321 incomplete cataract, anterior cortex                          |                     | 0         |       | 1         | 0.6%  |
| 100.322 incomplete cataract, posterior cortex                         |                     | 0         |       | 2         | 1.2%  |
| 100.328 y-suture tip opacities  |                     | 0         |       | 2         | 1.2%  |
| 100.330 generalized/complete cataract                                 |                     | 7         | 0.6%  | 0         |       |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 52        | 4.8%  | 19        | 11.3% |
| 100.375 <i>subluxation/luxation, unspecified</i>                      |                     | 1         | 0.1%  | 0         |       |
| <b>VITREOUS</b>   |                     |           |       |           |       |
| 110.120 persistent hyaloid artery/remnant                             |                     | 2         | 0.2%  | 0         |       |
| 110.135 PHPV/PTVL   |                     | 1         | 0.1%  | 0         |       |
| 110.200 vitreous degeneration-anterior chamber                        |                     | 0         |       | 1         | 0.6%  |
| 110.320 vitreal degeneration  |                     | 1         | 0.1%  | 0         |       |
| <b>RETINA</b>   |                     |           |       |           |       |
| 120.170 retinal dysplasia, folds                                      |                     | 45        | 4.2%  | 6         | 3.6%  |
| 120.180 retinal dysplasia, geographic                                 |                     | 3         | 0.3%  | 0         |       |

| <b>RETINA CONTINUED</b>                                   | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 120.310 generalized progressive retinal atrophy (PRA)     | 4 0.4%           | 0                |
| 120.400 retinal hemorrhage                                | 1 0.1%           | 0                |
| 120.910 retinal detachment without dialysis               | 2 0.2%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 2 0.2%           | 0                |
| 130.120 optic nerve hypoplasia                            | 3 0.3%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 13 1.2%          | 0                |
| 900.100 other, not inherited                              | 46 4.2%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 7 0.6%           | 5 3.0%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 735 67.8%        | 108 64.3%        |

# PYRENEAN SHEPHERD

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |
| B. | Cataract   | Not defined        | 1                | NO                     |
| C. | Choroidal hypoplasia                             | Not defined        | 1                | NO                     |
| D. | Retinal dysplasia – folds                        | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Choroidal hypoplasia

Inadequate development of the choroid present at birth and non-progressive. This condition is more commonly identified in the Collie breed where it is a manifestation of "Collie Eye Anomaly."

### D. Retinal dysplasia – folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Pyrenean Shepherd. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT PYRENEAN SHEPHERD

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015<br>421 |      | 2016-2020<br>276 |      |
|--|---------------------|------------------|------|------------------|------|
|  |                     | #                | %    | #                | %    |
| <b>GLOBE</b>   |                     |                  |      |                  |      |
| 0.110 microphthalmia   |                     | 0                |      | 2                | 0.7% |
| <b>EYELIDS</b>   |                     |                  |      |                  |      |
| 21.000 entropion, unspecified                                |                     | 0                |      | 1                | 0.4% |
| 25.110 distichiasis  |                     | 0                |      | 2                | 0.7% |
| <b>NASOLACRIMAL</b>  |                     |                  |      |                  |      |
| 32.110 imperforate lower nasolacrimal punctum                |                     | 1                | 0.2% | 1                | 0.4% |
| <b>NICTITANS</b>   |                     |                  |      |                  |      |
| 52.110 prolapsed gland of the third eyelid                   |                     | 1                | 0.2% | 0                |      |
| <b>CORNEA</b>  |                     |                  |      |                  |      |
| 70.700 corneal dystrophy                                     |                     | 1                | 0.2% | 2                | 0.7% |
| <b>UVEA</b>  |                     |                  |      |                  |      |
| 93.110 iris hypoplasia                                       |                     | 1                | 0.2% | 2                | 0.7% |
| 93.150 iris coloboma   |                     | 1                | 0.2% | 1                | 0.4% |
| 93.710 persistent pupillary membranes, iris to iris          |                     | 26               | 6.2% | 6                | 2.2% |
| 93.740 persistent pupillary membranes, iris sheets           |                     | 1                | 0.2% | 0                |      |
| <b>LENS</b>  |                     |                  |      |                  |      |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 12               | 2.9% | 4                | 1.4% |
| 100.301 punctate cataract, anterior cortex                   |                     | 2                | 0.5% | 1                | 0.4% |
| 100.302 punctate cataract, posterior cortex                  |                     | 1                | 0.2% | 1                | 0.4% |
| 100.303 punctate cataract, equatorial cortex                 |                     | 1                | 0.2% | 0                |      |
| 100.305 punctate cataract, posterior sutures                 |                     | 1                | 0.2% | 2                | 0.7% |
| 100.311 incipient cataract, anterior cortex                  |                     | 5                | 1.2% | 0                |      |
| 100.312 incipient cataract, posterior cortex                 |                     | 1                | 0.2% | 1                | 0.4% |
| 100.313 incipient cataract, equatorial cortex                |                     | 2                | 0.5% | 0                |      |
| 100.315 incipient cataract, posterior sutures                |                     | 1                | 0.2% | 0                |      |
| 100.316 incipient cataract, nucleus                          |                     | 3                | 0.7% | 4                | 1.4% |
| 100.322 incomplete cataract, posterior cortex                |                     | 3                | 0.7% | 1                | 0.4% |
| 100.326 incomplete cataract, nucleus                         |                     | 1                | 0.2% | 1                | 0.4% |
| 100.328 y-suture tip opacities                               |                     | 0                |      | 1                | 0.4% |
| 100.345 <i>significant cataracts (summary)</i>               |                     | 21               | 5.0% | 12               | 4.3% |
| 100.375 <i>subluxation/luxation, unspecified</i>             |                     | 1                | 0.2% | 0                |      |
| <b>VITREOUS</b>  |                     |                  |      |                  |      |
| 110.120 persistent hyaloid artery/remnant                    |                     | 4                | 1.0% | 2                | 0.7% |
| 110.135 PHPV/PTVL  |                     | 0                |      | 3                | 1.1% |
| 110.320 vitreal degeneration                                 |                     | 1                | 0.2% | 0                |      |
| <b>FUNDUS</b>  |                     |                  |      |                  |      |
| 97.110 choroidal hypoplasia                                  |                     | 17               | 4.0% | 6                | 2.2% |
| <b>RETINA</b>  |                     |                  |      |                  |      |
| 120.170 retinal dysplasia, folds                             |                     | 9                | 2.1% | 6                | 2.2% |
| 120.180 retinal dysplasia, geographic                        |                     | 1                | 0.2% | 0                |      |
| 120.310 generalized progressive retinal atrophy (PRA)        |                     | 1                | 0.2% | 0                |      |

| <b>RETINA CONTINUED</b>                                   | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 120.920 retinal detachment with dialysis                  | 0                | 1 0.4%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 0                | 2 0.7%           |
| 130.120 optic nerve hypoplasia                            | 0                | 1 0.4%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 9 2.1%           | 0                |
| 900.100 other, not inherited                              | 11 2.6%          | 0                |
| 900.110 other. suspect not inherited/significance unknown | 4 1.0%           | 16 5.8%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 351 83.4%        | 222 80.4%        |

# RAT TERRIER

|    | DISORDER                        | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|---------------------------------|---------------------|-----------|-----------------|--------------------------------------|
| A. | Retinal atrophy ( <i>prcd</i> ) | Autosomal recessive | 1         | NO              | Mutation in the <i>prcd</i> gene     |
| B. | Lens luxation                   | Autosomal recessive | 2, 3      | NO              | Mutation in the <i>ADAMTS17</i> gene |

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## Description and Comments

### A. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

### B. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Farias FH, Johnson GS, Taylor JF, et al. An *ADAMTS17* splice donor site mutation in dogs with primary lens luxation. *Invest Ophthalmol Vis Sci.* 2010;51:4716-4721.
3. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011;14:378-384.



# OCULAR DISORDERS REPORT RAT TERRIER

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015<br>267 |       | 2016-2020<br>67 |       |
|-----------------|--|------------------|-------|-----------------|-------|
|                 |  | #                | %     | #               | %     |
| <b>EYELIDS</b>  |  |                  |       |                 |       |
| 25.110          | distichiasis   | 4                | 1.5%  | 2               | 3.0%  |
| <b>UVEA</b>     |  |                  |       |                 |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 7                | 2.6%  | 2               | 3.0%  |
| 93.730          | persistent pupillary membranes, iris to cornea               | 1                | 0.4%  | 0               |       |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 2                | 0.7%  | 0               |       |
| <b>LENS</b>     |  |                  |       |                 |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 5                | 1.9%  | 0               |       |
| 100.303         | punctate cataract, equatorial cortex                         | 1                | 0.4%  | 0               |       |
| 100.311         | incipient cataract, anterior cortex                          | 3                | 1.1%  | 0               |       |
| 100.312         | incipient cataract, posterior cortex                         | 3                | 1.1%  | 0               |       |
| 100.313         | incipient cataract, equatorial cortex                        | 2                | 0.7%  | 0               |       |
| 100.315         | incipient cataract, posterior sutures                        | 1                | 0.4%  | 0               |       |
| 100.316         | incipient cataract, nucleus                                  | 1                | 0.4%  | 0               |       |
| 100.330         | generalized/complete cataract                                | 4                | 1.5%  | 0               |       |
| 100.345         | significant cataracts (summary)                              | 15               | 5.6%  | 0               |       |
| 100.375         | subluxation/luxation, unspecified                            | 3                | 1.1%  | 0               |       |
| <b>VITREOUS</b> |  |                  |       |                 |       |
| 110.200         | vitreal degeneration-anterior chamber                        | 1                | 0.4%  | 1               | 1.5%  |
| 110.320         | vitreal degeneration   | 3                | 1.1%  | 0               |       |
| <b>RETINA</b>   |  |                  |       |                 |       |
| 120.190         | retinal dysplasia, detached                                  | 1                | 0.4%  | 0               |       |
| 120.310         | generalized progressive retinal atrophy (PRA)                | 1                | 0.4%  | 0               |       |
| <b>OTHER</b>    |  |                  |       |                 |       |
| 900.000         | other, unspecified   | 3                | 1.1%  | 0               |       |
| 900.110         | other. suspect not inherited/significance unknown            | 1                | 0.4%  | 2               | 3.0%  |
| <b>NORMAL</b>   |  |                  |       |                 |       |
| 0.000           | normal globe   | 242              | 90.6% | 60              | 89.6% |

# **OCULAR DISORDERS REPORT REDBONE COONHOUND**

There are insufficient breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the REDBONE COONHOUND breed. Therefore, there are no conditions listed with breeding advice.

## OCULAR DISORDERS REPORT REDBONE COONHOUND

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|------------------|--|-----------|-------|-----------|-------|
|                  |  | #         | %     | #         | %     |
| <b>EYELIDS</b>   |  |           |       |           |       |
| 21.000           | entropion, unspecified                                       | 0         |       | 2         | 5.3%  |
| 25.110           | distichiasis   | 0         |       | 1         | 2.6%  |
| <b>NICTITANS</b> |  |           |       |           |       |
| 52.110           | prolapsed gland of the third eyelid                          | 0         |       | 1         | 2.6%  |
| <b>UVEA</b>      |  |           |       |           |       |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands | 1         | 5.0%  | 0         |       |
| <b>LENS</b>      |  |           |       |           |       |
| 100.210          | cataract. suspect not inherited/significance unknown         | 0         |       | 2         | 5.3%  |
| <b>RETINA</b>    |  |           |       |           |       |
| 120.170          | retinal dysplasia, folds                                     | 0         |       | 1         | 2.6%  |
| 120.310          | generalized progressive retinal atrophy (PRA)                | 1         | 5.0%  | 0         |       |
| 120.960          | retinopathy  | 0         |       | 1         | 2.6%  |
| <b>OTHER</b>     |  |           |       |           |       |
| 900.110          | other. suspect not inherited/significance unknown            | 1         | 5.0%  | 1         | 2.6%  |
| <b>NORMAL</b>    |  |           |       |           |       |
| 0.000            | normal globe   | 18        | 90.0% | 30        | 78.9% |

# RHODESIAN RIDGEBACK

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--------------------------------|-------------|-----------|-------------------------|
| A. | Distichiasis                   | Not defined | 1         | Breeder option          |
| B. | Entropion                      | Not defined | 2         | NO                      |
| C. | Persistent pupillary membranes |             |           |                         |
|    | - iris to iris                 | Not defined | 1         | Breeder option          |
|    | - lens pigment foci/no strands | Not defined | 1         | Passes with no notation |
| D. | Cataract                       | Not defined | 1         | NO                      |
| E. | Y-suture tip opacity           | Not defined | 1         | Breeder option          |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

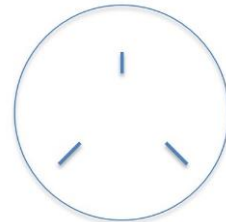
Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

#### D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

#### E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Rhodesian Ridgeback breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Breed club request to ACVO Genetics Committee, 2008.

# OCULAR DISORDERS REPORT RHODESIAN RIDGEBACK

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 0.110 microphthalmia  |                     | 2         | 0.0% | 0         |      |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 20.140 ectopic cilia  |                     | 0         |      | 1         | 0.1% |
| 21.000 entropion, unspecified   |                     | 15        | 0.3% | 2         | 0.1% |
| 22.000 ectropion, unspecified   |                     | 1         | 0.0% | 0         |      |
| 25.110 distichiasis   |                     | 129       | 2.9% | 27        | 1.9% |
| <b>NICTITANS</b>  |                     |           |      |           |      |
| 51.100 third eyelid cartilage anomaly                                 |                     | 5         | 0.1% | 1         | 0.1% |
| 52.110 prolapsed gland of the third eyelid                            |                     | 3         | 0.1% | 0         |      |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.210 corneal pannus   |                     | 6         | 0.1% | 0         |      |
| 70.700 corneal dystrophy  |                     | 24        | 0.5% | 7         | 0.5% |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.110 iris hypoplasia  |                     | 1         | 0.0% | 0         |      |
| 93.140 corneal endothelial pigment without PPM                        |                     | 4         | 0.1% | 0         |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 266       | 6.0% | 74        | 5.1% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 6         | 0.1% | 1         | 0.1% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 2         | 0.0% | 0         |      |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 1         | 0.0% | 0         |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 63        | 1.4% | 51        | 3.5% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 5         | 0.1% | 0         |      |
| 93.810 uveal melanoma   |                     | 2         | 0.0% | 1         | 0.1% |
| 93.999 uveal cysts  |                     | 5         | 0.1% | 1         | 0.1% |
| 97.150 chorioretinal coloboma, congenital                             |                     | 1         | 0.0% | 1         | 0.1% |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.200 cataract, unspecified   |                     | 4         | 0.1% | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 214       | 4.8% | 51        | 3.5% |
| 100.301 punctate cataract, anterior cortex                            |                     | 11        | 0.2% | 3         | 0.2% |
| 100.302 punctate cataract, posterior cortex                           |                     | 44        | 1.0% | 13        | 0.9% |
| 100.303 punctate cataract, equatorial cortex                          |                     | 2         | 0.0% | 0         |      |
| 100.304 punctate cataract, anterior sutures                           |                     | 0         |      | 2         | 0.1% |
| 100.305 punctate cataract, posterior sutures                          |                     | 21        | 0.5% | 6         | 0.4% |
| 100.307 punctate cataract, capsular                                   |                     | 9         | 0.2% | 5         | 0.3% |
| 100.311 incipient cataract, anterior cortex                           |                     | 6         | 0.1% | 4         | 0.3% |
| 100.312 incipient cataract, posterior cortex                          |                     | 82        | 1.8% | 21        | 1.5% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 9         | 0.2% | 2         | 0.1% |
| 100.315 incipient cataract, posterior sutures                         |                     | 14        | 0.3% | 5         | 0.3% |
| 100.316 incipient cataract, nucleus                                   |                     | 5         | 0.1% | 0         |      |
| 100.317 incipient cataract, capsular                                  |                     | 15        | 0.3% | 6         | 0.4% |
| 100.322 incomplete cataract, posterior cortex                         |                     | 1         | 0.0% | 5         | 0.3% |
| 100.324 incomplete cataract, anterior sutures                         |                     | 1         | 0.0% | 0         |      |
| 100.325 incomplete cataract, posterior sutures                        |                     | 1         | 0.0% | 1         | 0.1% |
| 100.328 y-suture tip opacities  |                     | 6         | 0.1% | 10        | 0.7% |
| 100.330 generalized/complete cataract                                 |                     | 3         | 0.1% | 0         |      |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.345 significant cataracts (summary)                   | 234              | 5.3%  | 83               | 5.8%  |
| 100.375 subluxation/luxation, unspecified                 | 3                | 0.1%  | 0                |       |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 2                | 0.0%  | 6                | 0.4%  |
| 110.135 PHPV/PTVL   | 1                | 0.0%  | 0                |       |
| 110.200 vitreous degeneration-anterior chamber            | 1                | 0.0%  | 4                | 0.3%  |
| 110.320 vitreal degeneration                              | 11               | 0.2%  | 3                | 0.2%  |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 6                | 0.1%  | 4                | 0.3%  |
| 120.180 retinal dysplasia, geographic                     | 1                | 0.0%  | 1                | 0.1%  |
| 120.190 retinal dysplasia, detached                       | 1                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 4                | 0.1%  | 3                | 0.2%  |
| 120.910 retinal detachment without dialysis               | 2                | 0.0%  | 0                |       |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 1                | 0.0%  | 0                |       |
| 130.120 optic nerve hypoplasia                            | 1                | 0.0%  | 0                |       |
| 130.150 optic disc coloboma                               | 5                | 0.1%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 51               | 1.1%  | 0                |       |
| 900.100 other, not inherited                              | 90               | 2.0%  | 2                | 0.1%  |
| 900.110 other. suspect not inherited/significance unknown | 45               | 1.0%  | 36               | 2.5%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 3,715            | 83.7% | 1,142            | 79.1% |

# ROTTWEILER

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE            |
|----|--|-------------|-----------|----------------------------|
| A. | Corneal dystrophy<br>- epithelial/stromal                              | Not defined | 1         | Breeder option             |
| B. | Uveal cysts  | Not defined | 1-3       | Breeder option             |
| C. | Persistent pupillary<br>membranes<br>- lens pigment foci/no<br>strands | Not defined | 1         | Passes with no<br>notation |
| D. | Cataract   | Not defined | 1, 2      | NO                         |
| E. | Retinal atrophy -<br>generalized                                       | Not defined | 1         | NO                         |

## Description and Comments

**A. Corneal dystrophy - epithelial/stromal**

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

**B. Uveal cysts**

A pigmented, fluid-filled epithelial-lined structure arising from the posterior iris or ciliary body epithelium. Cysts may remain attached to the pupil margin, iris, or ciliary body, or may detach and be free-floating within the anterior chamber. They may rupture and adhere to the cornea or anterior lens capsule. Uveal cysts may occur in any breed. Uveal cysts are commonly benign, although they may be associated with other pathologic conditions in various breeds.

**C. Persistent pupillary membranes (PPMs)**

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

**D. Cataract**

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are



complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

A variety of cataracts have been observed in this breed ranging from the posterior polar cataract similar to that in the Golden Retriever and cataracts involving multiple areas of the nucleus and cortex. Further studies need to be performed as to the exact mode of inheritance, but it is our recommendation that the individually afflicted dog should not be bred.

E. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. Except for X-linked PRA in the Siberian Husky, in all breeds studied to date, PRA is inherited as an autosomal recessive trait.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Bjerkas E. Progressive retinal atrophy in dogs in Norway. *Norsk Veterinaertidsskrift*. 1991;103:601-610.

# OCULAR DISORDERS REPORT ROTTWEILER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>14,699 |      | 2016-2020<br>2,990 |      |
|---------------------|--|---------------------|------|--------------------|------|
|                     |  | #                   | %    | #                  | %    |
| <b>GLOBE</b>        |  |                     |      |                    |      |
| 0.110               | microphthalmia   | 3                   | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                     |      |                    |      |
| 20.140              | ectopic cilia  | 1                   | 0.0% | 0                  |      |
| 20.160              | macropalpebral fissure   | 10                  | 0.1% | 0                  |      |
| 21.000              | entropion, unspecified   | 115                 | 0.8% | 23                 | 0.8% |
| 22.000              | ectropion, unspecified   | 29                  | 0.2% | 2                  | 0.1% |
| 25.110              | distichiasis   | 85                  | 0.6% | 20                 | 0.7% |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 0                   |      | 1                  | 0.0% |
| 40.910              | keratoconjunctivitis sicca                                     | 3                   | 0.0% | 0                  |      |
| <b>NICTITANS</b>    |  |                     |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 4                   | 0.0% | 0                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 15                  | 0.1% | 2                  | 0.1% |
| <b>CORNEA</b>       |  |                     |      |                    |      |
| 70.210              | corneal pannus   | 3                   | 0.0% | 0                  |      |
| 70.220              | pigmentary keratitis   | 2                   | 0.0% | 0                  |      |
| 70.700              | corneal dystrophy  | 134                 | 0.9% | 27                 | 0.9% |
| 70.730              | corneal endothelial degeneration                               | 7                   | 0.0% | 0                  |      |
| <b>UVEA</b>         |  |                     |      |                    |      |
| 93.110              | iris hypoplasia  | 11                  | 0.1% | 4                  | 0.1% |
| 93.140              | corneal endothelial pigment without PPM                        | 1                   | 0.0% | 0                  |      |
| 93.150              | iris coloboma  | 49                  | 0.3% | 9                  | 0.3% |
| 93.710              | persistent pupillary membranes, iris to iris                   | 119                 | 0.8% | 21                 | 0.7% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 38                  | 0.3% | 4                  | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 51                  | 0.3% | 10                 | 0.3% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 8                   | 0.1% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 113                 | 0.8% | 150                | 5.0% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 10                  | 0.1% | 6                  | 0.2% |
| 93.810              | uveal melanoma   | 3                   | 0.0% | 1                  | 0.0% |
| 93.999              | uveal cysts  | 264                 | 1.8% | 115                | 3.8% |
| <b>LENS</b>         |  |                     |      |                    |      |
| 100.200             | cataract, unspecified  | 229                 | 1.6% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 875                 | 6.0% | 200                | 6.7% |
| 100.301             | punctate cataract, anterior cortex                             | 101                 | 0.7% | 54                 | 1.8% |
| 100.302             | punctate cataract, posterior cortex                            | 260                 | 1.8% | 42                 | 1.4% |
| 100.303             | punctate cataract, equatorial cortex                           | 9                   | 0.1% | 3                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 13                  | 0.1% | 6                  | 0.2% |
| 100.305             | punctate cataract, posterior sutures                           | 82                  | 0.6% | 12                 | 0.4% |
| 100.306             | punctate cataract, nucleus                                     | 28                  | 0.2% | 5                  | 0.2% |
| 100.307             | punctate cataract, capsular                                    | 42                  | 0.3% | 25                 | 0.8% |
| 100.311             | incipient cataract, anterior cortex                            | 105                 | 0.7% | 21                 | 0.7% |
| 100.312             | incipient cataract, posterior cortex                           | 512                 | 3.5% | 87                 | 2.9% |
| 100.313             | incipient cataract, equatorial cortex                          | 37                  | 0.3% | 6                  | 0.2% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.314 incipient cataract, anterior sutures              | 11               | 0.1%  | 3                | 0.1%  |
| 100.315 incipient cataract, posterior sutures             | 72               | 0.5%  | 15               | 0.5%  |
| 100.316 incipient cataract, nucleus                       | 54               | 0.4%  | 6                | 0.2%  |
| 100.317 incipient cataract, capsular                      | 42               | 0.3%  | 7                | 0.2%  |
| 100.321 incomplete cataract, anterior cortex              | 2                | 0.0%  | 5                | 0.2%  |
| 100.322 incomplete cataract, posterior cortex             | 6                | 0.0%  | 10               | 0.3%  |
| 100.323 incomplete cataract, equatorial cortex            | 1                | 0.0%  | 0                |       |
| 100.325 incomplete cataract, posterior sutures            | 0                |       | 1                | 0.0%  |
| 100.326 incomplete cataract, nucleus                      | 0                |       | 2                | 0.1%  |
| 100.327 incomplete cataract, capsular                     | 3                | 0.0%  | 2                | 0.1%  |
| 100.328 y-suture tip opacities                            | 6                | 0.0%  | 21               | 0.7%  |
| 100.330 generalized/complete cataract                     | 48               | 0.3%  | 2                | 0.1%  |
| 100.345 <i>significant cataracts (summary)</i>            | 1,663            | 11.3% | 335              | 11.2% |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 3                | 0.0%  | 0                |       |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 21               | 0.1%  | 12               | 0.4%  |
| 110.135 PHPV/PTVL   | 7                | 0.0%  | 1                | 0.0%  |
| 110.200 vitreous degeneration-anterior chamber            | 2                | 0.0%  | 4                | 0.1%  |
| 110.320 vitreal degeneration                              | 66               | 0.4%  | 7                | 0.2%  |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 124              | 0.8%  | 27               | 0.9%  |
| 120.180 retinal dysplasia, geographic                     | 43               | 0.3%  | 11               | 0.4%  |
| 120.190 retinal dysplasia, detached                       | 1                | 0.0%  | 1                | 0.0%  |
| 120.310 generalized progressive retinal atrophy (PRA)     | 174              | 1.2%  | 9                | 0.3%  |
| 120.910 retinal detachment without dialysis               | 1                | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 1                | 0.0%  | 0                |       |
| 120.960 retinopathy                                       | 16               | 0.1%  | 13               | 0.4%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 15               | 0.1%  | 4                | 0.1%  |
| 130.120 optic nerve hypoplasia                            | 17               | 0.1%  | 1                | 0.0%  |
| 130.150 optic disc coloboma                               | 2                | 0.0%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 137              | 0.9%  | 0                |       |
| 900.100 other, not inherited                              | 330              | 2.2%  | 11               | 0.4%  |
| 900.110 other. suspect not inherited/significance unknown | 227              | 1.5%  | 163              | 5.5%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 11,699           | 79.6% | 2,065            | 69.1% |

# RUSSELL TERRIER

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|--|---------------------|-----------|-----------------|--------------------------------------|
| A. | Distichiasis                                     | Not defined         | 1         | Breeder option  |                                      |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option  |                                      |
| C. | Cataract   | Not defined         | 1         | NO              |                                      |
| D. | Lens luxation                                    | Autosomal recessive | 2         | NO              | Mutation in the <i>ADAMTS17</i> gene |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### D. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing

vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011; 14: 378-384.

# OCULAR DISORDERS REPORT RUSSELL TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED  |  | 1991-2015<br>267 |       | 2016-2020<br>284 |       |
|---------------------|--|--|------------------|-------|------------------|-------|
|                     |  |  | #                | %     | #                | %     |
| <b>EYELIDS</b>      |  |  |                  |       |                  |       |
| 25.110              | distichiasis   |  | 10               | 3.7%  | 8                | 2.8%  |
| <b>NASOLACRIMAL</b> |  |  |                  |       |                  |       |
| 32.110              | imperforate lower nasolacrimal punctum                       |  | 0                |       | 1                | 0.4%  |
| 40.910              | keratoconjunctivitis sicca                                   |  | 1                | 0.4%  | 0                |       |
| <b>CORNEA</b>       |  |  |                  |       |                  |       |
| 70.700              | corneal dystrophy  |  | 0                |       | 1                | 0.4%  |
| <b>UVEA</b>         |  |  |                  |       |                  |       |
| 93.150              | iris coloboma  |  | 1                | 0.4%  | 0                |       |
| 93.180              | iris sphincter dysplasia                                     |  | 1                | 0.4%  | 0                |       |
| 93.710              | persistent pupillary membranes, iris to iris                 |  | 10               | 3.7%  | 20               | 7.0%  |
| 93.720              | persistent pupillary membranes, iris to lens                 |  | 0                |       | 1                | 0.4%  |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands |  | 1                | 0.4%  | 1                | 0.4%  |
| 93.999              | uveal cysts  |  | 1                | 0.4%  | 0                |       |
| <b>LENS</b>         |  |  |                  |       |                  |       |
| 100.210             | cataract. suspect not inherited/significance unknown         |  | 8                | 3.0%  | 22               | 7.7%  |
| 100.305             | punctate cataract, posterior sutures                         |  | 0                |       | 1                | 0.4%  |
| 100.306             | punctate cataract, nucleus                                   |  | 0                |       | 1                | 0.4%  |
| 100.307             | punctate cataract, capsular                                  |  | 0                |       | 1                | 0.4%  |
| 100.321             | incomplete cataract, anterior cortex                         |  | 0                |       | 1                | 0.4%  |
| 100.322             | incomplete cataract, posterior cortex                        |  | 2                | 0.7%  | 2                | 0.7%  |
| 100.323             | incomplete cataract, equatorial cortex                       |  | 0                |       | 1                | 0.4%  |
| 100.325             | incomplete cataract, posterior sutures                       |  | 0                |       | 1                | 0.4%  |
| 100.328             | y-suture tip opacities                                       |  | 0                |       | 1                | 0.4%  |
| 100.345             | significant cataracts (summary)                              |  | 2                | 0.7%  | 9                | 3.2%  |
| <b>VITREOUS</b>     |  |  |                  |       |                  |       |
| 110.120             | persistent hyaloid artery/remnant                            |  | 0                |       | 2                | 0.7%  |
| <b>RETINA</b>       |  |  |                  |       |                  |       |
| 120.170             | retinal dysplasia, folds                                     |  | 2                | 0.7%  | 1                | 0.4%  |
| 120.180             | retinal dysplasia, geographic                                |  | 1                | 0.4%  | 0                |       |
| 120.310             | generalized progressive retinal atrophy (PRA)                |  | 1                | 0.4%  | 0                |       |
| <b>OPTIC NERVE</b>  |  |  |                  |       |                  |       |
| 130.110             | micropapilla   |  | 1                | 0.4%  | 0                |       |
| <b>OTHER</b>        |  |  |                  |       |                  |       |
| 900.000             | other, unspecified   |  | 2                | 0.7%  | 0                |       |
| 900.110             | other. suspect not inherited/significance unknown            |  | 7                | 2.6%  | 14               | 4.9%  |
| <b>NORMAL</b>       |  |  |                  |       |                  |       |
| 0.000               | normal globe   |  | 230              | 86.1% | 220              | 77.5% |

# RUSSIAN TOY

|    | <b>DISORDER</b>  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b>  |
|----|--|--------------------|------------------|-------------------------|
| A. | Persistent pupillary membranes<br>- lens pigment foci/no strands | Not defined        | 1                | Passes with no notation |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT RUSSIAN TOY

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 1         | 1.9%  | 0         |       |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.700          | corneal dystrophy  | 1         | 1.9%  | 0         |       |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                   | 1         | 1.9%  | 5         | 9.4%  |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands   | 1         | 1.9%  | 7         | 13.2% |
| 93.760          | persistent pupillary membranes, endothelial opacity/no strands | 1         | 1.9%  | 0         |       |
| 97.150          | chorioretinal coloboma, congenital                             | 1         | 1.9%  | 0         |       |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown           | 1         | 1.9%  | 2         | 3.8%  |
| 100.301         | punctate cataract, anterior cortex                             | 3         | 5.8%  | 0         |       |
| 100.303         | punctate cataract, equatorial cortex                           | 1         | 1.9%  | 0         |       |
| 100.305         | punctate cataract, posterior sutures                           | 0         |       | 1         | 1.9%  |
| 100.307         | punctate cataract, capsular                                    | 0         |       | 2         | 3.8%  |
| 100.328         | y-suture tip opacities   | 0         |       | 1         | 1.9%  |
| 100.345         | <i>significant cataracts (summary)</i>                         | 4         | 7.7%  | 4         | 7.5%  |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.120         | persistent hyaloid artery/remnant                              | 1         | 1.9%  | 0         |       |
| 110.200         | vitreal degeneration-anterior chamber                          | 0         |       | 1         | 1.9%  |
| 110.320         | vitreal degeneration   | 4         | 7.7%  | 2         | 3.8%  |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.180         | retinal dysplasia, geographic                                  | 0         |       | 1         | 1.9%  |
| 120.190         | retinal dysplasia, detached                                    | 0         |       | 1         | 1.9%  |
| 120.960         | retinopathy  | 1         | 1.9%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.000         | other, unspecified   | 2         | 3.8%  | 0         |       |
| 900.110         | other. suspect not inherited/significance unknown              | 3         | 5.8%  | 0         |       |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 41        | 78.8% | 35        | 66.0% |



## Russian Tsvetnaya Bolonka (Bolonka Zvetna)

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC<br>TEST                 |
|----|------------------------------------|------------------------|-----------|--------------------|---------------------------------|
| A. | Cataract                           | Not defined            | 1         | NO                 |                                 |
| B. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 2         | NO                 | Mutation in<br><i>prcd</i> gene |

### Description and Comments

#### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

#### B. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Russian Tsvetnaya Bolonka is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. However in the American Eskimo Dog the phenotype can be very variable in the age of onset. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

Other forms of retinal degeneration that are not *prcd* are recognized in the breed. The currently available genetic test will not detect these other forms of PRA.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006 Nov;88:551-563. PMID: 16938425

# OCULAR DISORDERS REPORT RUSSIAN TSVETNAYA BOLONKA

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>90 |       | 2016-2020<br>33 |       |
|---|---------------------|-----------------|-------|-----------------|-------|
|   |                     | #               | %     | #               | %     |
| <b>EYELIDS</b>  |                     |                 |       |                 |       |
| 25.110 distichiasis   |                     | 1               | 1.1%  | 0               |       |
| <b>NASOLACRIMAL</b>   |                     |                 |       |                 |       |
| 40.910 keratoconjunctivitis sicca                                   |                     | 1               | 1.1%  | 0               |       |
| <b>CORNEA</b>   |                     |                 |       |                 |       |
| 70.220 pigmentary keratitis   |                     | 2               | 2.2%  | 0               |       |
| <b>UVEA</b>   |                     |                 |       |                 |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 1               | 1.1%  | 0               |       |
| <b>LENS</b>   |                     |                 |       |                 |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 4               | 4.4%  | 1               | 3.0%  |
| 100.301 punctate cataract, anterior cortex                          |                     | 0               |       | 2               | 6.1%  |
| 100.313 incipient cataract, equatorial cortex                       |                     | 1               | 1.1%  | 0               |       |
| 100.315 incipient cataract, posterior sutures                       |                     | 0               |       | 3               | 9.1%  |
| 100.317 incipient cataract, capsular                                |                     | 0               |       | 1               | 3.0%  |
| 100.328 y-suture tip opacities                                      |                     | 0               |       | 3               | 9.1%  |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 1               | 1.1%  | 9               | 27.3% |
| 100.375 <i>subluxation/luxation, unspecified</i>                    |                     | 1               | 1.1%  | 0               |       |
| <b>VITREOUS</b>   |                     |                 |       |                 |       |
| 110.135 PHPV/PTVL   |                     | 1               | 1.1%  | 0               |       |
| 110.200 vitreous degeneration-anterior chamber                      |                     | 2               | 2.2%  | 2               | 6.1%  |
| 110.320 vitreal degeneration  |                     | 10              | 11.1% | 3               | 9.1%  |
| <b>OTHER</b>  |                     |                 |       |                 |       |
| 900.000 other, unspecified  |                     | 1               | 1.1%  | 0               |       |
| 900.100 other, not inherited  |                     | 2               | 2.2%  | 0               |       |
| 900.110 other. suspect not inherited/significance unknown           |                     | 0               |       | 4               | 12.1% |
| <b>NORMAL</b>   |                     |                 |       |                 |       |
| 0.000 normal globe  |                     | 77              | 85.6% | 19              | 57.6% |

## RUSSO-EUROPEAN LAIKA

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|------------------------------------|------------------------|-----------|--------------------|-------------------------------------|
| A. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>prcd</i> gene |

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### Description and Comments

#### A. Retinal atrophy – *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Russo-European Laika is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

### References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Russo-European Laika. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Zangerl, B., et al. (2006). "Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans." *Genomics* **88**(5): 551-563. PMID: 16938425

# SAMOYED

|    | DISORDER  | INHERITANCE  | REFERENCE | BREEDING<br>ADVICE  | GENETIC TESTS<br>AVAILABLE            |
|----|---|--|-----------|---|---------------------------------------|
| A. | Glaucoma  | Not defined  | 1-7       | NO  |                                       |
| B. | Distichiasis  | Not defined  | 1         | Breeder option  |                                       |
| C. | Corneal dystrophy -<br>epithelial/stromal                                       | Not defined  | 1, 8      | Breeder option  |                                       |
| D. | Persistent pupillary<br>membranes<br>- iris to iris                             | Not defined  | 1         | Breeder option  |                                       |
| E. | Cataract  | Not defined  | 1         | NO  |                                       |
| F. | Retinal atrophy<br>( <i>RPGR</i> )  | X-linked<br>recessive  | 1, 9, 10  | NO  | Mutation in the<br><i>RPGR</i> gene   |
| G. | Retinal dysplasia<br>- folds  | Presumed<br>autosomal<br>recessive                                     | 1, 11, 12 | NO<br>(Breeder option<br>with Normal DNA<br>test for folds) | Mutation in the<br><i>COL9A2</i> gene |
| H. | Retinal dysplasia<br>- folds/geographic/<br>detached (with<br>skeletal defects) | Autosomal<br>recessive with<br>incomplete<br>dominance for<br>the eyes | 1, 11-14  | NO  | Mutation in the<br><i>COL9A2</i> gene |
| I. | Uveodermatologic<br>syndrome  | Not defined  | 1, 15, 16 | NO  |                                       |

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## Description and Comments

### A. Glaucoma

An elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine breed eye screening exam.

B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

In the Samoyed, many of the PPMs identified on routine screening examinations bridge from the iris to the cornea where they may be associated with corneal opacity and vision impairment.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

F. Retinal atrophy - *RPGR*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. In the Samoyed, one form of PRA, known as XLPRA1, is due to a mutation in the *RPGR* gene and is inherited as a recessive, sex-linked trait. A DNA test is available.

G. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness.

In the Samoyed, the presence of retinal folds may be seen in the heterozygous state described in "I" below. Thus the recommendation against breeding. The breeding advice for

Labrador Retrievers and Samoyeds diagnosed with "retinal dysplasia - folds" will be changed from "No" to "Breeder option" if the owner of the dog provides the registering office with results of the DNA test for the affected dog showing that it is not a carrier of the *COL9A2* mutation.

#### H. Retinal dysplasia - folds with skeletal defects in homozygous affected dogs

This condition is also known as oculo-skeletal dysplasia (OSD) or dwarfism with retinal dysplasia type 2 (DRD2) in the Samoyed. A similar condition, DRD1, occurs in the Labrador Retriever. The condition is autosomal recessive and homozygous affected dogs have shortened forelimbs ("downhill" conformation) with valgus deformity. They have severe ocular defects including cataract, retinal folds/multifocal retinal dysplasia, vitreal degeneration and retinal detachment. The ocular abnormalities result in blindness in most dogs. Heterozygous dogs can have either a normal ocular exam or have multiple retinal folds, vitreal membranes, or vitreal degeneration suggesting a semi-dominant mechanism with respect to the eyes. It is important to note that generally the retinal folds present in heterozygous dogs tend to cluster around the major superior blood vessels of the central tapetal region. The condition is caused by a 1,267 bp deletion of *COL9A2*. A DNA test is available.

#### I. Uveodermatologic syndrome

Uveodermatologic syndrome in the Samoyed bears many similarities to a condition in people called Vogt-Koyanagi-Harada (or VKH) syndrome. Thus, the condition in dogs is often referred to as VKH or VKH-like syndrome. It is an immune-mediated disease in which pigmented cells (melanocytes) in the eye and in the skin are destroyed by white blood cells (lymphocytes). The first clinical signs are usually inflammation of the intraocular structures (or uveitis) in both eyes. Adhesions between the iris and lens (posterior synechiae) and the peripheral iris and cornea (peripheral anterior synechiae) develop rapidly. Other complications include cataract development, retinal degeneration, retinal separation or detachment, optic disc atrophy and secondary glaucoma. The uveitis is very difficult to control medically and ultimately results in blindness in most affected dogs. Whitening of the hair (poliosis) and skin (vitiligo) may also be noted in advanced cases. Some veterinary ophthalmologists feel there is a prevalence of this entity in the Samoyed. Additional studies are needed to validate this experience and explore the possibility of a genetic basis.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
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# OCULAR DISORDERS REPORT SAMOYED

| Diagnostic Name   | TOTAL DOGS EXAMINED |      | 1991-2015<br>23,045 |      | 2016-2020<br>5,526 |   |
|---|---------------------|------|---------------------|------|--------------------|---|
|   | #                   | %    | #                   | %    | #                  | % |
| <b>GLOBE</b>  |                     |      |                     |      |                    |   |
| 0.110 microphthalmia  | 20                  | 0.1% | 3                   | 0.1% |                    |   |
| 10.000 glaucoma   | 10                  | 0.0% | 1                   | 0.0% |                    |   |
| <b>EYELIDS</b>  |                     |      |                     |      |                    |   |
| 20.140 ectopic cilia  | 7                   | 0.0% | 2                   | 0.0% |                    |   |
| 20.160 macropalpebral fissure   | 1                   | 0.0% | 0                   |      |                    |   |
| 21.000 entropion, unspecified   | 6                   | 0.0% | 0                   |      |                    |   |
| 22.000 ectropion, unspecified   | 3                   | 0.0% | 1                   | 0.0% |                    |   |
| 25.110 distichiasis   | 1,331               | 5.8% | 226                 | 4.1% |                    |   |
| <b>NASOLACRIMAL</b>   |                     |      |                     |      |                    |   |
| 32.110 imperforate lower nasolacrimal punctum                         | 10                  | 0.0% | 15                  | 0.3% |                    |   |
| 40.910 keratoconjunctivitis sicca                                     | 14                  | 0.1% | 3                   | 0.1% |                    |   |
| <b>NICTITANS</b>  |                     |      |                     |      |                    |   |
| 51.100 third eyelid cartilage anomaly                                 | 4                   | 0.0% | 1                   | 0.0% |                    |   |
| <b>CORNEA</b>   |                     |      |                     |      |                    |   |
| 70.210 corneal pannus   | 4                   | 0.0% | 0                   |      |                    |   |
| 70.220 pigmentary keratitis   | 1                   | 0.0% | 1                   | 0.0% |                    |   |
| 70.700 corneal dystrophy  | 793                 | 3.4% | 235                 | 4.3% |                    |   |
| 70.730 corneal endothelial degeneration                               | 15                  | 0.1% | 2                   | 0.0% |                    |   |
| <b>UVEA</b>   |                     |      |                     |      |                    |   |
| 93.140 corneal endothelial pigment without PPM                        | 1                   | 0.0% | 0                   |      |                    |   |
| 93.150 iris coloboma  | 1                   | 0.0% | 0                   |      |                    |   |
| 93.710 persistent pupillary membranes, iris to iris                   | 456                 | 2.0% | 126                 | 2.3% |                    |   |
| 93.720 persistent pupillary membranes, iris to lens                   | 24                  | 0.1% | 9                   | 0.2% |                    |   |
| 93.730 persistent pupillary membranes, iris to cornea                 | 35                  | 0.2% | 8                   | 0.1% |                    |   |
| 93.740 persistent pupillary membranes, iris sheets                    | 16                  | 0.1% | 0                   |      |                    |   |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   | 10                  | 0.0% | 6                   | 0.1% |                    |   |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands | 12                  | 0.1% | 2                   | 0.0% |                    |   |
| 93.810 uveal melanoma   | 1                   | 0.0% | 0                   |      |                    |   |
| 93.999 uveal cysts  | 12                  | 0.1% | 4                   | 0.1% |                    |   |
| 97.150 chorioretinal coloboma, congenital                             | 0                   |      | 3                   | 0.1% |                    |   |
| <b>LENS</b>   |                     |      |                     |      |                    |   |
| 100.200 cataract, unspecified   | 100                 | 0.4% | 0                   |      |                    |   |
| 100.210 cataract. suspect not inherited/significance unknown          | 771                 | 3.3% | 158                 | 2.9% |                    |   |
| 100.301 punctate cataract, anterior cortex                            | 66                  | 0.3% | 25                  | 0.5% |                    |   |
| 100.302 punctate cataract, posterior cortex                           | 147                 | 0.6% | 21                  | 0.4% |                    |   |
| 100.303 punctate cataract, equatorial cortex                          | 14                  | 0.1% | 2                   | 0.0% |                    |   |
| 100.304 punctate cataract, anterior sutures                           | 8                   | 0.0% | 3                   | 0.1% |                    |   |
| 100.305 punctate cataract, posterior sutures                          | 58                  | 0.3% | 12                  | 0.2% |                    |   |
| 100.306 punctate cataract, nucleus                                    | 17                  | 0.1% | 3                   | 0.1% |                    |   |
| 100.307 punctate cataract, capsular                                   | 22                  | 0.1% | 20                  | 0.4% |                    |   |
| 100.311 incipient cataract, anterior cortex                           | 76                  | 0.3% | 28                  | 0.5% |                    |   |
| 100.312 incipient cataract, posterior cortex                          | 251                 | 1.1% | 46                  | 0.8% |                    |   |
| 100.313 incipient cataract, equatorial cortex                         | 23                  | 0.1% | 7                   | 0.1% |                    |   |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.314 incipient cataract, anterior sutures              | 7                | 0.0%  | 0                |       |
| 100.315 incipient cataract, posterior sutures             | 51               | 0.2%  | 7                | 0.1%  |
| 100.316 incipient cataract, nucleus                       | 33               | 0.1%  | 5                | 0.1%  |
| 100.317 incipient cataract, capsular                      | 28               | 0.1%  | 15               | 0.3%  |
| 100.321 incomplete cataract, anterior cortex              | 0                |       | 2                | 0.0%  |
| 100.322 incomplete cataract, posterior cortex             | 8                | 0.0%  | 16               | 0.3%  |
| 100.325 incomplete cataract, posterior sutures            | 1                | 0.0%  | 2                | 0.0%  |
| 100.326 incomplete cataract, nucleus                      | 1                | 0.0%  | 1                | 0.0%  |
| 100.327 incomplete cataract, capsular                     | 3                | 0.0%  | 4                | 0.1%  |
| 100.328 y-suture tip opacities                            | 5                | 0.0%  | 14               | 0.3%  |
| 100.330 generalized/complete cataract                     | 66               | 0.3%  | 0                |       |
| 100.340 resorbing/hypermature cataract                    | 1                | 0.0%  | 2                | 0.0%  |
| 100.345 <i>significant cataracts (summary)</i>            | 986              | 4.3%  | 235              | 4.3%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 3                | 0.0%  | 1                | 0.0%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 22               | 0.1%  | 7                | 0.1%  |
| 110.135 PHPV/PTVL   | 11               | 0.0%  | 3                | 0.1%  |
| 110.200 vitreous degeneration-anterior chamber            | 0                |       | 2                | 0.0%  |
| 110.320 vitreal degeneration                              | 94               | 0.4%  | 7                | 0.1%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 4                | 0.0%  | 0                |       |
| 97.120 coloboma   | 7                | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 483              | 2.1%  | 82               | 1.5%  |
| 120.180 retinal dysplasia, geographic                     | 168              | 0.7%  | 47               | 0.9%  |
| 120.190 retinal dysplasia, detached                       | 26               | 0.1%  | 6                | 0.1%  |
| 120.310 generalized progressive retinal atrophy (PRA)     | 56               | 0.2%  | 0                |       |
| 120.400 retinal hemorrhage                                | 2                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 10               | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 0                |       | 2                | 0.0%  |
| 120.960 retinopathy                                       | 2                | 0.0%  | 8                | 0.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 18               | 0.1%  | 1                | 0.0%  |
| 130.120 optic nerve hypoplasia                            | 13               | 0.1%  | 2                | 0.0%  |
| 130.150 optic disc coloboma                               | 70               | 0.3%  | 5                | 0.1%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 176              | 0.8%  | 0                |       |
| 900.100 other, not inherited                              | 447              | 1.9%  | 9                | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown | 225              | 1.0%  | 201              | 3.6%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 18,959           | 82.3% | 4,344            | 78.6% |

# SCHAPENDOES

|    | DISORDER                             | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE            |
|----|--------------------------------------|------------------------|-----------|--------------------|---------------------------------------|
| A. | Retinal atrophy<br>( <i>CCDC66</i> ) | Autosomal<br>recessive | 1, 2      | NO                 | Mutation in the<br><i>CCDC66</i> gene |

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## Description and Comments

### A. Retinal atrophy - *CCDC66*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

In the Schapendoes the age of onset is between 2-5 years of age. The causal mutation is a single base pair insertion in exon 6 of the gene coiled-coil domain containing 66 (*CCDC66*) that leads to a stop codon. The mutation is inherited as an autosomal recessive trait. A DNA test is available.

## References

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# OCULAR DISORDERS REPORT SCHAPENDOES

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | 76        |       | 47        |       |
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 1         | 1.3%  | 1         | 2.1%  |
| <b>CORNEA</b>   |  |           |       |           |       |
| 70.700          | corneal dystrophy  | 0         |       | 1         | 2.1%  |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 0         |       | 1         | 2.1%  |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 1         | 1.3%  | 1         | 2.1%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 3         | 3.9%  | 4         | 8.5%  |
| 100.301         | punctate cataract, anterior cortex                           | 1         | 1.3%  | 1         | 2.1%  |
| 100.311         | incipient cataract, anterior cortex                          | 0         |       | 2         | 4.3%  |
| 100.312         | incipient cataract, posterior cortex                         | 0         |       | 2         | 4.3%  |
| 100.315         | incipient cataract, posterior sutures                        | 1         | 1.3%  | 1         | 2.1%  |
| 100.328         | y-suture tip opacities                                       | 1         | 1.3%  | 2         | 4.3%  |
| 100.345         | <i>significant cataracts (summary)</i>                       | 3         | 3.9%  | 8         | 17.0% |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.120         | persistent hyaloid artery/remnant                            | 2         | 2.6%  | 1         | 2.1%  |
| 110.320         | vitreal degeneration   | 1         | 1.3%  | 0         |       |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.180         | retinal dysplasia, geographic                                | 1         | 1.3%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.100         | other, not inherited   | 6         | 7.9%  | 0         |       |
| 900.110         | other. suspect not inherited/significance unknown            | 0         |       | 2         | 4.3%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 65        | 85.5% | 30        | 63.8% |

# SCHIPPERKE

|    | DISORDER   | INHERITANCE                  | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE          |
|----|--|------------------------------|-----------|-----------------|----------------------------------|
| A. | Distichiasis                                     | Not defined                  | 1         | Breeder option  |                                  |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined                  | 1         | Breeder option  |                                  |
| C. | Cataract   | Not defined                  | 1         | NO              |                                  |
| D. | Vitreous degeneration                            | Not defined                  | 1         | Breeder option  |                                  |
| E. | Retinal atrophy - generalized ( <i>prcd</i> )    | Presumed autosomal recessive | 1, 2      | NO              | Mutation in the <i>prcd</i> gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

E. Retinal atrophy – generalized (*prcd*)

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Schipperke is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Zangerl, B., et al. (2006). "Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans." Genomics **88**(5): 551-563. PMID: 16938425

# OCULAR DISORDERS REPORT SCHIPPERKE

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 0.110 microphthalmia  |                     | 1         | 0.1% | 0         |      |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 25.110 distichiasis   |                     | 46        | 3.2% | 5         | 2.1% |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.210 corneal pannus   |                     | 1         | 0.1% | 0         |      |
| 70.220 pigmentary keratitis   |                     | 0         |      | 1         | 0.4% |
| 70.700 corneal dystrophy  |                     | 3         | 0.2% | 0         |      |
| 70.730 corneal endothelial degeneration                             |                     | 2         | 0.1% | 0         |      |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 126       | 8.6% | 21        | 8.7% |
| 93.720 persistent pupillary membranes, iris to lens                 |                     | 6         | 0.4% | 0         |      |
| 93.730 persistent pupillary membranes, iris to cornea               |                     | 2         | 0.1% | 0         |      |
| 93.740 persistent pupillary membranes, iris sheets                  |                     | 10        | 0.7% | 0         |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 7         | 0.5% | 1         | 0.4% |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.200 cataract, unspecified                                       |                     | 4         | 0.3% | 0         |      |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 62        | 4.3% | 13        | 5.4% |
| 100.301 punctate cataract, anterior cortex                          |                     | 13        | 0.9% | 2         | 0.8% |
| 100.302 punctate cataract, posterior cortex                         |                     | 1         | 0.1% | 0         |      |
| 100.303 punctate cataract, equatorial cortex                        |                     | 5         | 0.3% | 0         |      |
| 100.304 punctate cataract, anterior sutures                         |                     | 1         | 0.1% | 0         |      |
| 100.305 punctate cataract, posterior sutures                        |                     | 1         | 0.1% | 0         |      |
| 100.306 punctate cataract, nucleus                                  |                     | 7         | 0.5% | 1         | 0.4% |
| 100.311 incipient cataract, anterior cortex                         |                     | 20        | 1.4% | 2         | 0.8% |
| 100.312 incipient cataract, posterior cortex                        |                     | 10        | 0.7% | 0         |      |
| 100.313 incipient cataract, equatorial cortex                       |                     | 8         | 0.5% | 1         | 0.4% |
| 100.315 incipient cataract, posterior sutures                       |                     | 1         | 0.1% | 0         |      |
| 100.316 incipient cataract, nucleus                                 |                     | 5         | 0.3% | 1         | 0.4% |
| 100.317 incipient cataract, capsular                                |                     | 1         | 0.1% | 1         | 0.4% |
| 100.321 incomplete cataract, anterior cortex                        |                     | 1         | 0.1% | 0         |      |
| 100.322 incomplete cataract, posterior cortex                       |                     | 0         |      | 1         | 0.4% |
| 100.330 generalized/complete cataract                               |                     | 8         | 0.5% | 0         |      |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 86        | 5.9% | 9         | 3.7% |
| <b>VITREOUS</b>   |                     |           |      |           |      |
| 110.120 persistent hyaloid artery/remnant                           |                     | 1         | 0.1% | 0         |      |
| 110.135 PHPV/PTVL   |                     | 1         | 0.1% | 0         |      |
| 110.200 vitreous degeneration-anterior chamber                      |                     | 1         | 0.1% | 0         |      |
| 110.320 vitreal degeneration  |                     | 18        | 1.2% | 5         | 2.1% |
| <b>RETINA</b>   |                     |           |      |           |      |
| 120.170 retinal dysplasia, folds                                    |                     | 9         | 0.6% | 2         | 0.8% |
| 120.180 retinal dysplasia, geographic                               |                     | 4         | 0.3% | 0         |      |
| 120.310 generalized progressive retinal atrophy (PRA)               |                     | 16        | 1.1% | 1         | 0.4% |
| 120.920 retinal detachment with dialysis                            |                     | 0         |      | 1         | 0.4% |
| 120.960 retinopathy   |                     | 2         | 0.1% | 0         |      |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 16 1.1%     | 0         |
| 900.100 other, not inherited                              | 51 3.5%     | 0         |
| 900.110 other. suspect not inherited/significance unknown | 16 1.1%     | 18 7.5%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,170 80.2% | 182 75.5% |



# SCOTTISH TERRIER

|    | DISORDER                         | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|----------------------------------|-------------|-----------|-------------------------|
| A. | Persistent pupillary membranes   |             |           |                         |
|    | - iris to iris                   | Not defined | 1         | Breeder option          |
|    | - iris to lens                   | Not defined | 1         | NO                      |
|    | - lens pigment foci/no strands   | Not defined | 1         | Passes with no notation |
|    | - endothelial opacity/no strands | Not defined | 1         | NO                      |
| B. | Cataract                         | Not defined | 1         | NO                      |
| C. | Ligneous conjunctivitis          | Not defined | 2, 3      | NO                      |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Ligneous conjunctivitis

A rare type of conjunctivitis characterized by the formation of thick membranes covering conjunctiva of the nictitans and eyelids of affected dogs. This condition has been diagnosed in four unrelated Doberman Pinschers, three of which had life-threatening systemic disease. Ligneous conjunctivitis has also been reported in one Yorkshire Terrier.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Ramsey DT, Ketring K, Glaze MB, et al. Ligneous conjunctivitis in four Doberman Pinschers. *J Am Anim Hosp Assoc.* 1996; 32: 439-447.
3. Mason SL, McElroy P, Nuttall T. Ligneous membranitis in Scottish Terriers. *Vet Rec.* 2012; 171: 160.

# OCULAR DISORDERS REPORT SCOTTISH TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|---------------------|--|-----------|-------|-----------|-------|
|                     |  | #         | %     | #         | %     |
| <b>EYELIDS</b>      |  |           |       |           |       |
| 25.110              | distichiasis   | 3         | 0.4%  | 0         |       |
| <b>NASOLACRIMAL</b> |  |           |       |           |       |
| 40.910              | keratoconjunctivitis sicca                                     | 1         | 0.1%  | 0         |       |
| <b>NICTITANS</b>    |  |           |       |           |       |
| 52.110              | prolapsed gland of the third eyelid                            | 2         | 0.3%  | 0         |       |
| <b>CORNEA</b>       |  |           |       |           |       |
| 70.210              | corneal pannus   | 1         | 0.1%  | 0         |       |
| 70.220              | pigmentary keratitis   | 2         | 0.3%  | 0         |       |
| 70.700              | corneal dystrophy  | 5         | 0.7%  | 1         | 0.5%  |
| 70.730              | corneal endothelial degeneration                               | 2         | 0.3%  | 0         |       |
| <b>UVEA</b>         |  |           |       |           |       |
| 93.140              | corneal endothelial pigment without PPM                        | 3         | 0.4%  | 0         |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 227       | 30.0% | 49        | 26.5% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 38        | 5.0%  | 5         | 2.7%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 9         | 1.2%  | 2         | 1.1%  |
| 93.740              | persistent pupillary membranes, iris sheets                    | 3         | 0.4%  | 0         |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 57        | 7.5%  | 71        | 38.4% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 4         | 0.5%  | 6         | 3.2%  |
| <b>LENS</b>         |  |           |       |           |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 73        | 9.7%  | 10        | 5.4%  |
| 100.301             | punctate cataract, anterior cortex                             | 7         | 0.9%  | 0         |       |
| 100.302             | punctate cataract, posterior cortex                            | 2         | 0.3%  | 0         |       |
| 100.303             | punctate cataract, equatorial cortex                           | 2         | 0.3%  | 0         |       |
| 100.304             | punctate cataract, anterior sutures                            | 2         | 0.3%  | 0         |       |
| 100.305             | punctate cataract, posterior sutures                           | 1         | 0.1%  | 0         |       |
| 100.306             | punctate cataract, nucleus                                     | 3         | 0.4%  | 0         |       |
| 100.307             | punctate cataract, capsular                                    | 2         | 0.3%  | 1         | 0.5%  |
| 100.311             | incipient cataract, anterior cortex                            | 6         | 0.8%  | 1         | 0.5%  |
| 100.312             | incipient cataract, posterior cortex                           | 5         | 0.7%  | 1         | 0.5%  |
| 100.313             | incipient cataract, equatorial cortex                          | 3         | 0.4%  | 0         |       |
| 100.314             | incipient cataract, anterior sutures                           | 1         | 0.1%  | 0         |       |
| 100.315             | incipient cataract, posterior sutures                          | 1         | 0.1%  | 1         | 0.5%  |
| 100.316             | incipient cataract, nucleus                                    | 9         | 1.2%  | 0         |       |
| 100.317             | incipient cataract, capsular                                   | 2         | 0.3%  | 1         | 0.5%  |
| 100.321             | incomplete cataract, anterior cortex                           | 0         |       | 1         | 0.5%  |
| 100.322             | incomplete cataract, posterior cortex                          | 0         |       | 1         | 0.5%  |
| 100.326             | incomplete cataract, nucleus                                   | 0         |       | 1         | 0.5%  |
| 100.327             | incomplete cataract, capsular                                  | 0         |       | 1         | 0.5%  |
| 100.330             | generalized/complete cataract                                  | 4         | 0.5%  | 1         | 0.5%  |
| 100.345             | significant cataracts (summary)                                | 50        | 6.6%  | 10        | 5.4%  |
| 100.375             | subluxation/luxation, unspecified                              | 1         | 0.1%  | 0         |       |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>VITREOUS</b>   |           |           |
| 110.120 persistent hyaloid artery/remnant                 | 1 0.1%    | 0         |
| 110.320 vitreal degeneration                              | 5 0.7%    | 0         |
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 5 0.7%    | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 8 1.1%    | 0         |
| <b>OPTIC NERVE</b>  |           |           |
| 130.150 optic disc coloboma                               | 2 0.3%    | 0         |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 13 1.7%   | 0         |
| 900.100 other, not inherited                              | 60 7.9%   | 0         |
| 900.110 other. suspect not inherited/significance unknown | 19 2.5%   | 9 4.9%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 398 52.6% | 71 38.4%  |

# SEALYHAM TERRIER

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|----|--|---------------------|-----------|-----------------|--------------------------------------|
| A. | Distichiasis                                     | Not defined         | 1         | Breeder option  |                                      |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option  |                                      |
| C. | Cataract   | Not defined         | 1         | NO              |                                      |
| D. | Lens luxation                                    | Autosomal recessive | 1-5       | NO              | Mutation in the <i>ADAMTS17</i> gene |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### D. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to

be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Formston C. Observations on subluxation and luxation of the crystalline lens in the dog. *J Comp Pathol.* 1945;55:168-186.
3. Hodgman SFJ. Abnormalities and defects in pedigree dogs: I. An investigation into the existence of abnormalities in pedigree dogs in British Isles. *J Small Anim Pract.* 1963;4:447-456.
4. Curtis R, Barnett KC. Primary lens luxation in the dog. *J Small Anim Pract.* 1980;21:657-668.
5. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011;14:378-384.

# OCULAR DISORDERS REPORT SEALYHAM TERRIER

| Diagnostic Name    | TOTAL DOGS EXAMINED  | 1991-2015 |      | 2016-2020 |       |
|--------------------|--|-----------|------|-----------|-------|
|                    |  | #         | %    | #         | %     |
| <b>EYELIDS</b>     |  |           |      |           |       |
| 25.110             | distichiasis   | 27        | 5.5% | 1         | 2.7%  |
| <b>NICTITANS</b>   |  |           |      |           |       |
| 52.110             | prolapsed gland of the third eyelid                            | 1         | 0.2% | 0         |       |
| <b>UVEA</b>        |  |           |      |           |       |
| 93.710             | persistent pupillary membranes, iris to iris                   | 31        | 6.4% | 6         | 16.2% |
| 93.720             | persistent pupillary membranes, iris to lens                   | 2         | 0.4% | 0         |       |
| 93.730             | persistent pupillary membranes, iris to cornea                 | 1         | 0.2% | 0         |       |
| 93.740             | persistent pupillary membranes, iris sheets                    | 2         | 0.4% | 0         |       |
| 93.750             | persistent pupillary membranes, lens pigment foci/no strands   | 1         | 0.2% | 2         | 5.4%  |
| 93.760             | persistent pupillary membranes, endothelial opacity/no strands | 1         | 0.2% | 0         |       |
| <b>LENS</b>        |  |           |      |           |       |
| 100.200            | cataract, unspecified  | 2         | 0.4% | 0         |       |
| 100.210            | cataract. suspect not inherited/significance unknown           | 20        | 4.1% | 1         | 2.7%  |
| 100.301            | punctate cataract, anterior cortex                             | 4         | 0.8% | 0         |       |
| 100.302            | punctate cataract, posterior cortex                            | 3         | 0.6% | 0         |       |
| 100.303            | punctate cataract, equatorial cortex                           | 1         | 0.2% | 0         |       |
| 100.305            | punctate cataract, posterior sutures                           | 2         | 0.4% | 0         |       |
| 100.307            | punctate cataract, capsular                                    | 3         | 0.6% | 2         | 5.4%  |
| 100.311            | incipient cataract, anterior cortex                            | 3         | 0.6% | 0         |       |
| 100.312            | incipient cataract, posterior cortex                           | 8         | 1.6% | 0         |       |
| 100.313            | incipient cataract, equatorial cortex                          | 1         | 0.2% | 0         |       |
| 100.315            | incipient cataract, posterior sutures                          | 1         | 0.2% | 0         |       |
| 100.316            | incipient cataract, nucleus                                    | 2         | 0.4% | 0         |       |
| 100.317            | incipient cataract, capsular                                   | 2         | 0.4% | 0         |       |
| 100.330            | generalized/complete cataract                                  | 7         | 1.4% | 0         |       |
| 100.345            | <i>significant cataracts (summary)</i>                         | 39        | 8.0% | 2         | 5.4%  |
| 100.375            | <i>subluxation/luxation, unspecified</i>                       | 5         | 1.0% | 0         |       |
| <b>VITREOUS</b>    |  |           |      |           |       |
| 110.135            | PHPV/PTVL  | 2         | 0.4% | 0         |       |
| 110.320            | vitreal degeneration   | 6         | 1.2% | 0         |       |
| <b>FUNDUS</b>      |  |           |      |           |       |
| 97.120             | coloboma   | 1         | 0.2% | 0         |       |
| <b>RETINA</b>      |  |           |      |           |       |
| 120.170            | retinal dysplasia, folds                                       | 9         | 1.8% | 0         |       |
| 120.180            | retinal dysplasia, geographic                                  | 1         | 0.2% | 0         |       |
| 120.190            | retinal dysplasia, detached                                    | 1         | 0.2% | 0         |       |
| 120.310            | generalized progressive retinal atrophy (PRA)                  | 11        | 2.3% | 0         |       |
| 120.910            | retinal detachment without dialysis                            | 1         | 0.2% | 0         |       |
| 120.960            | retinopathy  | 0         |      | 1         | 2.7%  |
| <b>OPTIC NERVE</b> |  |           |      |           |       |
| 130.110            | micropapilla   | 0         |      | 1         | 2.7%  |
| 130.120            | optic nerve hypoplasia   | 1         | 0.2% | 0         |       |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 4 0.8%    | 0         |
| 900.100 other, not inherited                              | 10 2.1%   | 0         |
| 900.110 other. suspect not inherited/significance unknown | 2 0.4%    | 2 5.4%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 408 83.8% | 25 67.6%  |



# SERBIAN HOUND

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|------------------------------------|------------------------|-----------|--------------------|-------------------------------------|
| A. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 1         | NO                 | Mutation in the <i>prcd</i><br>gene |

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## Description and Comments

### A. Retinal atrophy – *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Serbian Hound is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Serbian Hound. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Zangerl, B., et al. (2006). "Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans." Genomics **88**(5): 551-563. PMID: 16938425

# SHETLAND SHEEPDOG

## (Sheltie)

|    | DISORDER  | INHERITANCE                | REFERENCE    | BREEDING ADVICE      | GENETIC TESTS AVAILABLE           |
|----|---|----------------------------|--------------|----------------------|-----------------------------------|
| A. | Distichiasis  | Not defined                | 1            | Breeder option       |                                   |
| B. | 1. Corneal dystrophy<br>2. Sheltie corneal dystrophy  | Not defined<br>Not defined | 1, 2<br>1, 2 | Breeder option<br>NO |                                   |
| C. | Persistent pupillary membranes<br>- iris to iris  | Not defined                | 1, 3         | Breeder option       |                                   |
| D. | Cataract  | Not defined                | 1            | NO                   |                                   |
| E. | Retinal atrophy<br>( <i>CNGA1</i> )   | Autosomal recessive        | 1, 4         | NO                   | Mutation in the <i>CNGA1</i> gene |
| F. | Slowly progressing retinopathy  | Not defined                | 5            | NO                   |                                   |
| G. | Choroidal hypoplasia<br>(Collie eye anomaly)<br>- optic nerve coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- staphyloma/coloboma | Autosomal recessive        | 1, 6, 7      | NO                   | Mutation in the <i>NHEJ1</i> gene |

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### Description and Comments

#### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded. Breeding discretion is advised.

Distichiasis in the Shetland Sheepdog usually involves stiff lashes which require permanent epilation.

B. 1. Corneal dystrophy

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

2. Sheltie corneal dystrophy

The corneal changes in the Shetland Sheepdog are characterized grossly by multifocal, central, subepithelial and superficial stromal, grey-white, circular or irregular rings. Some affected animals develop corneal erosions. The precocular tear film in the majority of dogs is unstable and requires symptomatic therapy to keep the patients comfortable. Further studies are necessary to define this disorder.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms are seen in the Shetland sheepdog and pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Retinal atrophy - *CNGA1*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. PRA is inherited as an autosomal recessive trait in most breeds.

One form of PRA in the Shetland Sheepdog is caused by a 4bp exonic deletion in *CNGA1*. However multiple forms of PRA exist in the breed and slowly progressive retinopathy is also not genetically linked to this mutation. A DNA test is available; however it will only detect this mutation.

F. Slowly progressing retinopathy

A syndrome as yet not well defined. May be a variant of PRA.

- G. Choroidal hypoplasia (Collie eye anomaly)
- staphyloma/coloboma
  - retinal detachment
  - retinal hemorrhage
  - optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie eye anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Crispin SM, Barnett KC. Dystrophy, degeneration and infiltration of the canine cornea. *J Small Anim Pract.* 1983;24:63-83.
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4. Wiik AC, Ropstad EO, Ekesten B, et al. Progressive retinal atrophy in Shetland Sheepdog is associated with a mutation in the *CNGA1* gene. *Anim Genet.* 2015;46:515-521.
5. Karlstam L, Hertel E, Zeiss C, et al. A slowly progressive retinopathy in the Shetland Sheepdog. *Vet Ophthalmol.* 2011;14:227-238.
6. Barnett KC, Stades FC. Collie eye anomaly in the Shetland Sheepdog in the Netherlands. *J Small Anim Pract.* 1979;20:321-329.
7. Parker HG, Kukekova AV, Akey DT, et al. Breed relationships facilitate fine-mapping studies: a 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. *Gen Res.* 2007;17:1562-1571.
8. Fredholm M, Larsen RC, Jönsson M, Söderlund MA, Hardon T, Proschowsky HF. Discrepancy in compliance between the clinical and genetic diagnosis of choroidal hypoplasia in Danish Rough Collies and Shetland Sheepdogs. *Anim Genet.* 2016 Apr; 47(2): 250-2.

# OCULAR DISORDERS REPORT SHETLAND SHEEPDOG

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>38,374 |      | 2016-2020<br>5,067 |      |
|---------------------|--|---------------------|------|--------------------|------|
|                     |  | #                   | %    | #                  | %    |
| <b>GLOBE</b>        |  |                     |      |                    |      |
| 0.110               | microphthalmia   | 64                  | 0.2% | 6                  | 0.1% |
| 10.000              | glaucoma   | 2                   | 0.0% | 0                  |      |
| <b>EYELIDS</b>      |  |                     |      |                    |      |
| 20.140              | ectopic cilia  | 9                   | 0.0% | 0                  |      |
| 21.000              | entropion, unspecified   | 7                   | 0.0% | 1                  | 0.0% |
| 22.000              | ectropion, unspecified   | 10                  | 0.0% | 0                  |      |
| 25.110              | distichiasis   | 2,491               | 6.5% | 251                | 5.0% |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 5                   | 0.0% | 2                  | 0.0% |
| 40.910              | keratoconjunctivitis sicca                                     | 7                   | 0.0% | 0                  |      |
| <b>NICTITANS</b>    |  |                     |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 7                   | 0.0% | 0                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 4                   | 0.0% | 0                  |      |
| <b>CORNEA</b>       |  |                     |      |                    |      |
| 70.210              | corneal pannus   | 9                   | 0.0% | 0                  |      |
| 70.220              | pigmentary keratitis   | 4                   | 0.0% | 0                  |      |
| 70.700              | corneal dystrophy  | 1,047               | 2.7% | 123                | 2.4% |
| 70.730              | corneal endothelial degeneration                               | 35                  | 0.1% | 1                  | 0.0% |
| <b>UVEA</b>         |  |                     |      |                    |      |
| 90.250              | pigmentary uveitis   | 1                   | 0.0% | 0                  |      |
| 93.110              | iris hypoplasia  | 5                   | 0.0% | 2                  | 0.0% |
| 93.140              | corneal endothelial pigment without PPM                        | 5                   | 0.0% | 0                  |      |
| 93.150              | iris coloboma  | 26                  | 0.1% | 2                  | 0.0% |
| 93.710              | persistent pupillary membranes, iris to iris                   | 1,599               | 4.2% | 273                | 5.4% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 119                 | 0.3% | 9                  | 0.2% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 192                 | 0.5% | 16                 | 0.3% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 29                  | 0.1% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 11                  | 0.0% | 9                  | 0.2% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 18                  | 0.0% | 9                  | 0.2% |
| 93.810              | uveal melanoma   | 1                   | 0.0% | 0                  |      |
| 93.999              | uveal cysts  | 24                  | 0.1% | 5                  | 0.1% |
| 97.150              | chorioretinal coloboma, congenital                             | 5                   | 0.0% | 6                  | 0.1% |
| <b>LENS</b>         |  |                     |      |                    |      |
| 100.200             | cataract, unspecified  | 73                  | 0.2% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 593                 | 1.5% | 111                | 2.2% |
| 100.301             | punctate cataract, anterior cortex                             | 71                  | 0.2% | 17                 | 0.3% |
| 100.302             | punctate cataract, posterior cortex                            | 61                  | 0.2% | 11                 | 0.2% |
| 100.303             | punctate cataract, equatorial cortex                           | 28                  | 0.1% | 3                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 4                   | 0.0% | 2                  | 0.0% |
| 100.305             | punctate cataract, posterior sutures                           | 8                   | 0.0% | 3                  | 0.1% |
| 100.306             | punctate cataract, nucleus                                     | 22                  | 0.1% | 2                  | 0.0% |
| 100.307             | punctate cataract, capsular                                    | 20                  | 0.1% | 6                  | 0.1% |
| 100.311             | incipient cataract, anterior cortex                            | 138                 | 0.4% | 17                 | 0.3% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.312 incipient cataract, posterior cortex              | 92               | 0.2%  | 14               | 0.3%  |
| 100.313 incipient cataract, equatorial cortex             | 55               | 0.1%  | 3                | 0.1%  |
| 100.314 incipient cataract, anterior sutures              | 5                | 0.0%  | 0                |       |
| 100.315 incipient cataract, posterior sutures             | 13               | 0.0%  | 0                |       |
| 100.316 incipient cataract, nucleus                       | 35               | 0.1%  | 2                | 0.0%  |
| 100.317 incipient cataract, capsular                      | 28               | 0.1%  | 6                | 0.1%  |
| 100.321 incomplete cataract, anterior cortex              | 1                | 0.0%  | 6                | 0.1%  |
| 100.322 incomplete cataract, posterior cortex             | 2                | 0.0%  | 5                | 0.1%  |
| 100.323 incomplete cataract, equatorial cortex            | 1                | 0.0%  | 3                | 0.1%  |
| 100.327 incomplete cataract, capsular                     | 1                | 0.0%  | 2                | 0.0%  |
| 100.328 y-suture tip opacities                            | 2                | 0.0%  | 4                | 0.1%  |
| 100.330 generalized/complete cataract                     | 44               | 0.1%  | 3                | 0.1%  |
| 100.340 resorbing/hypermature cataract                    | 1                | 0.0%  | 0                |       |
| 100.345 <i>significant cataracts (summary)</i>            | 705              | 1.8%  | 109              | 2.2%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 6                | 0.0%  | 1                | 0.0%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 90               | 0.2%  | 8                | 0.2%  |
| 110.135 PHPV/PTVL   | 17               | 0.0%  | 5                | 0.1%  |
| 110.200 vitreous degeneration-anterior chamber            | 1                | 0.0%  | 1                | 0.0%  |
| 110.320 vitreal degeneration                              | 133              | 0.3%  | 23               | 0.5%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 125              | 0.3%  | 25               | 0.5%  |
| 97.120 coloboma   | 82               | 0.2%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 86               | 0.2%  | 16               | 0.3%  |
| 120.180 retinal dysplasia, geographic                     | 16               | 0.0%  | 6                | 0.1%  |
| 120.190 retinal dysplasia, detached                       | 5                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 215              | 0.6%  | 8                | 0.2%  |
| 120.910 retinal detachment without dialysis               | 18               | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 1                | 0.0%  | 0                |       |
| 120.960 retinopathy                                       | 19               | 0.0%  | 5                | 0.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 17               | 0.0%  | 3                | 0.1%  |
| 130.120 optic nerve hypoplasia                            | 25               | 0.1%  | 0                |       |
| 130.150 optic disc coloboma                               | 191              | 0.5%  | 11               | 0.2%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 243              | 0.6%  | 0                |       |
| 900.100 other, not inherited                              | 570              | 1.5%  | 10               | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown | 279              | 0.7%  | 164              | 3.2%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 32,486           | 84.7% | 4,018            | 79.3% |

# SHIBA INU

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--------------------------------|-------------|-----------|-------------------------|
| A. | Glaucoma                       | Not defined | 1-3       | NO                      |
| B. | Distichiasis                   | Not defined | 1         | Breeder option          |
| C. | Persistent pupillary membranes |             |           |                         |
|    | - iris to iris                 | Not defined | 3         | Breeder option          |
|    | - lens pigment foci/no strands | Not defined | 3         | Passes with no notation |
| D. | Cataract                       | Not defined | 3         | NO                      |
| E. | Y-suture tip opacity           | Not defined | 3         | Breeder option          |

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## Description and Comments

### A. Glaucoma

An elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine breed eye screening exam.

A recent study found that a *SRBD1* polymorphism in exon 4 plays an important role in the development of glaucoma in the Shiba Inu. A genetic test is not yet available.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

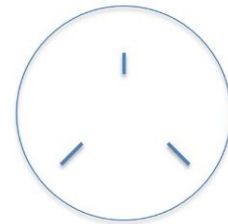
Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## References

1. ACVO Genetics Committee and Data from OFA All-Breeds Report.
2. Kanemaki N, Tchedre KT, Imayasu M, et al. Dogs and humans share a common susceptibility gene SRBD1 for glaucoma risk. *PLoS one*. 2013;8:e74372.
3. Kato K, Sasaki N, Matsunaga S, et al. Possible association of glaucoma with pectinate ligament dysplasia and narrowing of the iridocorneal angle in Shiba Inu dogs in Japan. *Vet Ophthalmol*. 2006;9:71-75.



# OCULAR DISORDERS REPORT SHIBA INU

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015 |      | 2016-2020 |      |
|---------------------|--|-----------|------|-----------|------|
|                     |  | #         | %    | #         | %    |
| <b>GLOBE</b>        |  |           |      |           |      |
| 10.000              | glaucoma   | 2         | 0.0% | 0         |      |
| <b>EYELIDS</b>      |  |           |      |           |      |
| 20.140              | ectopic cilia  | 4         | 0.1% | 0         |      |
| 20.160              | macropalpebral fissure   | 6         | 0.1% | 0         |      |
| 21.000              | entropion, unspecified   | 12        | 0.3% | 0         |      |
| 25.110              | distichiasis   | 105       | 2.4% | 19        | 1.8% |
| <b>NASOLACRIMAL</b> |  |           |      |           |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 2         | 0.0% | 0         |      |
| 40.910              | keratoconjunctivitis sicca                                     | 1         | 0.0% | 0         |      |
| <b>NICTITANS</b>    |  |           |      |           |      |
| 52.110              | prolapsed gland of the third eyelid                            | 2         | 0.0% | 0         |      |
| <b>CORNEA</b>       |  |           |      |           |      |
| 70.210              | corneal pannus   | 4         | 0.1% | 0         |      |
| 70.220              | pigmentary keratitis   | 10        | 0.2% | 1         | 0.1% |
| 70.700              | corneal dystrophy  | 32        | 0.7% | 4         | 0.4% |
| 70.730              | corneal endothelial degeneration                               | 10        | 0.2% | 1         | 0.1% |
| <b>UVEA</b>         |  |           |      |           |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 175       | 4.0% | 45        | 4.1% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 15        | 0.3% | 1         | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 1         | 0.0% | 0         |      |
| 93.740              | persistent pupillary membranes, iris sheets                    | 1         | 0.0% | 1         | 0.1% |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 21        | 0.5% | 43        | 4.0% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 3         | 0.1% | 0         |      |
| 93.999              | uveal cysts  | 0         |      | 2         | 0.2% |
| <b>LENS</b>         |  |           |      |           |      |
| 100.200             | cataract, unspecified  | 10        | 0.2% | 0         |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 187       | 4.2% | 65        | 6.0% |
| 100.301             | punctate cataract, anterior cortex                             | 6         | 0.1% | 3         | 0.3% |
| 100.302             | punctate cataract, posterior cortex                            | 15        | 0.3% | 4         | 0.4% |
| 100.303             | punctate cataract, equatorial cortex                           | 3         | 0.1% | 0         |      |
| 100.304             | punctate cataract, anterior sutures                            | 3         | 0.1% | 0         |      |
| 100.305             | punctate cataract, posterior sutures                           | 23        | 0.5% | 19        | 1.8% |
| 100.306             | punctate cataract, nucleus                                     | 1         | 0.0% | 0         |      |
| 100.307             | punctate cataract, capsular                                    | 1         | 0.0% | 3         | 0.3% |
| 100.311             | incipient cataract, anterior cortex                            | 32        | 0.7% | 4         | 0.4% |
| 100.312             | incipient cataract, posterior cortex                           | 24        | 0.5% | 2         | 0.2% |
| 100.313             | incipient cataract, equatorial cortex                          | 12        | 0.3% | 1         | 0.1% |
| 100.314             | incipient cataract, anterior sutures                           | 2         | 0.0% | 0         |      |
| 100.315             | incipient cataract, posterior sutures                          | 11        | 0.2% | 6         | 0.6% |
| 100.316             | incipient cataract, nucleus                                    | 4         | 0.1% | 2         | 0.2% |
| 100.317             | incipient cataract, capsular                                   | 2         | 0.0% | 0         |      |
| 100.322             | incomplete cataract, posterior cortex                          | 0         |      | 2         | 0.2% |
| 100.328             | y-suture tip opacities   | 3         | 0.1% | 20        | 1.8% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.330 generalized/complete cataract                     | 19 0.4%          | 0                |
| 100.345 significant cataracts (summary)                   | 171 3.9%         | 66 6.1%          |
| 100.375 subluxation/luxation, unspecified                 | 3 0.1%           | 1 0.1%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 19 0.4%          | 5 0.5%           |
| 110.135 PHPV/PTVL   | 4 0.1%           | 0                |
| 110.320 vitreal degeneration                              | 32 0.7%          | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 7 0.2%           | 4 0.4%           |
| 120.180 retinal dysplasia, geographic                     | 2 0.0%           | 0                |
| 120.190 retinal dysplasia, detached                       | 2 0.0%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 28 0.6%          | 1 0.1%           |
| 120.400 retinal hemorrhage                                | 1 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| 120.960 retinopathy                                       | 1 0.0%           | 1 0.1%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.120 optic nerve hypoplasia                            | 7 0.2%           | 1 0.1%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 31 0.7%          | 0                |
| 900.100 other, not inherited                              | 95 2.2%          | 1 0.1%           |
| 900.110 other. suspect not inherited/significance unknown | 39 0.9%          | 45 4.1%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 3,714 84.3%      | 834 76.9%        |

# SHIH TZU

|    | DISORDER                                      | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|---|-------------|-----------|-----------------|
| A. | Keratoconjunctivitis sicca                    | Not defined | 1, 2      | NO              |
| B. | Glaucoma                                      | Not defined | 3         | NO              |
| C. | Entropion                                     | Not defined | 1         | Breeder option  |
| D. | Distichiasis                                  | Not defined | 1         | Breeder option  |
| E. | Ectopic cilia                                 | Not defined | 1         | Breeder option  |
| F. | Corneal dystrophy - epithelial/stromal        | Not defined | 1         | Breeder option  |
| G. | Pigmentary keratitis                          | Not defined | 1         | Breeder option  |
| H. | Persistent pupillary membranes - iris to iris | Not defined | 1         | Breeder option  |
| I. | Cataract                                      | Not defined | 1         | NO              |
| J. | Vitreous degeneration                         | Not defined | 1, 4, 5   | Breeder option  |
| K. | Retinal detachment                            | Not defined | 4, 6      | NO              |
| L. | Retinal atrophy - generalized                 | Not defined | 1         | NO              |
| M. | Optic nerve hypoplasia                        | Not defined | 1, 7      | NO              |
| N. | Retinal degeneration                          | Not defined | 5         | NO              |

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## Description and Comments

### A. Keratoconjunctivitis sicca (KCS)

An abnormality of the tear film, most commonly a deficiency of the aqueous portion, although the mucin and/or lipid layers may be affected; results in ocular irritation and/or vision impairment.

B. Glaucoma

An elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine breed eye screening exam.

A recent study found that a *SRBD1* polymorphism in intron 1 plays an important role in the development of glaucoma in the Shih Tzu. A genetic test is not yet available.

C. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

D. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded. Breeding discretion is advised.

E. Ectopic cilia

Hair emerging through the eyelid conjunctiva. Ectopic cilia occur more frequently in younger dogs and cause discomfort and corneal disease.

F. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

G. Exposure/pigmentary keratitis

A condition characterized by variable degrees of superficial vascularization, fibrosis and/or pigmentation of the cornea. May be associated with excessive exposure/irritation of the globe due to shallow orbits, lower eyelid medial entropion, lagophthalmos and macropalpebral fissure.

H. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

I. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

J. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

K. Retinal detachment

A separation of the sensory retina from the underlying tissue. It results in blindness when complete.

L. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

M. Optic nerve hypoplasia

A congenital defect of the optic nerve which causes blindness and abnormal pupil response in the affected eye. May be unable to differentiate from micropapilla on a routine (dilated) screening ophthalmoscopic exam.

N. Retinal degeneration

A unilateral or bilateral retinal disease which can be progressive. When bilateral, the ophthalmoscopic lesions are sometimes asymmetrical, particularly in the early stages of the disease. Fundus examination shows initially single or multiple focal retinal lesions that appear active (local infiltrative inflammation or granulation) or inactive. The lesions can progress resulting in widespread retinal atrophy. The end-stage ophthalmoscopic lesions vary and may appear indistinguishable from PRA, or may be more characteristic of an inflammatory retinopathy. The asymmetry of the fundus abnormalities and the presence of inflammatory lesions in the retina and choroid help to differentiate this disorder from PRA. The mode of inheritance of this disease is not known; however, studies of different families suggest that it is possibly inherited. An intriguing aspect of the disease has been the preponderance of affected males compared to females. This has been confirmed in a recent unpublished survey.

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# OCULAR DISORDERS REPORT SHIH TZU

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>2,537 |       | 2016-2020<br>847 |       |
|---|---------------------|--------------------|-------|------------------|-------|
|   |                     | #                  | %     | #                | %     |
| <b>GLOBE</b>  |                     |                    |       |                  |       |
| 0.110 microphthalmia  |                     | 6                  | 0.2%  | 0                |       |
| <b>EYELIDS</b>  |                     |                    |       |                  |       |
| 20.140 ectopic cilia  |                     | 41                 | 1.6%  | 4                | 0.5%  |
| 20.160 macropalpebral fissure                                       |                     | 57                 | 2.2%  | 0                |       |
| 21.000 entropion, unspecified                                       |                     | 171                | 6.7%  | 97               | 11.5% |
| 22.000 ectropion, unspecified                                       |                     | 4                  | 0.2%  | 0                |       |
| 25.110 distichiasis   |                     | 478                | 18.8% | 103              | 12.2% |
| <b>NASOLACRIMAL</b>   |                     |                    |       |                  |       |
| 32.110 imperforate lower nasolacrimal punctum                       |                     | 4                  | 0.2%  | 3                | 0.4%  |
| 40.910 keratoconjunctivitis sicca                                   |                     | 17                 | 0.7%  | 12               | 1.4%  |
| <b>NICTITANS</b>  |                     |                    |       |                  |       |
| 51.100 third eyelid cartilage anomaly                               |                     | 1                  | 0.0%  | 0                |       |
| <b>CORNEA</b>   |                     |                    |       |                  |       |
| 70.210 corneal pannus   |                     | 25                 | 1.0%  | 0                |       |
| 70.220 pigmentary keratitis   |                     | 145                | 5.7%  | 55               | 6.5%  |
| 70.700 corneal dystrophy  |                     | 32                 | 1.3%  | 3                | 0.4%  |
| 70.730 corneal endothelial degeneration                             |                     | 4                  | 0.2%  | 0                |       |
| <b>UVEA</b>   |                     |                    |       |                  |       |
| 93.140 corneal endothelial pigment without PPM                      |                     | 1                  | 0.0%  | 0                |       |
| 93.150 iris coloboma  |                     | 4                  | 0.2%  | 1                | 0.1%  |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 36                 | 1.4%  | 20               | 2.4%  |
| 93.720 persistent pupillary membranes, iris to lens                 |                     | 4                  | 0.2%  | 1                | 0.1%  |
| 93.730 persistent pupillary membranes, iris to cornea               |                     | 3                  | 0.1%  | 2                | 0.2%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 1                  | 0.0%  | 2                | 0.2%  |
| 93.999 uveal cysts  |                     | 5                  | 0.2%  | 0                |       |
| <b>LENS</b>   |                     |                    |       |                  |       |
| 100.200 cataract, unspecified                                       |                     | 16                 | 0.6%  | 0                |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 58                 | 2.3%  | 16               | 1.9%  |
| 100.301 punctate cataract, anterior cortex                          |                     | 14                 | 0.6%  | 2                | 0.2%  |
| 100.302 punctate cataract, posterior cortex                         |                     | 7                  | 0.3%  | 2                | 0.2%  |
| 100.303 punctate cataract, equatorial cortex                        |                     | 1                  | 0.0%  | 0                |       |
| 100.304 punctate cataract, anterior sutures                         |                     | 1                  | 0.0%  | 0                |       |
| 100.305 punctate cataract, posterior sutures                        |                     | 9                  | 0.4%  | 2                | 0.2%  |
| 100.306 punctate cataract, nucleus                                  |                     | 1                  | 0.0%  | 1                | 0.1%  |
| 100.307 punctate cataract, capsular                                 |                     | 2                  | 0.1%  | 3                | 0.4%  |
| 100.311 incipient cataract, anterior cortex                         |                     | 21                 | 0.8%  | 3                | 0.4%  |
| 100.312 incipient cataract, posterior cortex                        |                     | 20                 | 0.8%  | 2                | 0.2%  |
| 100.313 incipient cataract, equatorial cortex                       |                     | 13                 | 0.5%  | 1                | 0.1%  |
| 100.314 incipient cataract, anterior sutures                        |                     | 1                  | 0.0%  | 0                |       |
| 100.315 incipient cataract, posterior sutures                       |                     | 6                  | 0.2%  | 3                | 0.4%  |
| 100.316 incipient cataract, nucleus                                 |                     | 8                  | 0.3%  | 0                |       |
| 100.317 incipient cataract, capsular                                |                     | 2                  | 0.1%  | 0                |       |
| 100.321 incomplete cataract, anterior cortex                        |                     | 1                  | 0.0%  | 2                | 0.2%  |
| 100.322 incomplete cataract, posterior cortex                       |                     | 1                  | 0.0%  | 1                | 0.1%  |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.323 incomplete cataract, equatorial cortex            | 0                | 2 0.2%           |
| 100.326 incomplete cataract, nucleus                      | 0                | 2 0.2%           |
| 100.328 y-suture tip opacities                            | 0                | 3 0.4%           |
| 100.330 generalized/complete cataract                     | 23 0.9%          | 3 0.4%           |
| 100.345 <i>significant cataracts (summary)</i>            | 147 5.8%         | 32 3.8%          |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 4 0.2%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 13 0.5%          | 7 0.8%           |
| 110.200 vitreous degeneration-anterior chamber            | 10 0.4%          | 10 1.2%          |
| 110.320 vitreal degeneration                              | 158 6.2%         | 20 2.4%          |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 1 0.0%           | 0                |
| 97.120 coloboma   | 2 0.1%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 11 0.4%          | 1 0.1%           |
| 120.180 retinal dysplasia, geographic                     | 4 0.2%           | 0                |
| 120.190 retinal dysplasia, detached                       | 0                | 1 0.1%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 41 1.6%          | 1 0.1%           |
| 120.910 retinal detachment without dialysis               | 9 0.4%           | 0                |
| 120.920 retinal detachment with dialysis                  | 1 0.0%           | 1 0.1%           |
| 120.960 retinopathy                                       | 2 0.1%           | 2 0.2%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.120 optic nerve hypoplasia                            | 10 0.4%          | 1 0.1%           |
| 130.150 optic disc coloboma                               | 4 0.2%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 43 1.7%          | 0                |
| 900.100 other, not inherited                              | 97 3.8%          | 5 0.6%           |
| 900.110 other. suspect not inherited/significance unknown | 89 3.5%          | 81 9.6%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,505 59.3%      | 490 57.9%        |



# SHIKOKU

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|--|--------------------|------------------|------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Shikoku breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT SHIKOKU

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 25.110 distichiasis   |                     | 0         |       | 1         | 2.1%  |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 3         | 27.3% | 22        | 45.8% |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 0         |       | 5         | 10.4% |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 1         | 9.1%  | 3         | 6.2%  |
| 100.307 punctate cataract, capsular                                 |                     | 1         | 9.1%  | 0         |       |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 1         | 9.1%  | 0         |       |
| <b>OTHER</b>  |                     |           |       |           |       |
| 900.110 other. suspect not inherited/significance unknown           |                     | 0         |       | 4         | 8.3%  |
| <b>NORMAL</b>   |                     |           |       |           |       |
| 0.000 normal globe  |                     | 4         | 36.4% | 20        | 41.7% |

# SHILOH SHEPHERD

|    | <b>DISORDER</b>                           | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|---|--------------------|------------------|------------------------|
| A. | Corneal dystrophy<br>- epithelial/stromal | Not defined        | 1                | Breeder option         |

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## Description and Comments

### A. Corneal Dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Shiloh Shepherd breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT SHILOH SHEPHERD

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015<br>243 |       | 2016-2020<br>76 |       |
|------------------|--|------------------|-------|-----------------|-------|
|                  |  | #                | %     | #               | %     |
| <b>EYELIDS</b>   |  |                  |       |                 |       |
| 25.110           | distichiasis   | 2                | 0.8%  | 1               | 1.3%  |
| <b>NICTITANS</b> |  |                  |       |                 |       |
| 50.210           | pannus of third eyelid                                       | 0                |       | 1               | 1.3%  |
| <b>CORNEA</b>    |  |                  |       |                 |       |
| 70.210           | corneal pannus   | 0                |       | 3               | 3.9%  |
| 70.700           | corneal dystrophy  | 29               | 11.9% | 8               | 10.5% |
| 70.730           | corneal endothelial degeneration                             | 1                | 0.4%  | 0               |       |
| <b>UVEA</b>      |  |                  |       |                 |       |
| 93.710           | persistent pupillary membranes, iris to iris                 | 3                | 1.2%  | 0               |       |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands | 0                |       | 1               | 1.3%  |
| 93.999           | uveal cysts  | 1                | 0.4%  | 1               | 1.3%  |
| <b>LENS</b>      |  |                  |       |                 |       |
| 100.210          | cataract. suspect not inherited/significance unknown         | 11               | 4.5%  | 2               | 2.6%  |
| 100.302          | punctate cataract, posterior cortex                          | 1                | 0.4%  | 0               |       |
| 100.307          | punctate cataract, capsular                                  | 1                | 0.4%  | 0               |       |
| 100.312          | incipient cataract, posterior cortex                         | 1                | 0.4%  | 0               |       |
| 100.314          | incipient cataract, anterior sutures                         | 1                | 0.4%  | 0               |       |
| 100.330          | generalized/complete cataract                                | 1                | 0.4%  | 0               |       |
| 100.345          | significant cataracts (summary)                              | 5                | 2.1%  | 0               |       |
| <b>RETINA</b>    |  |                  |       |                 |       |
| 120.180          | retinal dysplasia, geographic                                | 2                | 0.8%  | 0               |       |
| <b>OTHER</b>     |  |                  |       |                 |       |
| 900.000          | other, unspecified   | 1                | 0.4%  | 0               |       |
| 900.100          | other, not inherited   | 4                | 1.6%  | 0               |       |
| 900.110          | other. suspect not inherited/significance unknown            | 1                | 0.4%  | 2               | 2.6%  |
| <b>NORMAL</b>    |  |                  |       |                 |       |
| 0.000            | normal globe   | 208              | 85.6% | 60              | 78.9% |

# SIBERIAN HUSKY

|    | DISORDER  | INHERITANCE                        | REFERENCE | BREEDING ADVICE   | GENETIC TESTS AVAILABLE              |
|----|---|------------------------------------|-----------|-------------------|--------------------------------------|
| A. | Glaucoma  | Not defined                        | 1-4       | NO                |                                      |
| B. | Distichiasis  | Not defined                        | 1         | Breeder option    |                                      |
| C. | Corneal dystrophy<br>- epithelial/stromal           | Presumed<br>autosomal<br>recessive | 1, 5-8    | NO                |                                      |
| D. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined                        | 1         | Breeder<br>option |                                      |
| E. | Cataract  | Not defined                        | 1, 4      | NO                |                                      |
| F. | Retinal atrophy<br>( <i>RPGR</i> )                  | X-linked                           | 1, 9, 10  | NO                | Mutation in the<br><i>RPGR</i> gene  |
| G. | Cone degeneration<br>- (achromatopsia)              | Autosomal<br>recessive             | 10        | NO                | Mutation in the<br><i>CNGB3</i> gene |
| H. | Uveodermatologic<br>syndrome                        | Not defined                        | 1, 11-14  | NO                |                                      |

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## Description and Comments

### A. Glaucoma

An elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening exam.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds.

Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded. Breeding discretion is advised.

C. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

In the Siberian Husky, the opacities are bilaterally symmetrical, round to oval and ring shaped. They occur early in life (0.5-2 years) and may progress to cause significant vision loss. When seen, it may be beneficial to feed a low fat diet and recheck the eyes the following year to see if the opacities resolve, ruling out inherited corneal dystrophy.

D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Siberian Husky, cataracts begin in the axial posterior cortex at approximately one year of age. Progression is variable and vision impairment may occur. In cases with rapid progression, secondary lens-induced uveitis and glaucoma may be associated with partial cataract resorption.

F. Retinal atrophy – (*RPGR*)

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. In the Siberian Husky, one form of PRA, known as XLPRA1, is due to a mutation in the *RPGR* gene and is inherited as a recessive, sex-linked trait. A DNA test is available.

G. Cone degeneration - hemeralopia/achromatopsia

Autosomal recessively inherited early degeneration of the cone photoreceptors. Afflicted puppies develop day-blindness, colorblindness, and photophobia between 8 and 12 weeks of age. Afflicted dogs remain ophthalmoscopically normal their entire life. Electroretinography is required to definitively diagnose the disorder. A missense mutation in the same gene (*CNGB3*) that has been identified in CD-affected Alaskan Malamute-derived dogs has been detected in German Shorthaired Pointers affected with a clinically identical allelic disorder. A DNA test is available.

## H. Uveodermatologic syndrome

Uveodermatologic syndrome in the Siberian Husky bears many similarities to a condition in people called Vogt-Koyanagi-Harada (or VKH) syndrome. Thus, the condition in dogs is often referred to as VKH or VKH-like syndrome. It is an immune-mediated disease in which pigmented cells (melanocytes) in the eye and in the skin are destroyed by white blood cells (lymphocytes). The first clinical signs are usually inflammation of the intraocular structures (or uveitis) in both eyes. Adhesions between the iris and lens (posterior synechia) and the peripheral iris and cornea (peripheral anterior synechia) develop rapidly. Other complications include cataract development, retinal degeneration, retinal separation or detachment, optic disc atrophy and secondary glaucoma. The uveitis is very difficult to control medically and ultimately results in blindness in most affected dogs. Whitening of the hair (poliosis) and skin (vitiligo) may also be noted in advanced cases. The genetics of this condition are unclear, but some genetic predisposition is indicated by the higher prevalence of this disorder in Siberian Huskies compared with other dog breeds. Affected dogs are generally young, ranging in age between 1-1/2 to 4 years.

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# OCULAR DISORDERS REPORT SIBERIAN HUSKY

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>37,828 |      | 2016-2020<br>5,862 |      |
|---------------------|--|---------------------|------|--------------------|------|
|                     |  | #                   | %    | #                  | %    |
| <b>GLOBE</b>        |  |                     |      |                    |      |
| 0.110               | microphthalmia   | 7                   | 0.0% | 0                  |      |
| 10.000              | glaucoma   | 12                  | 0.0% | 3                  | 0.1% |
| <b>EYELIDS</b>      |  |                     |      |                    |      |
| 20.110              | eyelid dermoid   | 4                   | 0.0% | 0                  |      |
| 20.140              | ectopic cilia  | 3                   | 0.0% | 0                  |      |
| 20.160              | macropalpebral fissure   | 1                   | 0.0% | 0                  |      |
| 21.000              | entropion, unspecified   | 19                  | 0.1% | 1                  | 0.0% |
| 22.000              | ectropion, unspecified   | 4                   | 0.0% | 0                  |      |
| 25.110              | distichiasis   | 396                 | 1.0% | 61                 | 1.0% |
| <b>NASOLACRIMAL</b> |  |                     |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 1                   | 0.0% | 3                  | 0.1% |
| 40.910              | keratoconjunctivitis sicca                                     | 3                   | 0.0% | 0                  |      |
| <b>NICTITANS</b>    |  |                     |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 2                   | 0.0% | 0                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 2                   | 0.0% | 0                  |      |
| <b>CORNEA</b>       |  |                     |      |                    |      |
| 70.210              | corneal pannus   | 21                  | 0.1% | 1                  | 0.0% |
| 70.220              | pigmentary keratitis   | 3                   | 0.0% | 0                  |      |
| 70.700              | corneal dystrophy  | 997                 | 2.6% | 100                | 1.7% |
| 70.730              | corneal endothelial degeneration                               | 37                  | 0.1% | 1                  | 0.0% |
| <b>UVEA</b>         |  |                     |      |                    |      |
| 93.110              | iris hypoplasia  | 3                   | 0.0% | 2                  | 0.0% |
| 93.140              | corneal endothelial pigment without PPM                        | 1                   | 0.0% | 0                  |      |
| 93.150              | iris coloboma  | 8                   | 0.0% | 1                  | 0.0% |
| 93.710              | persistent pupillary membranes, iris to iris                   | 891                 | 2.4% | 186                | 3.2% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 26                  | 0.1% | 3                  | 0.1% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 50                  | 0.1% | 7                  | 0.1% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 5                   | 0.0% | 1                  | 0.0% |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 14                  | 0.0% | 12                 | 0.2% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 17                  | 0.0% | 2                  | 0.0% |
| 93.810              | uveal melanoma   | 1                   | 0.0% | 0                  |      |
| 93.999              | uveal cysts  | 20                  | 0.1% | 3                  | 0.1% |
| 97.150              | chorioretinal coloboma, congenital                             | 3                   | 0.0% | 1                  | 0.0% |
| <b>LENS</b>         |  |                     |      |                    |      |
| 100.200             | cataract, unspecified  | 576                 | 1.5% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 688                 | 1.8% | 138                | 2.4% |
| 100.301             | punctate cataract, anterior cortex                             | 67                  | 0.2% | 19                 | 0.3% |
| 100.302             | punctate cataract, posterior cortex                            | 199                 | 0.5% | 21                 | 0.4% |
| 100.303             | punctate cataract, equatorial cortex                           | 36                  | 0.1% | 5                  | 0.1% |
| 100.304             | punctate cataract, anterior sutures                            | 10                  | 0.0% | 3                  | 0.1% |
| 100.305             | punctate cataract, posterior sutures                           | 103                 | 0.3% | 9                  | 0.2% |
| 100.306             | punctate cataract, nucleus                                     | 23                  | 0.1% | 10                 | 0.2% |
| 100.307             | punctate cataract, capsular                                    | 29                  | 0.1% | 15                 | 0.3% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.311 incipient cataract, anterior cortex               | 131              | 0.3%  | 22               | 0.4%  |
| 100.312 incipient cataract, posterior cortex              | 1,281            | 3.4%  | 104              | 1.8%  |
| 100.313 incipient cataract, equatorial cortex             | 63               | 0.2%  | 10               | 0.2%  |
| 100.314 incipient cataract, anterior sutures              | 18               | 0.0%  | 0                |       |
| 100.315 incipient cataract, posterior sutures             | 260              | 0.7%  | 5                | 0.1%  |
| 100.316 incipient cataract, nucleus                       | 91               | 0.2%  | 11               | 0.2%  |
| 100.317 incipient cataract, capsular                      | 87               | 0.2%  | 19               | 0.3%  |
| 100.321 incomplete cataract, anterior cortex              | 5                | 0.0%  | 8                | 0.1%  |
| 100.322 incomplete cataract, posterior cortex             | 58               | 0.2%  | 85               | 1.5%  |
| 100.323 incomplete cataract, equatorial cortex            | 1                | 0.0%  | 9                | 0.2%  |
| 100.324 incomplete cataract, anterior sutures             | 1                | 0.0%  | 2                | 0.0%  |
| 100.325 incomplete cataract, posterior sutures            | 6                | 0.0%  | 3                | 0.1%  |
| 100.326 incomplete cataract, nucleus                      | 10               | 0.0%  | 16               | 0.3%  |
| 100.327 incomplete cataract, capsular                     | 7                | 0.0%  | 7                | 0.1%  |
| 100.328 y-suture tip opacities                            | 6                | 0.0%  | 10               | 0.2%  |
| 100.330 generalized/complete cataract                     | 463              | 1.2%  | 14               | 0.2%  |
| 100.340 resorbing/hypermature cataract                    | 1                | 0.0%  | 1                | 0.0%  |
| 100.345 <i>significant cataracts (summary)</i>            | 3,532            | 9.3%  | 408              | 7.0%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 13               | 0.0%  | 2                | 0.0%  |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 47               | 0.1%  | 17               | 0.3%  |
| 110.135 PHPV/PTVL   | 6                | 0.0%  | 1                | 0.0%  |
| 110.200 vitreous degeneration-anterior chamber            | 0                |       | 1                | 0.0%  |
| 110.320 vitreal degeneration                              | 37               | 0.1%  | 4                | 0.1%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 47               | 0.1%  | 16               | 0.3%  |
| 97.120 coloboma   | 16               | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 91               | 0.2%  | 10               | 0.2%  |
| 120.180 retinal dysplasia, geographic                     | 50               | 0.1%  | 10               | 0.2%  |
| 120.190 retinal dysplasia, detached                       | 13               | 0.0%  | 1                | 0.0%  |
| 120.310 generalized progressive retinal atrophy (PRA)     | 162              | 0.4%  | 11               | 0.2%  |
| 120.400 retinal hemorrhage                                | 7                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 27               | 0.1%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 2                | 0.0%  | 1                | 0.0%  |
| 120.960 retinopathy                                       | 21               | 0.1%  | 27               | 0.5%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 3                | 0.0%  | 0                |       |
| 130.120 optic nerve hypoplasia                            | 7                | 0.0%  | 0                |       |
| 130.150 optic disc coloboma                               | 3                | 0.0%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 354              | 0.9%  | 0                |       |
| 900.100 other, not inherited                              | 753              | 2.0%  | 11               | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown | 478              | 1.3%  | 324              | 5.5%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 31,998           | 84.6% | 4,689            | 80.0% |

# SILKEN WINDHOUND

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|--|---------------------|-----------|-----------------|-----------------------------------|
| A. | Cataract   | Not defined         | 1         | NO              |                                   |
| B. | Vitreous degeneration  | Not defined         | 1         | Breeder option  |                                   |
| C. | Choroidal hypoplasia (Collie Eye Anomaly)<br>- staphyloma/coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- optic nerve coloboma | Autosomal recessive | 1, 2      | NO              | Mutation in the <i>NHEJ1</i> gene |
| D. | Y-suture tip opacity   | Not defined         | 1         | Breeder option  |                                   |
| E. | Retinopathy  | Not defined         | 1         | Breeder option  |                                   |

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## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### B. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

### C. Choroidal hypoplasia (Collie Eye Anomaly)

- Staphyloma/coloboma
- Retinal detachment
- Retinal hemorrhage
- Optic nerve coloboma

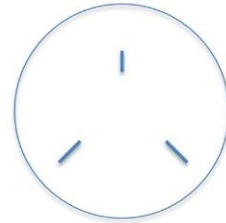
A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a

recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

#### D. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

#### E. Retinopathy

Patchy focal unilateral or bilateral hyper reflective tapetal lesions most frequently peripheral but occasionally central around a pigmented spot, usually non progressive. Not usually present prior to 3 months of age but usually present by 18 months of age.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Parker HG, Kukekova AV, Akey DT, et al. Breed relationships facilitate fine-mapping studies: a 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. *Gen Res.* 2007;17:1562-1571.

# OCULAR DISORDERS REPORT SILKEN WINDHOUND

| Diagnostic Name  | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|--|---------------------|-----------|-------|-----------|-------|
|  |                     | #         | %     | #         | %     |
| <b>GLOBE</b>   |                     |           |       |           |       |
| 0.110 microphthalmia   |                     | 0         |       | 1         | 0.2%  |
| <b>EYELIDS</b>   |                     |           |       |           |       |
| 25.110 distichiasis  |                     | 3         | 1.1%  | 4         | 1.0%  |
| <b>UVEA</b>  |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris          |                     | 1         | 0.4%  | 3         | 0.7%  |
| <b>LENS</b>  |                     |           |       |           |       |
| 100.210 cataract. suspect not inherited/significance unknown |                     | 16        | 5.7%  | 9         | 2.2%  |
| 100.302 punctate cataract, posterior cortex                  |                     | 1         | 0.4%  | 0         |       |
| 100.305 punctate cataract, posterior sutures                 |                     | 1         | 0.4%  | 0         |       |
| 100.307 punctate cataract, capsular                          |                     | 2         | 0.7%  | 0         |       |
| 100.311 incipient cataract, anterior cortex                  |                     | 1         | 0.4%  | 4         | 1.0%  |
| 100.312 incipient cataract, posterior cortex                 |                     | 0         |       | 1         | 0.2%  |
| 100.315 incipient cataract, posterior sutures                |                     | 1         | 0.4%  | 3         | 0.7%  |
| 100.317 incipient cataract, capsular                         |                     | 1         | 0.4%  | 0         |       |
| 100.328 y-suture tip opacities                               |                     | 1         | 0.4%  | 2         | 0.5%  |
| 100.345 <i>significant cataracts (summary)</i>               |                     | 8         | 2.9%  | 10        | 2.5%  |
| <b>VITREOUS</b>  |                     |           |       |           |       |
| 110.200 vitreous degeneration-anterior chamber               |                     | 0         |       | 3         | 0.7%  |
| 110.320 vitreal degeneration                                 |                     | 5         | 1.8%  | 4         | 1.0%  |
| <b>FUNDUS</b>  |                     |           |       |           |       |
| 97.110 choroidal hypoplasia                                  |                     | 1         | 0.4%  | 0         |       |
| <b>RETINA</b>  |                     |           |       |           |       |
| 120.180 retinal dysplasia, geographic                        |                     | 3         | 1.1%  | 1         | 0.2%  |
| 120.310 generalized progressive retinal atrophy (PRA)        |                     | 1         | 0.4%  | 0         |       |
| 120.960 retinopathy  |                     | 0         |       | 5         | 1.2%  |
| <b>OTHER</b>   |                     |           |       |           |       |
| 900.000 other, unspecified                                   |                     | 2         | 0.7%  | 0         |       |
| 900.100 other, not inherited                                 |                     | 0         |       | 1         | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown    |                     | 7         | 2.5%  | 20        | 4.9%  |
| <b>NORMAL</b>  |                     |           |       |           |       |
| 0.000 normal globe   |                     | 255       | 91.1% | 351       | 86.7% |

# SILKY TERRIER

|    | DISORDER  | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|---|------------------------|-----------|--------------------|-------------------------------------|
| A. | Entropion   | Not defined            | 1         | Breeder option     |                                     |
| B. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined            | 1         | Breeder option     |                                     |
| C. | Cataract  | Not defined            | 1-3       | NO                 |                                     |
| D. | Vitreous degeneration                               | Not defined            | 1, 2, 4   | Breeder option     |                                     |
| E. | Retinal atrophy<br>( <i>prcd</i> )                  | Autosomal<br>recessive | 5         | NO                 | Mutation in the<br><i>prcd</i> gene |

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## Description and Comments

### A. Entropion

A conformational defect resulting in an "in rolling" of one or more of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally in the neonatal period. These strands may bridge from iris to iris, iris to cornea, iris to lens, or from sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membranes, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### D. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

E. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Silky Terrier is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gelatt KN, Samuelson DA, Barrie KP, et al. Biometry and clinical characteristics of congenital cataracts and microphthalmia in the Miniature Schnauzer. *J Am Vet Med Assoc.* 1983;183:99-102.
3. Gelatt KN, Mackay EO. Prevalence of primary breed-related cataracts in the dog in North America. *Vet Ophthalmol.* 2005;8:101-111.
4. Koch SA. Cataracts in interrelated old English Sheepdogs. *J Am Vet Med Assoc.* 1972;160:299-301.
5. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics.* 2006;88:551-563. PMID: 16938425

# OCULAR DISORDERS REPORT SILKY TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>690 |       | 2016-2020<br>234 |       |
|---------------------|--|------------------|-------|------------------|-------|
|                     |  | #                | %     | #                | %     |
| <b>EYELIDS</b>      |  |                  |       |                  |       |
| 21.000              | entropion, unspecified   | 1                | 0.1%  | 8                | 3.4%  |
| 25.110              | distichiasis   | 3                | 0.4%  | 0                |       |
| <b>NASOLACRIMAL</b> |  |                  |       |                  |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 0                |       | 2                | 0.9%  |
| <b>NICTITANS</b>    |  |                  |       |                  |       |
| 52.110              | prolapsed gland of the third eyelid                            | 1                | 0.1%  | 0                |       |
| <b>CORNEA</b>       |  |                  |       |                  |       |
| 70.220              | pigmentary keratitis   | 0                |       | 1                | 0.4%  |
| 70.700              | corneal dystrophy  | 8                | 1.2%  | 0                |       |
| <b>UVEA</b>         |  |                  |       |                  |       |
| 93.140              | corneal endothelial pigment without PPM                        | 1                | 0.1%  | 0                |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 45               | 6.5%  | 16               | 6.8%  |
| 93.720              | persistent pupillary membranes, iris to lens                   | 1                | 0.1%  | 0                |       |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 3                | 0.4%  | 0                |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 1                | 0.1%  | 1                | 0.4%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 1                | 0.1%  | 1                | 0.4%  |
| <b>LENS</b>         |  |                  |       |                  |       |
| 100.200             | cataract, unspecified  | 4                | 0.6%  | 0                |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 35               | 5.1%  | 16               | 6.8%  |
| 100.301             | punctate cataract, anterior cortex                             | 9                | 1.3%  | 2                | 0.9%  |
| 100.302             | punctate cataract, posterior cortex                            | 4                | 0.6%  | 3                | 1.3%  |
| 100.303             | punctate cataract, equatorial cortex                           | 6                | 0.9%  | 0                |       |
| 100.304             | punctate cataract, anterior sutures                            | 1                | 0.1%  | 0                |       |
| 100.305             | punctate cataract, posterior sutures                           | 0                |       | 3                | 1.3%  |
| 100.306             | punctate cataract, nucleus                                     | 1                | 0.1%  | 3                | 1.3%  |
| 100.307             | punctate cataract, capsular                                    | 0                |       | 1                | 0.4%  |
| 100.311             | incipient cataract, anterior cortex                            | 12               | 1.7%  | 4                | 1.7%  |
| 100.312             | incipient cataract, posterior cortex                           | 18               | 2.6%  | 5                | 2.1%  |
| 100.313             | incipient cataract, equatorial cortex                          | 9                | 1.3%  | 1                | 0.4%  |
| 100.314             | incipient cataract, anterior sutures                           | 1                | 0.1%  | 0                |       |
| 100.315             | incipient cataract, posterior sutures                          | 2                | 0.3%  | 1                | 0.4%  |
| 100.316             | incipient cataract, nucleus                                    | 0                |       | 1                | 0.4%  |
| 100.317             | incipient cataract, capsular                                   | 1                | 0.1%  | 1                | 0.4%  |
| 100.321             | incomplete cataract, anterior cortex                           | 1                | 0.1%  | 1                | 0.4%  |
| 100.322             | incomplete cataract, posterior cortex                          | 1                | 0.1%  | 1                | 0.4%  |
| 100.323             | incomplete cataract, equatorial cortex                         | 0                |       | 1                | 0.4%  |
| 100.328             | y-suture tip opacities   | 0                |       | 4                | 1.7%  |
| 100.330             | generalized/complete cataract                                  | 22               | 3.2%  | 1                | 0.4%  |
| 100.345             | <i>significant cataracts (summary)</i>                         | 92               | 13.3% | 33               | 14.1% |
| <b>VITREOUS</b>     |  |                  |       |                  |       |
| 110.135             | PHPV/PTVL  | 0                |       | 1                | 0.4%  |
| 110.200             | vitreal degeneration-anterior chamber                          | 0                |       | 4                | 1.7%  |
| 110.320             | vitreal degeneration   | 29               | 4.2%  | 14               | 6.0%  |



|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>FUNDUS</b>   |           |           |
| 97.110 choroidal hypoplasia                               | 2 0.3%    | 1 0.4%    |
| <b>RETINA</b>   |           |           |
| 120.170 retinal dysplasia, folds                          | 4 0.6%    | 1 0.4%    |
| 120.180 retinal dysplasia, geographic                     | 1 0.1%    | 1 0.4%    |
| 120.310 generalized progressive retinal atrophy (PRA)     | 8 1.2%    | 1 0.4%    |
| 120.910 retinal detachment without dialysis               | 1 0.1%    | 0         |
| 120.960 retinopathy                                       | 0         | 1 0.4%    |
| <b>OPTIC NERVE</b>  |           |           |
| 130.110 micropapilla                                      | 1 0.1%    | 1 0.4%    |
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 12 1.7%   | 0         |
| 900.100 other, not inherited                              | 24 3.5%   | 0         |
| 900.110 other. suspect not inherited/significance unknown | 9 1.3%    | 10 4.3%   |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 519 75.2% | 160 68.4% |

# SLOUGHI

|    | DISORDER                            | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE           |
|----|-------------------------------------|------------------------|-----------|--------------------|--------------------------------------|
| A. | Retinal atrophy<br>( <i>rcd1a</i> ) | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>PDE6B</i> gene |

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## Description and Comments

### A. Retinal atrophy - *rcd1a*

A later onset degenerative disease of the retinal visual cells with visual deficits detectable at 2 to 3 years of age and which progresses to blindness. This abnormality may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. It is inherited as an autosomal recessive trait.

In the Sloughi, the disease is due to an 8-bp insertion in exon 21 of the *PDE6B* gene causing the *rcd1a* form of PRA. The disease is genetically distinct from that in the Irish Setter and has a later age of onset. A DNA test is available.

## References

1. Dekomien G, Runte M, Godde R, et al. Generalized progressive retinal atrophy of Sloughi dogs is due to an 8-bp insertion in exon 21 of the *PDE6B* gene. *Cytogenet Cell Genet.* 2000;90:261-267.

# OCULAR DISORDERS REPORT SLOUGHI

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 25.110 distichiasis   |                     | 0         |       | 2         | 5.0%  |
| <b>NICTITANS</b>  |                     |           |       |           |       |
| 51.100 third eyelid cartilage anomaly                               |                     | 1         | 3.1%  | 0         |       |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 2         | 6.2%  | 0         |       |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 1         | 3.1%  | 3         | 7.5%  |
| <b>VITREOUS</b>   |                     |           |       |           |       |
| 110.320 vitreal degeneration  |                     | 1         | 3.1%  | 0         |       |
| <b>OTHER</b>  |                     |           |       |           |       |
| 900.000 other, unspecified  |                     | 1         | 3.1%  | 0         |       |
| <b>NORMAL</b>   |                     |           |       |           |       |
| 0.000 normal globe  |                     | 31        | 96.9% | 35        | 87.5% |

## SMOOTH FOX TERRIER\*

\*The Smooth Fox Terrier and the Wire Fox Terrier were originally considered two varieties of the same breed. They became separate breeds in 1985. It is likely that the same genetic diseases exist in both breeds.

|    | DISORDER      | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|---------------|------------------------|-----------|--------------------|---|
| A. | Glaucoma      | Not defined            | 1, 2      | NO                 |   |
| B. | Lens luxation | Autosomal<br>recessive | 1, 3-7    | NO                 | Mutation in the<br><i>ADAMTS17</i> gene |

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### Description and Comments

#### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of the intraocular pressure (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening exam.

#### B. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

### References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Martin CL and Wyman M. Primary glaucoma in the dog. *Vet Clin North Am.* 1978 May;8:257-286.
3. Lawson DD. Luxation of the crystalline lens in the dog. *J Small Anim Pract.* 1969;10:461.
4. Curtis R and Barnett KC. Primary lens luxation in the dog. *J Small Anim Pract.* 1980 Dec;21:657-668.

5. Hodgman SFJ. Abnormalities and defects in pedigree dogs: I. An investigation into the existence of abnormalities in pedigree dogs in British Isles. *J Small Anim Pract.* 1963;4:447.
6. Formston C. Observations on subluxation and luxation of the crystalline lens in the dog. *Journal of Comparative Pathology.* 1945;55:168.
7. Gould D, Pettitt L, McLaughlin B, et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011; 14: 378-384.

# OCULAR DISORDERS REPORT SMOOTH FOX TERRIER

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015<br>270 |       | 2016-2020<br>67 |       |
|-----------------|--|------------------|-------|-----------------|-------|
|                 |  | #                | %     | #               | %     |
| <b>CORNEA</b>   |  |                  |       |                 |       |
| 70.700          | corneal dystrophy  | 0                |       | 1               | 1.5%  |
| <b>UVEA</b>     |  |                  |       |                 |       |
| 93.710          | persistent pupillary membranes, iris to iris                   | 13               | 4.8%  | 1               | 1.5%  |
| 93.720          | persistent pupillary membranes, iris to lens                   | 0                |       | 1               | 1.5%  |
| 93.730          | persistent pupillary membranes, iris to cornea                 | 0                |       | 1               | 1.5%  |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands   | 1                | 0.4%  | 0               |       |
| 93.760          | persistent pupillary membranes, endothelial opacity/no strands | 1                | 0.4%  | 0               |       |
| <b>LENS</b>     |  |                  |       |                 |       |
| 100.210         | cataract. suspect not inherited/significance unknown           | 3                | 1.1%  | 0               |       |
| 100.311         | incipient cataract, anterior cortex                            | 1                | 0.4%  | 0               |       |
| 100.312         | incipient cataract, posterior cortex                           | 2                | 0.7%  | 0               |       |
| 100.330         | generalized/complete cataract                                  | 2                | 0.7%  | 0               |       |
| 100.345         | significant cataracts (summary)                                | 5                | 1.9%  | 0               |       |
| <b>VITREOUS</b> |  |                  |       |                 |       |
| 110.320         | vitreal degeneration   | 3                | 1.1%  | 1               | 1.5%  |
| <b>RETINA</b>   |  |                  |       |                 |       |
| 120.170         | retinal dysplasia, folds                                       | 1                | 0.4%  | 2               | 3.0%  |
| 120.310         | generalized progressive retinal atrophy (PRA)                  | 2                | 0.7%  | 1               | 1.5%  |
| 120.960         | retinopathy  | 0                |       | 1               | 1.5%  |
| <b>OTHER</b>    |  |                  |       |                 |       |
| 900.000         | other, unspecified   | 1                | 0.4%  | 0               |       |
| 900.100         | other, not inherited   | 6                | 2.2%  | 0               |       |
| 900.110         | other. suspect not inherited/significance unknown              | 3                | 1.1%  | 3               | 4.5%  |
| <b>NORMAL</b>   |  |                  |       |                 |       |
| 0.000           | normal globe   | 240              | 88.9% | 56              | 83.6% |

# SOFT-COATED WHEATEN TERRIER

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--------------------------------|-------------|-----------|-------------------------|
| A. | Distichiasis                   | Not defined | 1         | Breeder option          |
| B. | Persistent pupillary membranes |             |           |                         |
|    | - iris to iris                 | Not defined | 1, 2      | Breeder option          |
|    | - lens pigment foci/no strands | Not defined | 1         | Passes with no notation |
| C. | Cataract                       | Not defined | 1, 2      | NO                      |
| D. | Retinal dysplasia - folds      | Not defined | 1         | Breeder option          |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore is not noted on the certificate.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

D. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Van der Woerd A. Multiple ocular anomalies in two related litters of Soft-Coated Wheaten Terriers. *Prog Vet Comp Ophthal.* 1995;5:78.



# OCULAR DISORDERS REPORT SOFT COATED WHEATEN TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>7,291 |      | 2016-2020<br>1,235 |      |
|---|---------------------|--------------------|------|--------------------|------|
|   |                     | #                  | %    | #                  | %    |
| <b>GLOBE</b>  |                     |                    |      |                    |      |
| 10.000 glaucoma   |                     | 2                  | 0.0% | 0                  |      |
| <b>EYELIDS</b>  |                     |                    |      |                    |      |
| 20.160 macropalpebral fissure   |                     | 1                  | 0.0% | 0                  |      |
| 21.000 entropion, unspecified   |                     | 1                  | 0.0% | 0                  |      |
| 25.110 distichiasis   |                     | 130                | 1.8% | 38                 | 3.1% |
| <b>NASOLACRIMAL</b>   |                     |                    |      |                    |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 6                  | 0.1% | 4                  | 0.3% |
| <b>NICTITANS</b>  |                     |                    |      |                    |      |
| 52.110 prolapsed gland of the third eyelid                            |                     | 3                  | 0.0% | 0                  |      |
| <b>CORNEA</b>   |                     |                    |      |                    |      |
| 70.700 corneal dystrophy  |                     | 54                 | 0.7% | 6                  | 0.5% |
| <b>UVEA</b>   |                     |                    |      |                    |      |
| 93.140 corneal endothelial pigment without PPM                        |                     | 3                  | 0.0% | 0                  |      |
| 93.150 iris coloboma  |                     | 1                  | 0.0% | 0                  |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 240                | 3.3% | 67                 | 5.4% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 18                 | 0.2% | 0                  |      |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 3                  | 0.0% | 0                  |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 42                 | 0.6% | 55                 | 4.5% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 4                  | 0.1% | 4                  | 0.3% |
| 93.999 uveal cysts  |                     | 15                 | 0.2% | 5                  | 0.4% |
| <b>LENS</b>   |                     |                    |      |                    |      |
| 100.200 cataract, unspecified   |                     | 24                 | 0.3% | 0                  |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 341                | 4.7% | 69                 | 5.6% |
| 100.301 punctate cataract, anterior cortex                            |                     | 27                 | 0.4% | 13                 | 1.1% |
| 100.302 punctate cataract, posterior cortex                           |                     | 9                  | 0.1% | 3                  | 0.2% |
| 100.303 punctate cataract, equatorial cortex                          |                     | 13                 | 0.2% | 2                  | 0.2% |
| 100.304 punctate cataract, anterior sutures                           |                     | 4                  | 0.1% | 4                  | 0.3% |
| 100.305 punctate cataract, posterior sutures                          |                     | 4                  | 0.1% | 1                  | 0.1% |
| 100.306 punctate cataract, nucleus                                    |                     | 4                  | 0.1% | 0                  |      |
| 100.307 punctate cataract, capsular                                   |                     | 12                 | 0.2% | 6                  | 0.5% |
| 100.311 incipient cataract, anterior cortex                           |                     | 29                 | 0.4% | 9                  | 0.7% |
| 100.312 incipient cataract, posterior cortex                          |                     | 29                 | 0.4% | 4                  | 0.3% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 17                 | 0.2% | 2                  | 0.2% |
| 100.314 incipient cataract, anterior sutures                          |                     | 2                  | 0.0% | 1                  | 0.1% |
| 100.315 incipient cataract, posterior sutures                         |                     | 11                 | 0.2% | 2                  | 0.2% |
| 100.316 incipient cataract, nucleus                                   |                     | 17                 | 0.2% | 1                  | 0.1% |
| 100.317 incipient cataract, capsular                                  |                     | 12                 | 0.2% | 0                  |      |
| 100.321 incomplete cataract, anterior cortex                          |                     | 0                  |      | 2                  | 0.2% |
| 100.322 incomplete cataract, posterior cortex                         |                     | 0                  |      | 3                  | 0.2% |
| 100.328 y-suture tip opacities  |                     | 1                  | 0.0% | 3                  | 0.2% |
| 100.330 generalized/complete cataract                                 |                     | 35                 | 0.5% | 0                  |      |
| 100.340 resorbing/hypermature cataract                                |                     | 0                  |      | 1                  | 0.1% |
| 100.345 significant cataracts (summary)                               |                     | 250                | 3.4% | 57                 | 4.6% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.375 subluxation/luxation, unspecified                 | 4 0.1%           | 1 0.1%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 68 0.9%          | 8 0.6%           |
| 110.135 PHPV/PTVL   | 6 0.1%           | 0                |
| 110.320 vitreal degeneration                              | 13 0.2%          | 2 0.2%           |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 17 0.2%          | 0                |
| 97.120 coloboma   | 1 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 70 1.0%          | 4 0.3%           |
| 120.180 retinal dysplasia, geographic                     | 3 0.0%           | 1 0.1%           |
| 120.190 retinal dysplasia, detached                       | 2 0.0%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 14 0.2%          | 1 0.1%           |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| 120.960 retinopathy                                       | 2 0.0%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 14 0.2%          | 0                |
| 130.120 optic nerve hypoplasia                            | 5 0.1%           | 0                |
| 130.150 optic disc coloboma                               | 9 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 49 0.7%          | 0                |
| 900.100 other, not inherited                              | 183 2.5%         | 2 0.2%           |
| 900.110 other. suspect not inherited/significance unknown | 58 0.8%          | 57 4.6%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 6,330 86.8%      | 930 75.3%        |

# SPANISH WATER DOG

|    | DISORDER   | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|--|------------------------|-----------|--------------------|-------------------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined            | 1         | Breeder option     |                                     |
| B. | Retinal atrophy<br>( <i>prcd</i> )               | Autosomal<br>recessive | 1, 2      | NO                 | Mutation in the<br><i>prcd</i> gene |
| C. | Retinal dysplasia<br>- folds                     | Not defined            | 1         | Breeder option     |                                     |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that one form of PRA in the Spanish Water Dog is PRCD which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

### C. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in

their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and Data from OFA All-Breeds Report.
2. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006;88:551-563. PMID: 16938425

# OCULAR DISORDERS REPORT SPANISH WATER DOG

| Diagnostic Name    | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|--------------------|--|-----------|-------|-----------|-------|
|                    |  | #         | %     | #         | %     |
| <b>EYELIDS</b>     |  |           |       |           |       |
| 25.110             | distichiasis   | 2         | 0.9%  | 2         | 0.9%  |
| <b>NICTITANS</b>   |  |           |       |           |       |
| 52.110             | prolapsed gland of the third eyelid                          | 1         | 0.5%  | 0         |       |
| <b>CORNEA</b>      |  |           |       |           |       |
| 70.700             | corneal dystrophy  | 2         | 0.9%  | 1         | 0.5%  |
| <b>UVEA</b>        |  |           |       |           |       |
| 93.710             | persistent pupillary membranes, iris to iris                 | 7         | 3.3%  | 6         | 2.8%  |
| 93.750             | persistent pupillary membranes, lens pigment foci/no strands | 1         | 0.5%  | 0         |       |
| 93.999             | uveal cysts  | 0         |       | 1         | 0.5%  |
| <b>LENS</b>        |  |           |       |           |       |
| 100.210            | cataract. suspect not inherited/significance unknown         | 15        | 7.0%  | 10        | 4.7%  |
| 100.302            | punctate cataract, posterior cortex                          | 1         | 0.5%  | 0         |       |
| 100.305            | punctate cataract, posterior sutures                         | 0         |       | 1         | 0.5%  |
| 100.306            | punctate cataract, nucleus                                   | 2         | 0.9%  | 1         | 0.5%  |
| 100.313            | incipient cataract, equatorial cortex                        | 1         | 0.5%  | 1         | 0.5%  |
| 100.316            | incipient cataract, nucleus                                  | 1         | 0.5%  | 0         |       |
| 100.317            | incipient cataract, capsular                                 | 1         | 0.5%  | 0         |       |
| 100.345            | <i>significant cataracts (summary)</i>                       | 6         | 2.8%  | 3         | 1.4%  |
| <b>VITREOUS</b>    |  |           |       |           |       |
| 110.120            | persistent hyaloid artery/remnant                            | 1         | 0.5%  | 0         |       |
| 110.320            | vitreal degeneration   | 0         |       | 2         | 0.9%  |
| <b>RETINA</b>      |  |           |       |           |       |
| 120.170            | retinal dysplasia, folds                                     | 5         | 2.3%  | 3         | 1.4%  |
| 120.180            | retinal dysplasia, geographic                                | 1         | 0.5%  | 3         | 1.4%  |
| 120.190            | retinal dysplasia, detached                                  | 1         | 0.5%  | 0         |       |
| 120.310            | generalized progressive retinal atrophy (PRA)                | 5         | 2.3%  | 3         | 1.4%  |
| <b>OPTIC NERVE</b> |  |           |       |           |       |
| 130.110            | micropapilla   | 0         |       | 1         | 0.5%  |
| <b>OTHER</b>       |  |           |       |           |       |
| 900.000            | other, unspecified   | 4         | 1.9%  | 0         |       |
| 900.100            | other, not inherited   | 7         | 3.3%  | 0         |       |
| 900.110            | other. suspect not inherited/significance unknown            | 2         | 0.9%  | 16        | 7.5%  |
| <b>NORMAL</b>      |  |           |       |           |       |
| 0.000              | normal globe   | 181       | 84.2% | 172       | 80.8% |

# SPINONE ITALIANO

|    | DISORDER                       | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|----|--------------------------------|-------------|-----------|-------------------------|
| A. | Ectropion                      | Not defined | 1         | Breeder option          |
| B. | Distichiasis                   | Not defined | 1         | Breeder option          |
| C. | Persistent pupillary membranes |             |           |                         |
|    | - iris to iris                 | Not defined | 1         | Breeder options         |
|    | - lens pigment foci/no strands | Not defined | 1         | Passes with no notation |
| D. | Cataract                       | Not defined | 1         | NO                      |
| E. | Y-suture tip opacity           | Not defined | 1         | Breeder option          |

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## Description and Comments

### A. Ectropion

A conformational defect resulting in eversion of the eyelids, which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded. Breeding discretion is advised.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

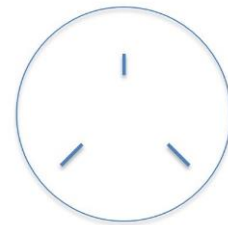
Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

#### D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

#### E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Spinone Italiano breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT SPINONE ITALIANO

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>2,060 |      | 2016-2020<br>407 |      |
|---|---------------------|--------------------|------|------------------|------|
|   |                     | #                  | %    | #                | %    |
| <b>GLOBE</b>  |                     |                    |      |                  |      |
| 0.110 microphthalmia  |                     | 1                  | 0.0% | 0                |      |
| <b>EYELIDS</b>  |                     |                    |      |                  |      |
| 20.160 macropalpebral fissure                                       |                     | 3                  | 0.1% | 0                |      |
| 21.000 entropion, unspecified                                       |                     | 30                 | 1.5% | 2                | 0.5% |
| 22.000 ectropion, unspecified                                       |                     | 12                 | 0.6% | 6                | 1.5% |
| 25.110 distichiasis   |                     | 25                 | 1.2% | 8                | 2.0% |
| <b>NASOLACRIMAL</b>   |                     |                    |      |                  |      |
| 40.910 keratoconjunctivitis sicca                                   |                     | 1                  | 0.0% | 0                |      |
| <b>NICTITANS</b>  |                     |                    |      |                  |      |
| 51.100 third eyelid cartilage anomaly                               |                     | 3                  | 0.1% | 0                |      |
| 52.110 prolapsed gland of the third eyelid                          |                     | 3                  | 0.1% | 0                |      |
| <b>UVEA</b>   |                     |                    |      |                  |      |
| 90.250 pigmentary uveitis   |                     | 1                  | 0.0% | 0                |      |
| 93.150 iris coloboma  |                     | 1                  | 0.0% | 0                |      |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 86                 | 4.2% | 34               | 8.4% |
| 93.720 persistent pupillary membranes, iris to lens                 |                     | 2                  | 0.1% | 2                | 0.5% |
| 93.730 persistent pupillary membranes, iris to cornea               |                     | 1                  | 0.0% | 0                |      |
| 93.740 persistent pupillary membranes, iris sheets                  |                     | 2                  | 0.1% | 1                | 0.2% |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 6                  | 0.3% | 2                | 0.5% |
| 93.999 uveal cysts  |                     | 3                  | 0.1% | 0                |      |
| <b>LENS</b>   |                     |                    |      |                  |      |
| 100.200 cataract, unspecified                                       |                     | 2                  | 0.1% | 0                |      |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 107                | 5.2% | 23               | 5.7% |
| 100.301 punctate cataract, anterior cortex                          |                     | 6                  | 0.3% | 0                |      |
| 100.302 punctate cataract, posterior cortex                         |                     | 3                  | 0.1% | 0                |      |
| 100.303 punctate cataract, equatorial cortex                        |                     | 1                  | 0.0% | 1                | 0.2% |
| 100.304 punctate cataract, anterior sutures                         |                     | 3                  | 0.1% | 0                |      |
| 100.305 punctate cataract, posterior sutures                        |                     | 2                  | 0.1% | 3                | 0.7% |
| 100.306 punctate cataract, nucleus                                  |                     | 14                 | 0.7% | 0                |      |
| 100.307 punctate cataract, capsular                                 |                     | 3                  | 0.1% | 1                | 0.2% |
| 100.311 incipient cataract, anterior cortex                         |                     | 13                 | 0.6% | 3                | 0.7% |
| 100.312 incipient cataract, posterior cortex                        |                     | 6                  | 0.3% | 0                |      |
| 100.313 incipient cataract, equatorial cortex                       |                     | 5                  | 0.2% | 0                |      |
| 100.314 incipient cataract, anterior sutures                        |                     | 1                  | 0.0% | 0                |      |
| 100.315 incipient cataract, posterior sutures                       |                     | 5                  | 0.2% | 1                | 0.2% |
| 100.316 incipient cataract, nucleus                                 |                     | 7                  | 0.3% | 5                | 1.2% |
| 100.317 incipient cataract, capsular                                |                     | 0                  |      | 2                | 0.5% |
| 100.322 incomplete cataract, posterior cortex                       |                     | 0                  |      | 1                | 0.2% |
| 100.328 y-suture tip opacities                                      |                     | 0                  |      | 3                | 0.7% |
| 100.330 generalized/complete cataract                               |                     | 5                  | 0.2% | 0                |      |
| 100.345 significant cataracts (summary)                             |                     | 76                 | 3.7% | 20               | 4.9% |
| 100.375 subluxation/luxation, unspecified                           |                     | 3                  | 0.1% | 0                |      |



|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>VITREOUS</b>   |             |           |
| 110.120 persistent hyaloid artery/remnant                 | 2 0.1%      | 0         |
| 110.200 vitreous degeneration-anterior chamber            | 2 0.1%      | 1 0.2%    |
| 110.320 vitreal degeneration                              | 19 0.9%     | 0         |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 10 0.5%     | 0         |
| 120.180 retinal dysplasia, geographic                     | 0           | 1 0.2%    |
| 120.310 generalized progressive retinal atrophy (PRA)     | 1 0.0%      | 0         |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 0           | 1 0.2%    |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 22 1.1%     | 0         |
| 900.100 other, not inherited                              | 62 3.0%     | 0         |
| 900.110 other. suspect not inherited/significance unknown | 15 0.7%     | 17 4.2%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,801 87.4% | 306 75.2% |

# ST. BERNARD

|    | DISORDER  | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|---|-------------|-----------|-----------------|
| A. | Ectropion                                       | Not defined | 1         | Breeder option  |
| B. | Entropion                                       | Not defined | 1, 2      | Breeder option  |
| C. | Distichiasis                                    | Not defined | 1         | Breeder option  |
| D. | Persistent pupillary membrane<br>- iris to iris | Not defined | 1         | Breeder option  |
| E. | Cataract  | Not defined | 1         | NO              |

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## Description and Comments

### A. Ectropion

A conformational defect resulting in eversion of the eyelids which may cause ocular irritation. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull. In this breed, entropion is associated with an exceptionally large palpebral fissure.

### C. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### D. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest

threat to vision and when severe, vision impairment or blindness may occur.

E. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Priester WA. Congenital ocular defects in cattle, horses, cats, and dogs. *J Am Vet Med Assoc.* 1972 Jun 1;160:1504-1511.

# OCULAR DISORDERS REPORT ST. BERNARD

| Diagnostic Name    | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|--------------------|--|-----------|-------|-----------|-------|
|                    |  | 213       |       | 128       |       |
|                    |  | #         | %     | #         | %     |
| <b>EYELIDS</b>     |  |           |       |           |       |
| 20.160             | macropalpebral fissure   | 21        | 9.9%  | 0         |       |
| 21.000             | entropion, unspecified   | 47        | 22.1% | 33        | 25.8% |
| 22.000             | ectropion, unspecified   | 73        | 34.3% | 34        | 26.6% |
| 25.110             | distichiasis   | 14        | 6.6%  | 5         | 3.9%  |
| <b>NICTITANS</b>   |  |           |       |           |       |
| 51.100             | third eyelid cartilage anomaly                                 | 1         | 0.5%  | 0         |       |
| 52.110             | prolapsed gland of the third eyelid                            | 1         | 0.5%  | 0         |       |
| <b>CORNEA</b>      |  |           |       |           |       |
| 70.220             | pigmentary keratitis   | 0         |       | 1         | 0.8%  |
| 70.700             | corneal dystrophy  | 2         | 0.9%  | 0         |       |
| <b>UVEA</b>        |  |           |       |           |       |
| 93.710             | persistent pupillary membranes, iris to iris                   | 20        | 9.4%  | 13        | 10.2% |
| 93.720             | persistent pupillary membranes, iris to lens                   | 0         |       | 1         | 0.8%  |
| 93.730             | persistent pupillary membranes, iris to cornea                 | 0         |       | 2         | 1.6%  |
| 93.760             | persistent pupillary membranes, endothelial opacity/no strands | 0         |       | 1         | 0.8%  |
| 93.999             | uveal cysts  | 1         | 0.5%  | 1         | 0.8%  |
| <b>LENS</b>        |  |           |       |           |       |
| 100.210            | cataract. suspect not inherited/significance unknown           | 12        | 5.6%  | 5         | 3.9%  |
| 100.302            | punctate cataract, posterior cortex                            | 2         | 0.9%  | 1         | 0.8%  |
| 100.303            | punctate cataract, equatorial cortex                           | 1         | 0.5%  | 0         |       |
| 100.305            | punctate cataract, posterior sutures                           | 1         | 0.5%  | 0         |       |
| 100.306            | punctate cataract, nucleus                                     | 1         | 0.5%  | 0         |       |
| 100.307            | punctate cataract, capsular                                    | 1         | 0.5%  | 0         |       |
| 100.311            | incipient cataract, anterior cortex                            | 2         | 0.9%  | 2         | 1.6%  |
| 100.312            | incipient cataract, posterior cortex                           | 3         | 1.4%  | 1         | 0.8%  |
| 100.313            | incipient cataract, equatorial cortex                          | 5         | 2.3%  | 1         | 0.8%  |
| 100.316            | incipient cataract, nucleus                                    | 4         | 1.9%  | 0         |       |
| 100.317            | incipient cataract, capsular                                   | 0         |       | 1         | 0.8%  |
| 100.321            | incomplete cataract, anterior cortex                           | 1         | 0.5%  | 0         |       |
| 100.326            | incomplete cataract, nucleus                                   | 1         | 0.5%  | 0         |       |
| 100.328            | y-suture tip opacities   | 1         | 0.5%  | 0         |       |
| 100.330            | generalized/complete cataract                                  | 8         | 3.8%  | 0         |       |
| 100.345            | <i>significant cataracts (summary)</i>                         | 31        | 14.6% | 6         | 4.7%  |
| <b>VITREOUS</b>    |  |           |       |           |       |
| 110.120            | persistent hyaloid artery/remnant                              | 3         | 1.4%  | 0         |       |
| 110.135            | PHPV/PTVL  | 1         | 0.5%  | 0         |       |
| <b>RETINA</b>      |  |           |       |           |       |
| 120.170            | retinal dysplasia, folds                                       | 5         | 2.3%  | 0         |       |
| <b>OPTIC NERVE</b> |  |           |       |           |       |
| 130.110            | micropapilla   | 1         | 0.5%  | 0         |       |
| 130.120            | optic nerve hypoplasia   | 1         | 0.5%  | 0         |       |

|   | 1991-2015 | 2016-2020 |
|---|-----------|-----------|
| <b>OTHER</b>  |           |           |
| 900.000 other, unspecified                                | 3 1.4%    | 0         |
| 900.100 other, not inherited                              | 6 2.8%    | 4 3.1%    |
| 900.110 other. suspect not inherited/significance unknown | 9 4.2%    | 8 6.2%    |
| <b>NORMAL</b>   |           |           |
| 0.000 normal globe  | 84 39.4%  | 57 44.5%  |

# STAFFORDSHIRE BULL TERRIER\*

\* Please note that since 1972 the AKC considers the Staffordshire Bull Terrier a different breed from the American Staffordshire Terrier.

|    | DISORDER   | INHERITANCE                | REFERENCE | BREEDING ADVICE                           | GENETIC TESTS AVAILABLE          |
|----|--|----------------------------|-----------|---|----------------------------------|
| A. | Distichiasis   | Not defined                | 1         | Breeder option                            |                                  |
| B. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined | 1<br>1    | Breeder option<br>Passes with no notation |                                  |
| C. | Cataract   | Autosomal recessive        | 1-4       | NO  | Mutation in the <i>HSF4</i> gene |
| D. | Persistent hyperplastic primary vitreous (PHPV)                                    | Not defined                | 1, 5, 6   | NO  |                                  |
| E. | Vitreous degeneration  | Not defined                | 1         | Breeder option                            |                                  |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore is not noted on the certificate.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Staffordshire Bull Terrier, cataracts usually develop by one year of age. There is initial opacification of the suture lines progressing to nuclear and cortical cataract formation; complete cataracts and blindness develop by three years of age. The condition is inherited as an autosomal recessive mutation in the *HSF4* gene (*HSF4-1*). A DNA test is available.

### D. Persistent hyperplastic primary vitreous (PHPV)

A congenital defect resulting from abnormalities in the development and regression of the hyaloid artery (the primary vitreous) and the interaction of this blood vessel with the posterior lens capsule/cortex during embryogenesis. This condition is often associated with persistent tunica vasculosa lentis (PTVL) which results from failure of regression of the embryologic vascular network which surrounds the developing lens.

The majority of affected dogs have a retrolental fibrovascular plaque and variable lenticular defects which include posterior lenticonus/globus, colobomata, intralenticular hemorrhage and/or secondary cataracts. Vision impairment may result. The disease is an inherited disorder in the breed, but the mode of inheritance has not been defined. The results of current studies cannot rule out autosomal recessive or a dominant trait with incomplete penetrance.

### E. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Barnett KC. Hereditary cataract in the dog. *J Small Anim Pract.* 1978;19:109-120.
3. Barnett KC. The diagnosis and differential diagnosis of cataract in the dog. *J Small Anim Pract.* 1985;26:305-316.
4. Mellersh CS, McLaughlin B, Ahonen S, et al. Mutation in *HSF4* is associated with hereditary cataract in the Australian Shepherd. *Vet Ophthalmol.* 2009;12:372-378.
5. Curtis R, Barnett KC, Leon A. Persistent hyperplastic primary vitreous in the Staffordshire Bull Terrier. *Vet Rec.* 1984;115:385.
6. Leon A, Curtis R, Barnett K. Hereditary persistent hyperplastic primary vitreous in the Staffordshire Bull Terrier. *J Am Anim Hosp Assoc.* 1986;22:765-774.

# OCULAR DISORDERS REPORT STAFFORDSHIRE BULL TERRIER

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015<br>846 |       | 2016-2020<br>484 |       |
|-----------------|--|------------------|-------|------------------|-------|
|                 |  | #                | %     | #                | %     |
| <b>EYELIDS</b>  |  |                  |       |                  |       |
| 25.110          | distichiasis   | 73               | 8.6%  | 29               | 6.0%  |
| <b>CORNEA</b>   |  |                  |       |                  |       |
| 70.700          | corneal dystrophy  | 1                | 0.1%  | 3                | 0.6%  |
| <b>UVEA</b>     |  |                  |       |                  |       |
| 93.710          | persistent pupillary membranes, iris to iris                   | 20               | 2.4%  | 16               | 3.3%  |
| 93.720          | persistent pupillary membranes, iris to lens                   | 2                | 0.2%  | 0                |       |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands   | 3                | 0.4%  | 13               | 2.7%  |
| 93.760          | persistent pupillary membranes, endothelial opacity/no strands | 1                | 0.1%  | 0                |       |
| 93.999          | uveal cysts  | 6                | 0.7%  | 0                |       |
| <b>LENS</b>     |  |                  |       |                  |       |
| 100.210         | cataract. suspect not inherited/significance unknown           | 32               | 3.8%  | 17               | 3.5%  |
| 100.301         | punctate cataract, anterior cortex                             | 5                | 0.6%  | 5                | 1.0%  |
| 100.302         | punctate cataract, posterior cortex                            | 1                | 0.1%  | 0                |       |
| 100.303         | punctate cataract, equatorial cortex                           | 1                | 0.1%  | 1                | 0.2%  |
| 100.304         | punctate cataract, anterior sutures                            | 1                | 0.1%  | 0                |       |
| 100.305         | punctate cataract, posterior sutures                           | 0                |       | 4                | 0.8%  |
| 100.307         | punctate cataract, capsular                                    | 2                | 0.2%  | 1                | 0.2%  |
| 100.311         | incipient cataract, anterior cortex                            | 2                | 0.2%  | 4                | 0.8%  |
| 100.312         | incipient cataract, posterior cortex                           | 6                | 0.7%  | 3                | 0.6%  |
| 100.313         | incipient cataract, equatorial cortex                          | 4                | 0.5%  | 1                | 0.2%  |
| 100.314         | incipient cataract, anterior sutures                           | 0                |       | 1                | 0.2%  |
| 100.315         | incipient cataract, posterior sutures                          | 1                | 0.1%  | 2                | 0.4%  |
| 100.317         | incipient cataract, capsular                                   | 2                | 0.2%  | 1                | 0.2%  |
| 100.321         | incomplete cataract, anterior cortex                           | 0                |       | 1                | 0.2%  |
| 100.328         | y-suture tip opacities   | 0                |       | 3                | 0.6%  |
| 100.330         | generalized/complete cataract                                  | 1                | 0.1%  | 0                |       |
| 100.345         | <i>significant cataracts (summary)</i>                         | 26               | 3.1%  | 27               | 5.6%  |
| <b>VITREOUS</b> |  |                  |       |                  |       |
| 110.120         | persistent hyaloid artery/remnant                              | 4                | 0.5%  | 2                | 0.4%  |
| 110.200         | vitreal degeneration-anterior chamber                          | 0                |       | 1                | 0.2%  |
| 110.320         | vitreal degeneration   | 18               | 2.1%  | 5                | 1.0%  |
| <b>RETINA</b>   |  |                  |       |                  |       |
| 120.170         | retinal dysplasia, folds                                       | 5                | 0.6%  | 3                | 0.6%  |
| 120.180         | retinal dysplasia, geographic                                  | 4                | 0.5%  | 2                | 0.4%  |
| 120.310         | generalized progressive retinal atrophy (PRA)                  | 1                | 0.1%  | 2                | 0.4%  |
| <b>OTHER</b>    |  |                  |       |                  |       |
| 900.000         | other, unspecified   | 9                | 1.1%  | 0                |       |
| 900.100         | other, not inherited   | 20               | 2.4%  | 0                |       |
| 900.110         | other. suspect not inherited/significance unknown              | 15               | 1.8%  | 13               | 2.7%  |
| <b>NORMAL</b>   |  |                  |       |                  |       |
| 0.000           | normal globe   | 695              | 82.2% | 375              | 77.5% |



# STANDARD SCHNAUZER

|    | DISORDER                  | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|---------------------------|-------------|-----------|-----------------|
| A. | Distichiasis              | Not defined | 1         | Breeder option  |
| B. | Cataract                  | Not defined | 1         | NO              |
| C. | Y-suture tip opacity      | Not defined | 1         | Breeder option  |
| D. | Retinal dysplasia - folds | Not defined | 1         | Breeder option  |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

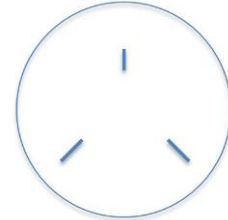
### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membranes, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

There are apparently several forms of cataracts in the Standard Schnauzer: 1) posterior cortex and posterior/total nucleus involvement, with slow progression; 2) dense posterior polar opacity near the sub-capsular region which progresses rapidly to very dense posterior polar plaques in young animals; 3) dense posterior polar opacity like that reported in young animals but found in older animals with variable progression.

### C. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

### D. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Standard Schnauzer breed. The conditions listed are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT STANDARD SCHNAUZER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>3,074 |      | 2016-2020<br>587 |      |
|---------------------|--|--------------------|------|------------------|------|
|                     |  | #                  | %    | #                | %    |
| <b>GLOBE</b>        |  |                    |      |                  |      |
| 0.110               | microphthalmia   | 1                  | 0.0% | 0                |      |
| 10.000              | glaucoma   | 2                  | 0.1% | 0                |      |
| <b>EYELIDS</b>      |  |                    |      |                  |      |
| 20.140              | ectopic cilia  | 0                  |      | 1                | 0.2% |
| 25.110              | distichiasis   | 64                 | 2.1% | 6                | 1.0% |
| <b>NASOLACRIMAL</b> |  |                    |      |                  |      |
| 40.910              | keratoconjunctivitis sicca                                     | 1                  | 0.0% | 0                |      |
| <b>NICTITANS</b>    |  |                    |      |                  |      |
| 51.100              | third eyelid cartilage anomaly                                 | 2                  | 0.1% | 1                | 0.2% |
| 52.110              | prolapsed gland of the third eyelid                            | 2                  | 0.1% | 0                |      |
| <b>CORNEA</b>       |  |                    |      |                  |      |
| 70.700              | corneal dystrophy  | 21                 | 0.7% | 6                | 1.0% |
| 70.730              | corneal endothelial degeneration                               | 1                  | 0.0% | 0                |      |
| <b>UVEA</b>         |  |                    |      |                  |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 14                 | 0.5% | 2                | 0.3% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 3                  | 0.1% | 2                | 0.3% |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 3                  | 0.1% | 0                |      |
| 93.740              | persistent pupillary membranes, iris sheets                    | 2                  | 0.1% | 0                |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 7                  | 0.2% | 7                | 1.2% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 1                  | 0.0% | 0                |      |
| 93.999              | uveal cysts  | 2                  | 0.1% | 0                |      |
| <b>LENS</b>         |  |                    |      |                  |      |
| 100.200             | cataract, unspecified  | 2                  | 0.1% | 0                |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 113                | 3.7% | 31               | 5.3% |
| 100.301             | punctate cataract, anterior cortex                             | 10                 | 0.3% | 6                | 1.0% |
| 100.302             | punctate cataract, posterior cortex                            | 5                  | 0.2% | 4                | 0.7% |
| 100.303             | punctate cataract, equatorial cortex                           | 5                  | 0.2% | 0                |      |
| 100.304             | punctate cataract, anterior sutures                            | 1                  | 0.0% | 1                | 0.2% |
| 100.305             | punctate cataract, posterior sutures                           | 10                 | 0.3% | 4                | 0.7% |
| 100.306             | punctate cataract, nucleus                                     | 4                  | 0.1% | 3                | 0.5% |
| 100.307             | punctate cataract, capsular                                    | 11                 | 0.4% | 2                | 0.3% |
| 100.311             | incipient cataract, anterior cortex                            | 11                 | 0.4% | 4                | 0.7% |
| 100.312             | incipient cataract, posterior cortex                           | 12                 | 0.4% | 2                | 0.3% |
| 100.313             | incipient cataract, equatorial cortex                          | 16                 | 0.5% | 1                | 0.2% |
| 100.314             | incipient cataract, anterior sutures                           | 2                  | 0.1% | 0                |      |
| 100.315             | incipient cataract, posterior sutures                          | 1                  | 0.0% | 1                | 0.2% |
| 100.316             | incipient cataract, nucleus                                    | 9                  | 0.3% | 0                |      |
| 100.317             | incipient cataract, capsular                                   | 4                  | 0.1% | 1                | 0.2% |
| 100.321             | incomplete cataract, anterior cortex                           | 0                  |      | 1                | 0.2% |
| 100.322             | incomplete cataract, posterior cortex                          | 0                  |      | 1                | 0.2% |
| 100.323             | incomplete cataract, equatorial cortex                         | 0                  |      | 1                | 0.2% |
| 100.328             | y-suture tip opacities   | 1                  | 0.0% | 8                | 1.4% |
| 100.330             | generalized/complete cataract                                  | 13                 | 0.4% | 1                | 0.2% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.345 significant cataracts (summary)                   | 117 3.8%         | 41 7.0%          |
| 100.375 subluxation/luxation, unspecified                 | 1 0.0%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 3 0.1%           | 1 0.2%           |
| 110.320 vitreal degeneration                              | 18 0.6%          | 1 0.2%           |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 30 1.0%          | 2 0.3%           |
| 120.180 retinal dysplasia, geographic                     | 4 0.1%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 23 0.7%          | 1 0.2%           |
| 120.910 retinal detachment without dialysis               | 1 0.0%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 5 0.2%           | 1 0.2%           |
| 130.120 optic nerve hypoplasia                            | 3 0.1%           | 1 0.2%           |
| 130.150 optic disc coloboma                               | 0                | 1 0.2%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 31 1.0%          | 0                |
| 900.100 other, not inherited                              | 71 2.3%          | 1 0.2%           |
| 900.110 other. suspect not inherited/significance unknown | 22 0.7%          | 42 7.2%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 2,745 89.3%      | 471 80.2%        |

# SUSSEX SPANIEL

|    | DISORDER                  | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|---------------------------|-------------|-----------|-----------------|
| A. | Ectropion                 | Not defined | 1         | Breeder option  |
| B. | Distichiasis              | Not defined | 1         | Breeder option  |
| C. | Cataract                  | Not defined | 1         | NO              |
| D. | Persistent hyaloid artery | Not defined | 1         | Breeder option  |
| E. | Retinal dysplasia - folds | Not defined | 1         | Breeder option  |

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## Description and Comments

### A. Ectropion

A conformational defect resulting in eversion of the eyelid(s), which may cause ocular irritation due to exposure. It is likely that ectropion is influenced by several genes (polygenic) defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membranes, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### D. Persistent hyaloid artery (PHA)

Congenital defect resulting from abnormalities in the development and regression of the hyaloid artery. The blood vessel remnant can be present in the vitreous as a small patent vascular

strand (PHA) or as a non-vascular strand that appears gray-white (persistent hyaloid remnant).

E. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Sussex Spaniel breed. The conditions listed are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT SUSSEX SPANIEL

| Diagnostic Name    | TOTAL DOGS EXAMINED  | 1991-2015 |      | 2016-2020 |      |
|--------------------|--|-----------|------|-----------|------|
|                    |  | #         | %    | #         | %    |
| <b>EYELIDS</b>     |  |           |      |           |      |
| 20.160             | macropalpebral fissure                                       | 23        | 5.4% | 0         |      |
| 21.000             | entropion, unspecified                                       | 1         | 0.2% | 0         |      |
| 22.000             | ectropion, unspecified                                       | 30        | 7.1% | 3         | 4.5% |
| 25.110             | distichiasis   | 24        | 5.7% | 5         | 7.5% |
| <b>NICTITANS</b>   |  |           |      |           |      |
| 52.110             | prolapsed gland of the third eyelid                          | 0         |      | 1         | 1.5% |
| <b>CORNEA</b>      |  |           |      |           |      |
| 70.700             | corneal dystrophy  | 2         | 0.5% | 0         |      |
| <b>UVEA</b>        |  |           |      |           |      |
| 93.110             | iris hypoplasia  | 2         | 0.5% | 1         | 1.5% |
| 93.150             | iris coloboma  | 7         | 1.7% | 1         | 1.5% |
| 93.710             | persistent pupillary membranes, iris to iris                 | 2         | 0.5% | 3         | 4.5% |
| 93.720             | persistent pupillary membranes, iris to lens                 | 6         | 1.4% | 1         | 1.5% |
| 93.740             | persistent pupillary membranes, iris sheets                  | 1         | 0.2% | 0         |      |
| 93.750             | persistent pupillary membranes, lens pigment foci/no strands | 2         | 0.5% | 1         | 1.5% |
| <b>LENS</b>        |  |           |      |           |      |
| 100.210            | cataract. suspect not inherited/significance unknown         | 15        | 3.5% | 2         | 3.0% |
| 100.302            | punctate cataract, posterior cortex                          | 1         | 0.2% | 0         |      |
| 100.305            | punctate cataract, posterior sutures                         | 0         |      | 1         | 1.5% |
| 100.307            | punctate cataract, capsular                                  | 1         | 0.2% | 0         |      |
| 100.312            | incipient cataract, posterior cortex                         | 2         | 0.5% | 0         |      |
| 100.315            | incipient cataract, posterior sutures                        | 2         | 0.5% | 0         |      |
| 100.316            | incipient cataract, nucleus                                  | 2         | 0.5% | 0         |      |
| 100.317            | incipient cataract, capsular                                 | 4         | 0.9% | 1         | 1.5% |
| 100.322            | incomplete cataract, posterior cortex                        | 0         |      | 1         | 1.5% |
| 100.328            | y-suture tip opacities                                       | 0         |      | 1         | 1.5% |
| 100.330            | generalized/complete cataract                                | 2         | 0.5% | 0         |      |
| 100.345            | <i>significant cataracts (summary)</i>                       | 14        | 3.3% | 4         | 6.0% |
| <b>VITREOUS</b>    |  |           |      |           |      |
| 110.120            | persistent hyaloid artery/remnant                            | 36        | 8.5% | 4         | 6.0% |
| 110.135            | PHPV/PTVL  | 4         | 0.9% | 0         |      |
| 110.320            | vitreal degeneration   | 1         | 0.2% | 0         |      |
| <b>RETINA</b>      |  |           |      |           |      |
| 120.170            | retinal dysplasia, folds                                     | 42        | 9.9% | 3         | 4.5% |
| 120.180            | retinal dysplasia, geographic                                | 2         | 0.5% | 0         |      |
| <b>OPTIC NERVE</b> |  |           |      |           |      |
| 130.110            | micropapilla   | 1         | 0.2% | 0         |      |
| 130.120            | optic nerve hypoplasia                                       | 1         | 0.2% | 0         |      |
| 130.150            | optic disc coloboma  | 3         | 0.7% | 0         |      |
| <b>OTHER</b>       |  |           |      |           |      |
| 900.000            | other, unspecified   | 10        | 2.4% | 0         |      |
| 900.100            | other, not inherited   | 20        | 4.7% | 0         |      |

| <b>OTHER CONTINUED</b>                                    | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 900.110 other. suspect not inherited/significance unknown | 5 1.2%           | 4 6.0%           |
| <b>NORMAL</b><br>0.000 normal globe                       | 265 62.6%        | 48 71.6%         |



# SWEDISH LAPPHUND

|    | DISORDER                           | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE          |
|----|------------------------------------|------------------------|-----------|--------------------|-------------------------------------|
| A. | Retinal atrophy<br>( <i>prcd</i> ) | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>prcd</i> gene |

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## Description and Comments

### A. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Swedish Lapphund is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Swedish Lapphund. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Zangerl B, Goldstein O, Philp AR, et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*. 2006 Nov;88:551-563. PMID: 16938425

# OCULAR DISORDERS REPORT SWEDISH LAPPHUND

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |        | 2016-2020 |       |
|---|---------------------|-----------|--------|-----------|-------|
|   |                     | #         | %      | #         | %     |
| <b>UVEA</b>   |                     | 3         |        | 9         |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 0         |        | 1         | 11.1% |
| <b>LENS</b>   |                     |           |        |           |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 1         | 33.3%  | 0         |       |
| 100.305 punctate cataract, posterior sutures                        |                     | 1         | 33.3%  | 0         |       |
| 100.315 incipient cataract, posterior sutures                       |                     | 1         | 33.3%  | 0         |       |
| 100.328 y-suture tip opacities                                      |                     | 1         | 33.3%  | 0         |       |
| 100.345 significant cataracts (summary)                             |                     | 3         | 100.0% | 0         |       |
| <b>RETINA</b>   |                     |           |        |           |       |
| 120.310 generalized progressive retinal atrophy (PRA)               |                     | 1         | 33.3%  | 0         |       |
| <b>NORMAL</b>   |                     |           |        |           |       |
| 0.000 normal globe  |                     | 0         |        | 8         | 88.9% |

# SWEDISH VALLHUND

|    | DISORDER                                  | INHERITANCE                        | REFERENCE | BREEDING<br>ADVICE         | GENETIC TESTS<br>AVAILABLE           |
|----|---|------------------------------------|-----------|----------------------------|--------------------------------------|
| A. | Distichiasis                              | Not defined                        | 1         | Breeder option             |                                      |
| B. | Corneal dystrophy -<br>epithelial/stromal | Not defined                        | 1         | Breeder option             |                                      |
| C. | Persistent pupillary<br>membranes         |                                    |           |                            |                                      |
|    | - iris to iris                            | Not defined                        | 1         | Breeder option             |                                      |
|    | - iris to lens                            | Not defined                        | 1         | NO                         |                                      |
|    | - lens pigment<br>foci/no strands         | Not defined                        | 1         | Passes with no<br>notation |                                      |
| D. | Cataract                                  | Not defined                        | 1         | NO                         |                                      |
| E. | Y-suture tip opacity                      | Not defined                        | 1         | Breeder option             |                                      |
| F. | Vitreous<br>degeneration                  | Not defined                        | 1         | Breeder option             |                                      |
| G. | Retinopathy                               | Presumed<br>autosomal<br>recessive | 1-4       | NO                         | Mutation in the<br><i>MERTK</i> gene |
| H. | Retinal dysplasia<br>- folds              | Not defined                        | 1         | Breeder option             |                                      |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral. In the Swedish Vallhund, lesions are circular or semicircular central crystalline deposits in the anterior corneal stroma that appear

between 2 and 5 years of age. It may be associated with exophthalmos and lagophthalmos common in these dogs.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

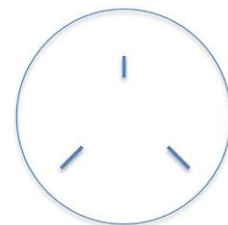
Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

F. Vitreous degeneration

Liquefaction of the vitreous gel which may predispose to retinal detachment.

G. Retinopathy

Swedish Vallhunds have a unique form of retinal degeneration compared to most forms of PRA. The condition is multifocal rather than diffuse and the age of onset and rate of progression vary dramatically, even between littermates. The clinical signs progress in three stages. (A. Komaromy, personal communication 2016)

- Stage one usually occurs between 2-3 years of age and is characterized by mottling or multifocal brown discoloration of the tapetal fundus – this should be marked as retinopathy even though visual deficits are not yet noted.
- In stage two, geographic thinning of the retina can be seen and subtle night vision deficits are observed.
- In stage three, the retinal thinning becomes more generalized with small islands of retinal sparing and deficits are noted in both photopic and scotopic vision. The disease has been associated with a mutation in the *MERTK* gene on canine chromosome 17. Dogs homozygous for the mutation have an 18 fold increased risk of developing the retinopathy. However, the actual causative mutation has not yet been identified.

H. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Cooper AE, Ahonen S, Rowlan JS, et al. A novel form of progressive retinal atrophy in Swedish Vallhund dogs. *PLoS one*. 2014;9:e106610.
3. Ahonen SJ, Arumilli M, Seppala E, et al. Increased expression of MERTK is associated with a unique form of canine retinopathy. *PLoS one*. 2014;9:e114552.
4. Everson R, Pettitt L, Forman OP, et al. An intronic LINE-1 insertion in MERTK is strongly associated with retinopathy in Swedish Vallhund Dogs. *PLoS one*. 2017; 12(8):e0183021

# OCULAR DISORDERS REPORT SWEDISH VALLHUND

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>1,398 |       | 2016-2020<br>343 |       |
|---------------------|--|--------------------|-------|------------------|-------|
|                     |  | #                  | %     | #                | %     |
| <b>EYELIDS</b>      |  |                    |       |                  |       |
| 20.140              | ectopic cilia  | 1                  | 0.1%  | 0                |       |
| 25.110              | distichiasis   | 36                 | 2.6%  | 2                | 0.6%  |
| <b>NASOLACRIMAL</b> |  |                    |       |                  |       |
| 40.910              | keratoconjunctivitis sicca                                     | 1                  | 0.1%  | 0                |       |
| <b>CORNEA</b>       |  |                    |       |                  |       |
| 70.700              | corneal dystrophy  | 16                 | 1.1%  | 7                | 2.0%  |
| <b>UVEA</b>         |  |                    |       |                  |       |
| 93.140              | corneal endothelial pigment without PPM                        | 1                  | 0.1%  | 0                |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 238                | 17.0% | 77               | 22.4% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 7                  | 0.5%  | 3                | 0.9%  |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 2                  | 0.1%  | 1                | 0.3%  |
| 93.740              | persistent pupillary membranes, iris sheets                    | 1                  | 0.1%  | 0                |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 5                  | 0.4%  | 7                | 2.0%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 1                  | 0.1%  | 0                |       |
| 93.810              | uveal melanoma   | 2                  | 0.1%  | 0                |       |
| 93.999              | uveal cysts  | 5                  | 0.4%  | 1                | 0.3%  |
| <b>LENS</b>         |  |                    |       |                  |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 209                | 14.9% | 33               | 9.6%  |
| 100.301             | punctate cataract, anterior cortex                             | 8                  | 0.6%  | 1                | 0.3%  |
| 100.302             | punctate cataract, posterior cortex                            | 3                  | 0.2%  | 0                |       |
| 100.303             | punctate cataract, equatorial cortex                           | 2                  | 0.1%  | 2                | 0.6%  |
| 100.305             | punctate cataract, posterior sutures                           | 10                 | 0.7%  | 3                | 0.9%  |
| 100.306             | punctate cataract, nucleus                                     | 10                 | 0.7%  | 6                | 1.7%  |
| 100.307             | punctate cataract, capsular                                    | 1                  | 0.1%  | 2                | 0.6%  |
| 100.311             | incipient cataract, anterior cortex                            | 14                 | 1.0%  | 7                | 2.0%  |
| 100.312             | incipient cataract, posterior cortex                           | 3                  | 0.2%  | 1                | 0.3%  |
| 100.313             | incipient cataract, equatorial cortex                          | 7                  | 0.5%  | 0                |       |
| 100.314             | incipient cataract, anterior sutures                           | 2                  | 0.1%  | 2                | 0.6%  |
| 100.315             | incipient cataract, posterior sutures                          | 6                  | 0.4%  | 0                |       |
| 100.316             | incipient cataract, nucleus                                    | 13                 | 0.9%  | 2                | 0.6%  |
| 100.321             | incomplete cataract, anterior cortex                           | 1                  | 0.1%  | 1                | 0.3%  |
| 100.322             | incomplete cataract, posterior cortex                          | 0                  |       | 1                | 0.3%  |
| 100.323             | incomplete cataract, equatorial cortex                         | 1                  | 0.1%  | 0                |       |
| 100.328             | y-suture tip opacities   | 0                  |       | 4                | 1.2%  |
| 100.330             | generalized/complete cataract                                  | 7                  | 0.5%  | 0                |       |
| 100.345             | significant cataracts (summary)                                | 88                 | 6.3%  | 32               | 9.3%  |
| <b>VITREOUS</b>     |  |                    |       |                  |       |
| 110.135             | PHPV/PTVL  | 1                  | 0.1%  | 0                |       |
| 110.200             | vitreous degeneration-anterior chamber                         | 1                  | 0.1%  | 1                | 0.3%  |
| 110.320             | vitreal degeneration   | 45                 | 3.2%  | 8                | 2.3%  |
| <b>FUNDUS</b>       |  |                    |       |                  |       |
| 97.110              | choroidal hypoplasia   | 0                  |       | 1                | 0.3%  |

|   | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 23 1.6%          | 3 0.9%           |
| 120.180 retinal dysplasia, geographic                     | 4 0.3%           | 2 0.6%           |
| 120.190 retinal dysplasia, detached                       | 1 0.1%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 45 3.2%          | 2 0.6%           |
| 120.960 retinopathy                                       | 40 2.9%          | 15 4.4%          |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 1 0.1%           | 4 1.2%           |
| 130.150 optic disc coloboma                               | 1 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 47 3.4%          | 0                |
| 900.100 other, not inherited                              | 71 5.1%          | 2 0.6%           |
| 900.110 other. suspect not inherited/significance unknown | 50 3.6%          | 31 9.0%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 897 64.2%        | 167 48.7%        |

# TEDDY ROOSEVELT TERRIER

|    | DISORDER      | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|---------------|------------------------|-----------|--------------------|---|
| A. | Lens luxation | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>ADAMTS17</i> gene |

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## Description and Comments

### A. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Teddy Roosevelt Terrier. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011; 14: 378-384.



# OCULAR DISORDERS REPORT TEDDY ROOSEVELT TERRIER

| Diagnostic Name | TOTAL DOGS EXAMINED                               | 1991-2015 |       | 2016-2020 |       |
|-----------------|---|-----------|-------|-----------|-------|
|                 |   | #         | %     | #         | %     |
| <b>LENS</b>     |   |           |       |           |       |
| 100.311         | incipient cataract, anterior cortex               | 1         | 25.0% | 0         |       |
| 100.312         | incipient cataract, posterior cortex              | 1         | 25.0% | 0         |       |
| 100.313         | incipient cataract, equatorial cortex             | 1         | 25.0% | 0         |       |
| 100.345         | <i>significant cataracts (summary)</i>            | 3         | 75.0% | 0         |       |
| <b>VITREOUS</b> |   |           |       |           |       |
| 110.200         | vitreous degeneration-anterior chamber            | 0         |       | 1         | 50.0% |
| <b>OTHER</b>    |   |           |       |           |       |
| 900.110         | other. suspect not inherited/significance unknown | 2         | 50.0% | 0         |       |
| <b>NORMAL</b>   |   |           |       |           |       |
| 0.000           | normal globe                                      | 1         | 25.0% | 1         | 50.0% |

# TENTERFIELD TERRIER

|    | DISORDER      | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|---------------|------------------------|-----------|--------------------|---|
| A. | Lens luxation | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>ADAMTS17</i> gene |

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## Description and Comments

### A. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Tenterfield Terrier. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011; 14: 378-384.

# TIBETAN MASTIFF

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| B. | Cataract   | Not defined | 1         | NO              |

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## Description and Comments

### A. Persistent pupillary membranes (PPM)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Tibetan Mastiff breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT TIBETAN MASTIFF

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015<br>44 |       | 2016-2020<br>46 |       |
|-----------------|--|-----------------|-------|-----------------|-------|
|                 |  | #               | %     | #               | %     |
| <b>EYELIDS</b>  |  |                 |       |                 |       |
| 21.000          | entropion, unspecified                                       | 3               | 6.8%  | 0               |       |
| 22.000          | ectropion, unspecified                                       | 0               |       | 1               | 2.2%  |
| 25.110          | distichiasis   | 1               | 2.3%  | 4               | 8.7%  |
| <b>CORNEA</b>   |  |                 |       |                 |       |
| 70.700          | corneal dystrophy  | 1               | 2.3%  | 0               |       |
| <b>UVEA</b>     |  |                 |       |                 |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 4               | 9.1%  | 7               | 15.2% |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 5               | 11.4% | 0               |       |
| <b>LENS</b>     |  |                 |       |                 |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 1               | 2.3%  | 2               | 4.3%  |
| 100.301         | punctate cataract, anterior cortex                           | 1               | 2.3%  | 0               |       |
| 100.302         | punctate cataract, posterior cortex                          | 0               |       | 1               | 2.2%  |
| 100.307         | punctate cataract, capsular                                  | 0               |       | 1               | 2.2%  |
| 100.315         | incipient cataract, posterior sutures                        | 0               |       | 1               | 2.2%  |
| 100.317         | incipient cataract, capsular                                 | 0               |       | 2               | 4.3%  |
| 100.345         | <i>significant cataracts (summary)</i>                       | 1               | 2.3%  | 5               | 10.9% |
| <b>VITREOUS</b> |  |                 |       |                 |       |
| 110.120         | persistent hyaloid artery/remnant                            | 0               |       | 1               | 2.2%  |
| <b>OTHER</b>    |  |                 |       |                 |       |
| 900.000         | other, unspecified   | 2               | 4.5%  | 0               |       |
| 900.110         | other. suspect not inherited/significance unknown            | 0               |       | 3               | 6.5%  |
| <b>NORMAL</b>   |  |                 |       |                 |       |
| 0.000           | normal globe   | 32              | 72.7% | 31              | 67.4% |

# TIBETAN SPANIEL

|    | DISORDER                          | INHERITANCE         | REFERENCE | BREEDING ADVICE         | GENETIC TESTS AVAILABLE             |
|----|-----------------------------------|---------------------|-----------|-------------------------|-------------------------------------|
| A. | Entropion                         | Not defined         | 1         | Breeder option          |                                     |
| B. | Distichiasis                      | Not defined         | 1         | Breeder option          |                                     |
| C. | Persistent pupillary membranes    |                     |           |                         |                                     |
|    | - iris to iris                    | Not defined         | 1         | Breeder option          |                                     |
|    | - lens pigment foci/no strands    | Not defined         | 1         | Passes with no notation |                                     |
| D. | Cataract                          | Not defined         | 1         | NO                      |                                     |
| E. | Y-suture tip opacity              | Not defined         | 1         | Breeder option          |                                     |
| F. | Retinal atrophy<br><i>FAM161A</i> | Autosomal recessive | 1-3       | NO                      | Mutation in the <i>FAM161A</i> gene |

## Descriptions and Comments

### A. Entropion

A conformational defect resulting in an "in rolling" of one or more of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic), defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time. It is difficult to make a strong recommendation with regards to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded and breeding discretion is advised.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

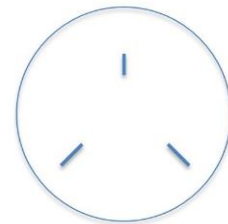
Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

F. Retinal atrophy - *FAM161A*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. In most breeds PRA is inherited as an autosomal recessive trait.

In the Tibetan Spaniel, a mutation in *FAM161A* causes a later onset (4-5 years) of PRA. This form is being called progressive retinal atrophy 3 (PRA3) and appears to be the causative mutation in about 60% of Tibetan Spaniels with PRA. This form is inherited as an autosomal recessive trait. A DNA test for PRA3 is available. This test will not detect PRA caused by other genetic mutations. At least one other form of PRA appears to be present in the Tibetan

Spaniel.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Bjerkas E. Progressive retinal atrophy in dogs in Norway. *Norsk Veterinaertidsskrift*. 1991;103:601-610.
3. Downs LM, Mellersh CS. An Intronic SINE insertion in FAM161A that causes exon-skipping is associated with progressive retinal atrophy in Tibetan Spaniels and Tibetan Terriers. *PLoS One*. 2014;9:e93990.

# OCULAR DISORDERS REPORT TIBETAN SPANIEL

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>3,208 |      | 2016-2020<br>362 |      |
|---|---------------------|--------------------|------|------------------|------|
|   |                     | #                  | %    | #                | %    |
| <b>GLOBE</b>  |                     |                    |      |                  |      |
| 0.110 microphthalmia  |                     | 2                  | 0.1% | 0                |      |
| <b>EYELIDS</b>  |                     |                    |      |                  |      |
| 20.140 ectopic cilia  |                     | 4                  | 0.1% | 0                |      |
| 20.160 macropalpebral fissure   |                     | 5                  | 0.2% | 0                |      |
| 21.000 entropion, unspecified   |                     | 89                 | 2.8% | 3                | 0.8% |
| 22.000 ectropion, unspecified   |                     | 2                  | 0.1% | 0                |      |
| 25.110 distichiasis   |                     | 278                | 8.7% | 22               | 6.1% |
| <b>NASOLACRIMAL</b>   |                     |                    |      |                  |      |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 1                  | 0.0% | 0                |      |
| 40.910 keratoconjunctivitis sicca                                     |                     | 2                  | 0.1% | 1                | 0.3% |
| <b>NICTITANS</b>  |                     |                    |      |                  |      |
| 51.100 third eyelid cartilage anomaly                                 |                     | 2                  | 0.1% | 0                |      |
| 52.110 prolapsed gland of the third eyelid                            |                     | 6                  | 0.2% | 1                | 0.3% |
| <b>CORNEA</b>   |                     |                    |      |                  |      |
| 70.210 corneal pannus   |                     | 8                  | 0.2% | 0                |      |
| 70.220 pigmentary keratitis   |                     | 18                 | 0.6% | 3                | 0.8% |
| 70.700 corneal dystrophy  |                     | 10                 | 0.3% | 0                |      |
| 70.730 corneal endothelial degeneration                               |                     | 1                  | 0.0% | 0                |      |
| <b>UVEA</b>   |                     |                    |      |                  |      |
| 93.110 iris hypoplasia  |                     | 1                  | 0.0% | 0                |      |
| 93.150 iris coloboma  |                     | 4                  | 0.1% | 0                |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 58                 | 1.8% | 11               | 3.0% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 4                  | 0.1% | 1                | 0.3% |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 4                  | 0.1% | 0                |      |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 2                  | 0.1% | 6                | 1.7% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 1                  | 0.0% | 0                |      |
| 93.810 uveal melanoma   |                     | 2                  | 0.1% | 0                |      |
| 93.999 uveal cysts  |                     | 2                  | 0.1% | 1                | 0.3% |
| <b>LENS</b>   |                     |                    |      |                  |      |
| 100.200 cataract, unspecified   |                     | 9                  | 0.3% | 0                |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 80                 | 2.5% | 8                | 2.2% |
| 100.301 punctate cataract, anterior cortex                            |                     | 4                  | 0.1% | 3                | 0.8% |
| 100.302 punctate cataract, posterior cortex                           |                     | 2                  | 0.1% | 0                |      |
| 100.303 punctate cataract, equatorial cortex                          |                     | 2                  | 0.1% | 0                |      |
| 100.304 punctate cataract, anterior sutures                           |                     | 1                  | 0.0% | 0                |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 9                  | 0.3% | 2                | 0.6% |
| 100.306 punctate cataract, nucleus                                    |                     | 1                  | 0.0% | 0                |      |
| 100.307 punctate cataract, capsular                                   |                     | 1                  | 0.0% | 0                |      |
| 100.311 incipient cataract, anterior cortex                           |                     | 21                 | 0.7% | 0                |      |
| 100.312 incipient cataract, posterior cortex                          |                     | 12                 | 0.4% | 0                |      |
| 100.313 incipient cataract, equatorial cortex                         |                     | 6                  | 0.2% | 0                |      |
| 100.314 incipient cataract, anterior sutures                          |                     | 2                  | 0.1% | 0                |      |
| 100.315 incipient cataract, posterior sutures                         |                     | 4                  | 0.1% | 1                | 0.3% |



| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.316 incipient cataract, nucleus                       | 7                | 0.2%  | 0                |       |
| 100.317 incipient cataract, capsular                      | 2                | 0.1%  | 0                |       |
| 100.325 incomplete cataract, posterior sutures            | 1                | 0.0%  | 0                |       |
| 100.328 y-suture tip opacities                            | 5                | 0.2%  | 7                | 1.9%  |
| 100.330 generalized/complete cataract                     | 1                | 0.0%  | 0                |       |
| 100.345 <i>significant cataracts (summary)</i>            | 90               | 2.8%  | 13               | 3.6%  |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 1                | 0.0%  | 0                |       |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 8                | 0.2%  | 1                | 0.3%  |
| 110.135 PHPV/PTVL   | 1                | 0.0%  | 0                |       |
| 110.200 vitreous degeneration-anterior chamber            | 1                | 0.0%  | 1                | 0.3%  |
| 110.320 vitreal degeneration                              | 13               | 0.4%  | 2                | 0.6%  |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 9                | 0.3%  | 0                |       |
| 120.180 retinal dysplasia, geographic                     | 1                | 0.0%  | 3                | 0.8%  |
| 120.190 retinal dysplasia, detached                       | 2                | 0.1%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 28               | 0.9%  | 1                | 0.3%  |
| 120.960 retinopathy                                       | 1                | 0.0%  | 4                | 1.1%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.120 optic nerve hypoplasia                            | 2                | 0.1%  | 0                |       |
| 130.150 optic disc coloboma                               | 7                | 0.2%  | 0                |       |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 32               | 1.0%  | 0                |       |
| 900.100 other, not inherited                              | 75               | 2.3%  | 2                | 0.6%  |
| 900.110 other. suspect not inherited/significance unknown | 32               | 1.0%  | 21               | 5.8%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 2,612            | 81.4% | 280              | 77.3% |

# TIBETAN TERRIER

|    | DISORDER   | INHERITANCE                | REFERENCE  | BREEDING ADVICE                           | GENETIC TESTS AVAILABLE              |
|----|--|----------------------------|------------|---|--------------------------------------|
| A. | Distichiasis   | Not defined                | 1          | Breeder option                            |                                      |
| B. | Corneal dystrophy - epithelial/stromal   | Not defined                | 1          | Breeder option                            |                                      |
| C. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined | 1<br>1     | Breeder option<br>Passes with no notation |                                      |
| D. | Cataract   | Not defined                | 1          | NO  |                                      |
| E. | Lens luxation  | Autosomal recessive        | 1, 2-7     | NO  | Mutation in the <i>ADAMTS17</i> gene |
| F. | Retinal atrophy<br><i>FAM161A</i>  | Autosomal recessive        | 1, 3, 8-11 | NO  | Mutation in the <i>FAM161A</i> gene  |
| G. | Retinal atrophy - Rod-cone dysplasia ( <i>rcd4</i> )                               | Autosomal recessive        | 14         | NO  | Mutation in the <i>C2orf71</i> gene  |
| H. | Ceroid lipofuscinosis  | Not defined                | 13, 14     | NO  |                                      |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time. It is difficult to make a strong recommendation with regards to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded and breeding discretion is advised.

### B. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

F. Retinal atrophy - *FAM161A*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. Except for X-linked PRA in the Siberian Husky and Samoyed, in most breeds studied to date, PRA is inherited as an autosomal recessive trait.

There are ERG studies to indicate that there is depression of the B wave at 10-12 weeks of age in the second variety and slower depression in the first variety. Some may have no obvious signs at 5-6 years of age, only to develop clinical signs at 6-7 years of age. It is logical that any animal found with signs of bilateral atrophy should not be bred. Members of the family of the affected animal should be carefully screened. Perhaps, ERG in animals less than 4 years of age is logical, especially if the animal is intended for breed foundation.

In the Tibetan Terrier a mutation in *FAM161A* causes a later onset (4-5 years) of PRA. This form is being called progressive retinal atrophy 3 (PRA3). This form is inherited as an autosomal recessive trait. A DNA test for PRA3 is available. This test will not detect PRA caused by other genetic mutations. At least one other form of PRA appears to be present in the Tibetan Terrier.

G. Rod-cone dysplasia, type 4 (*rcd4*)

A form of PRA initially identified in the Gordon and Irish Setter breeds. Clinical night blindness is observed on average as late as 10 years of age and progresses to total blindness. This form of PRA has been referred to as late-onset PRA (LOPRA). The disorder is caused by a mutation present in the *C2orf71* gene. A mutation-based gene test is now available that will unequivocally identify genetically normal, affected and carrier dogs. The test is accurate only for this mutation and is of no value in identifying other forms of PRA.

H. Ceroid Lipofuscinosis

An inherited disease of man and animal characterized by the accumulation of lipopigment in various tissues of the body including the eye. It results in progressive neurologic disease. In the Tibetan Terrier, moderate visual impairment can occur in low-light conditions.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Willis MB, Curtis R, Barnett KC, et al. Genetic aspects of lens luxation in the Tibetan Terrier. *Vet Rec.* 1979;104:409-412.
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12. Downs LM, Bell JS, Freeman J, et al. Late-onset progressive retinal atrophy in the Gordon and Irish Setter breeds is associated with a frameshift mutation in C2orf71. *Anim Genet.* 2012;44:169-177.
13. Katz ML, Narfstrom K, Johnson GS, et al. Assessment of retinal function and characterization of lysosomal storage body accumulation in the retinas and brains of Tibetan Terriers with ceroid-lipofuscinosis. *Am J Vet Res.* 2005;66:67-76.
14. Drogemuller C, Wohlke A, Distl O. Characterization of candidate genes for neuronal ceroid lipofuscinosis in dog. *J Hered.* 2005;96:735-738.

# OCULAR DISORDERS REPORT TIBETAN TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED  |     | 1991-2015<br>8,299 |    | 2016-2020<br>1,093 |   |
|---------------------|--|-----|--------------------|----|--------------------|---|
|                     | #  | %   | #                  | %  | #                  | % |
| <b>GLOBE</b>        |  |     |                    |    |                    |   |
| 0.110               | microphthalmia   | 4   | 0.0%               | 0  |                    |   |
| 10.000              | glaucoma   | 3   | 0.0%               | 0  |                    |   |
| <b>EYELIDS</b>      |  |     |                    |    |                    |   |
| 21.000              | entropion, unspecified   | 1   | 0.0%               | 0  |                    |   |
| 25.110              | distichiasis   | 120 | 1.4%               | 5  | 0.5%               |   |
| <b>NASOLACRIMAL</b> |  |     |                    |    |                    |   |
| 32.110              | imperforate lower nasolacrimal punctum                         | 1   | 0.0%               | 4  | 0.4%               |   |
| <b>NICTITANS</b>    |  |     |                    |    |                    |   |
| 52.110              | prolapsed gland of the third eyelid                            | 4   | 0.0%               | 0  |                    |   |
| <b>CORNEA</b>       |  |     |                    |    |                    |   |
| 70.220              | pigmentary keratitis   | 3   | 0.0%               | 1  | 0.1%               |   |
| 70.700              | corneal dystrophy  | 88  | 1.1%               | 8  | 0.7%               |   |
| 70.730              | corneal endothelial degeneration                               | 1   | 0.0%               | 0  |                    |   |
| <b>UVEA</b>         |  |     |                    |    |                    |   |
| 93.710              | persistent pupillary membranes, iris to iris                   | 487 | 5.9%               | 63 | 5.8%               |   |
| 93.720              | persistent pupillary membranes, iris to lens                   | 21  | 0.3%               | 1  | 0.1%               |   |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 40  | 0.5%               | 0  |                    |   |
| 93.740              | persistent pupillary membranes, iris sheets                    | 10  | 0.1%               | 0  |                    |   |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 34  | 0.4%               | 37 | 3.4%               |   |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 11  | 0.1%               | 4  | 0.4%               |   |
| 93.810              | uveal melanoma   | 0   |                    | 1  | 0.1%               |   |
| 93.999              | uveal cysts  | 0   |                    | 1  | 0.1%               |   |
| <b>LENS</b>         |  |     |                    |    |                    |   |
| 100.200             | cataract, unspecified  | 34  | 0.4%               | 0  |                    |   |
| 100.210             | cataract. suspect not inherited/significance unknown           | 380 | 4.6%               | 64 | 5.9%               |   |
| 100.301             | punctate cataract, anterior cortex                             | 71  | 0.9%               | 9  | 0.8%               |   |
| 100.302             | punctate cataract, posterior cortex                            | 36  | 0.4%               | 3  | 0.3%               |   |
| 100.303             | punctate cataract, equatorial cortex                           | 11  | 0.1%               | 5  | 0.5%               |   |
| 100.304             | punctate cataract, anterior sutures                            | 12  | 0.1%               | 0  |                    |   |
| 100.305             | punctate cataract, posterior sutures                           | 6   | 0.1%               | 2  | 0.2%               |   |
| 100.306             | punctate cataract, nucleus                                     | 9   | 0.1%               | 1  | 0.1%               |   |
| 100.307             | punctate cataract, capsular                                    | 12  | 0.1%               | 5  | 0.5%               |   |
| 100.311             | incipient cataract, anterior cortex                            | 62  | 0.7%               | 8  | 0.7%               |   |
| 100.312             | incipient cataract, posterior cortex                           | 67  | 0.8%               | 7  | 0.6%               |   |
| 100.313             | incipient cataract, equatorial cortex                          | 36  | 0.4%               | 2  | 0.2%               |   |
| 100.314             | incipient cataract, anterior sutures                           | 12  | 0.1%               | 1  | 0.1%               |   |
| 100.315             | incipient cataract, posterior sutures                          | 13  | 0.2%               | 1  | 0.1%               |   |
| 100.316             | incipient cataract, nucleus                                    | 9   | 0.1%               | 2  | 0.2%               |   |
| 100.317             | incipient cataract, capsular                                   | 5   | 0.1%               | 0  |                    |   |
| 100.321             | incomplete cataract, anterior cortex                           | 5   | 0.1%               | 6  | 0.5%               |   |
| 100.322             | incomplete cataract, posterior cortex                          | 3   | 0.0%               | 3  | 0.3%               |   |
| 100.323             | incomplete cataract, equatorial cortex                         | 2   | 0.0%               | 3  | 0.3%               |   |
| 100.326             | incomplete cataract, nucleus                                   | 0   |                    | 1  | 0.1%               |   |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> |       | <b>2016-2020</b> |       |
|---|------------------|-------|------------------|-------|
| 100.330 generalized/complete cataract                     | 38               | 0.5%  | 3                | 0.3%  |
| 100.340 resorbing/hypermature cataract                    | 1                | 0.0%  | 1                | 0.1%  |
| 100.345 significant cataracts (summary)                   | 444              | 5.4%  | 63               | 5.8%  |
| 100.375 subluxation/luxation, unspecified                 | 17               | 0.2%  | 0                |       |
| <b>VITREOUS</b>   |                  |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                 | 4                | 0.0%  | 2                | 0.2%  |
| 110.135 PHPV/PTVL   | 2                | 0.0%  | 0                |       |
| 110.320 vitreal degeneration                              | 39               | 0.5%  | 2                | 0.2%  |
| <b>FUNDUS</b>   |                  |       |                  |       |
| 97.110 choroidal hypoplasia                               | 1                | 0.0%  | 0                |       |
| 97.120 coloboma   | 1                | 0.0%  | 0                |       |
| <b>RETINA</b>   |                  |       |                  |       |
| 120.170 retinal dysplasia, folds                          | 10               | 0.1%  | 2                | 0.2%  |
| 120.180 retinal dysplasia, geographic                     | 4                | 0.0%  | 1                | 0.1%  |
| 120.190 retinal dysplasia, detached                       | 4                | 0.0%  | 0                |       |
| 120.310 generalized progressive retinal atrophy (PRA)     | 124              | 1.5%  | 0                |       |
| 120.400 retinal hemorrhage                                | 3                | 0.0%  | 0                |       |
| 120.910 retinal detachment without dialysis               | 3                | 0.0%  | 0                |       |
| 120.920 retinal detachment with dialysis                  | 0                |       | 1                | 0.1%  |
| 120.960 retinopathy                                       | 3                | 0.0%  | 7                | 0.6%  |
| <b>OPTIC NERVE</b>  |                  |       |                  |       |
| 130.110 micropapilla                                      | 2                | 0.0%  | 0                |       |
| 130.120 optic nerve hypoplasia                            | 4                | 0.0%  | 1                | 0.1%  |
| <b>OTHER</b>  |                  |       |                  |       |
| 900.000 other, unspecified                                | 82               | 1.0%  | 0                |       |
| 900.100 other, not inherited                              | 147              | 1.8%  | 2                | 0.2%  |
| 900.110 other. suspect not inherited/significance unknown | 61               | 0.7%  | 43               | 3.9%  |
| <b>NORMAL</b>   |                  |       |                  |       |
| 0.000 normal globe  | 7,077            | 85.3% | 860              | 78.7% |

## TOY AUSTRALIAN SHEPHERD

It is recommended that this breed be examined prior to pharmacological dilation to best facilitate identification of iris coloboma.

|    | DISORDER   | INHERITANCE   | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE         |
|----|--|---|-----------|--------------------|------------------------------------|
| A. | Microphthalmia with multiple ocular defects  | Presumed autosomal recessive with incomplete penetrance | 1-6       | NO                 |                                    |
| B. | Distichiasis   | Not defined   | 1         | Breeder option     |                                    |
| C. | Iris coloboma  | Not defined   | 1         | NO                 |                                    |
| D. | Iris hypoplasia  | Not defined   | 1         | Breeder option     |                                    |
| E. | Persistent pupillary membranes<br>- iris to iris   | Not defined   | 1         | Breeder option     |                                    |
| F. | Cataract   | Autosomal co-dominant                                   | 1, 7, 8   | NO                 | Mutation in the <i>HSF4-2</i> gene |
| G. | Retinal atrophy ( <i>prcd</i> )  | Autosomal recessive                                     | 1, 9      | NO                 | Mutation in the <i>prcd</i> gene   |
| H. | Cone degeneration - day blindness  | Autosomal recessive                                     | 10        | NO                 | Mutation in the <i>CNGB3</i> gene  |
| I. | Multifocal retinopathy - <i>cmr1</i>   | Autosomal recessive                                     | 11        | Breeder option     | Mutation in the <i>BEST1</i> gene  |
| J. | Choroidal hypoplasia (Collie Eye Anomaly)<br>- Optic nerve coloboma<br>- Retinal detachment<br>- Retinal hemorrhage<br>- Staphyloma/coloboma | Autosomal recessive                                     | 1, 12     | NO                 | Mutation in the <i>NHEJ1</i> gene  |



## Description and Comments

### A. Microphthalmia with multiple ocular defects

Microphthalmia is a congenital defect characterized by a small eye with associated defects of the cornea, iris (coloboma), anterior chamber, lens (cataract) and/or retina (dysplasia). In the Australian Shepherd, microphthalmia has long been suspected to be associated with merled coat coloration but a definitive genetic relationship has not been established. The eyes of affected homozygous merle (usually white) dogs have extreme forms of this entity and are usually blind at birth. Affected heterozygous merle-coated dogs demonstrate less severe manifestations.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### C. Iris coloboma

A congenital abnormality in iris development usually characterized by a full-thickness defect in iris tissue, commonly (though not exclusively) located at the 6 o'clock position associated with failure of closure of the optic fissure. A partial-thickness defect in iris tissue should be recorded as iris hypoplasia on the OFA form.

### D. Iris hypoplasia

A congenital abnormality in iris development usually characterized by a reduced quantity of tissue identified as a partial-thickness defect in iris tissue. Full-thickness iris hypoplasia is rare and should be recorded as an iris coloboma on the OFA form.

### E. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### F. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Australian Shepherd, a mutation in *HSF4* (heat shock transcription factor 4), the *HSF4-2* mutation, has been shown to increase the likelihood of cataract formation. The mutation is inherited in a co-dominant manner. Dogs with one copy of the mutation develop bilateral posterior cataracts and homozygotes develop a nuclear cataract that typically progresses to a mature cataract. A DNA test is available for this mutation. Other genetic factors can contribute to cataract formation in this breed and will not be detected by this test.

G. Retinal atrophy - *prcd*

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. With limited exceptions, most PRAs are recessively inherited.

Studies have shown that the principal form of PRA in the Toy Australian Shepherd is *prcd* which is a late-onset form of PRA inherited as autosomal recessive. The mutation is allelic to that present in Miniature Poodles, Labrador Retrievers, English and American Cocker Spaniels, and others. The locus is termed the progressive rod-cone degeneration (*prcd*) gene and at least 30+ breeds are affected. In most affected dogs to date, the disease is recognized clinically in dogs 3-6 years of age or older. This photoreceptor degeneration is characterized by slow death of visual cells following their normal development. The disease begins clinically with signs of night blindness followed by day blindness. A DNA test is available.

H. Cone degeneration - day blindness or hemeralopia

Autosomal recessively inherited early degeneration of the cone photoreceptors. Affected puppies develop day-blindness, colorblindness, and photophobia between 8 and 12 weeks of age. Affected dogs remain ophthalmoscopically normal their entire life. Electroretinography is required to definitively diagnose the disorder. Genetically, the condition results from a mutation in the *CNGB3* gene. A DNA test is available.

I. Multifocal retinopathy

Canine Multifocal Retinopathy type 1 (*cmr1*) is characterized by numerous distinct (i.e. multifocal), roughly circular patches of elevated retina (multifocal bullous retinal detachments). There may be a serous subretinal fluid, or accumulation of subretinal material that produces gray-tan-pink colored lesions. These lesions, looking somewhat like blisters, vary in location and size, although typically they are present in both eyes of the affected dog.

The disease generally develops in young dogs between 11-20 weeks of age and there is minimal progression after 1 year of age. The lesions may flatten, leaving areas of retinal thinning and RPE hypertrophy, hyperplasia, and pigmentation. Discrete areas of tapetal hyper-reflectivity may be seen in areas of previous retinal and RPE detachments. Most dogs exhibit no noticeable problem with vision or electroretinographic abnormalities despite their abnormal appearing retinas.

Canine Multifocal Retinopathy type 1 is caused by a mutation in the Bestrophin 1 gene

(*BEST1*) and is described to be recessively inherited in the Great Pyrenees, Dogue de Bordeaux, Bullmastiff, and Mastiff.

- J. Choroidal hypoplasia (Collie Eye Anomaly)
- staphyloma/coloboma
  - retinal detachment
  - retinal hemorrhage
  - optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly." The choroidal hypoplasia component is caused by a 7799 base pair deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

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1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
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6. Gelatt KN, Powell NG, Huston K. Inheritance of microphthalmia with coloboma in the Australian Shepherd dog. *Am J Vet Res.* 1981;42:1686-1690.
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# OCULAR DISORDERS REPORT TOY AUSTRALIAN SHEPHERD

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |      |
|---|---------------------|-----------|-------|-----------|------|
|   |                     | #         | %     | #         | %    |
| <b>GLOBE</b>  |                     |           |       |           |      |
| 0.110 microphthalmia  |                     | 4         | 0.4%  | 0         |      |
| <b>EYELIDS</b>  |                     |           |       |           |      |
| 25.110 distichiasis   |                     | 41        | 4.4%  | 9         | 5.8% |
| <b>CORNEA</b>   |                     |           |       |           |      |
| 70.700 corneal dystrophy  |                     | 1         | 0.1%  | 3         | 1.9% |
| <b>UVEA</b>   |                     |           |       |           |      |
| 93.110 iris hypoplasia  |                     | 15        | 1.6%  | 7         | 4.5% |
| 93.150 iris coloboma  |                     | 18        | 2.0%  | 1         | 0.6% |
| 93.180 iris sphincter dysplasia                                       |                     | 3         | 0.3%  | 0         |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 103       | 11.2% | 13        | 8.3% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 7         | 0.8%  | 0         |      |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 2         | 0.2%  | 1         | 0.6% |
| 93.740 persistent pupillary membranes, iris sheets                    |                     | 0         |       | 1         | 0.6% |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 1         | 0.1%  | 0         |      |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 0         |       | 1         | 0.6% |
| <b>LENS</b>   |                     |           |       |           |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 12        | 1.3%  | 3         | 1.9% |
| 100.302 punctate cataract, posterior cortex                           |                     | 1         | 0.1%  | 0         |      |
| 100.303 punctate cataract, equatorial cortex                          |                     | 1         | 0.1%  | 0         |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 1         | 0.1%  | 1         | 0.6% |
| 100.306 punctate cataract, nucleus                                    |                     | 0         |       | 1         | 0.6% |
| 100.311 incipient cataract, anterior cortex                           |                     | 4         | 0.4%  | 0         |      |
| 100.312 incipient cataract, posterior cortex                          |                     | 1         | 0.1%  | 0         |      |
| 100.313 incipient cataract, equatorial cortex                         |                     | 2         | 0.2%  | 0         |      |
| 100.317 incipient cataract, capsular                                  |                     | 2         | 0.2%  | 0         |      |
| 100.330 generalized/complete cataract                                 |                     | 1         | 0.1%  | 0         |      |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 13        | 1.4%  | 2         | 1.3% |
| <b>VITREOUS</b>   |                     |           |       |           |      |
| 110.120 persistent hyaloid artery/remnant                             |                     | 5         | 0.5%  | 0         |      |
| 110.135 PHPV/PTVL   |                     | 2         | 0.2%  | 0         |      |
| 110.320 vitreal degeneration  |                     | 2         | 0.2%  | 1         | 0.6% |
| <b>RETINA</b>   |                     |           |       |           |      |
| 120.170 retinal dysplasia, folds                                      |                     | 3         | 0.3%  | 0         |      |
| 120.180 retinal dysplasia, geographic                                 |                     | 1         | 0.1%  | 0         |      |
| 120.310 generalized progressive retinal atrophy (PRA)                 |                     | 1         | 0.1%  | 0         |      |
| <b>OPTIC NERVE</b>  |                     |           |       |           |      |
| 130.110 micropapilla  |                     | 10        | 1.1%  | 1         | 0.6% |
| 130.120 optic nerve hypoplasia  |                     | 2         | 0.2%  | 0         |      |
| <b>OTHER</b>  |                     |           |       |           |      |
| 900.000 other, unspecified  |                     | 6         | 0.7%  | 0         |      |
| 900.100 other, not inherited  |                     | 8         | 0.9%  | 0         |      |

| <b>OTHER CONTINUED</b>                                    | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 900.110 other. suspect not inherited/significance unknown | 6 0.7%           | 5 3.2%           |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 786 85.2%        | 115 73.7%        |

# TOY FOX TERRIER

| DISORDER  | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE              |
|---|---------------------|-----------|-----------------|--------------------------------------|
| A. Persistent pupillary membranes<br>- iris to iris | Not defined         | 1         | Breeder option  |                                      |
| B. Cataract   | Not defined         | 1         | NO              |                                      |
| C. Lens luxation                                    | Autosomal recessive | 2         | NO              | Mutation in the <i>ADAMTS17</i> gene |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

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2. Gould D, Pettitt L, McLaughlin B, et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011 Nov;14:378-384.



# OCULAR DISORDERS REPORT TOY FOX TERRIER

| Diagnostic Name     | TOTAL DOGS EXAMINED                                  | 1991-2015 |       | 2016-2020 |       |
|---------------------|--|-----------|-------|-----------|-------|
|                     |  | #         | %     | #         | %     |
| <b>EYELIDS</b>      |  |           |       |           |       |
| 25.110              | distichiasis   | 2         | 1.0%  | 0         |       |
| <b>NASOLACRIMAL</b> |  |           |       |           |       |
| 32.110              | imperforate lower nasolacrimal punctum               | 0         |       | 1         | 3.1%  |
| <b>CORNEA</b>       |  |           |       |           |       |
| 70.700              | corneal dystrophy                                    | 0         |       | 1         | 3.1%  |
| 70.730              | corneal endothelial degeneration                     | 1         | 0.5%  | 0         |       |
| <b>UVEA</b>         |  |           |       |           |       |
| 93.710              | persistent pupillary membranes, iris to iris         | 19        | 9.8%  | 1         | 3.1%  |
| 93.720              | persistent pupillary membranes, iris to lens         | 2         | 1.0%  | 0         |       |
| 93.730              | persistent pupillary membranes, iris to cornea       | 1         | 0.5%  | 0         |       |
| <b>LENS</b>         |  |           |       |           |       |
| 100.210             | cataract. suspect not inherited/significance unknown | 3         | 1.6%  | 1         | 3.1%  |
| 100.311             | incipient cataract, anterior cortex                  | 5         | 2.6%  | 1         | 3.1%  |
| 100.312             | incipient cataract, posterior cortex                 | 1         | 0.5%  | 1         | 3.1%  |
| 100.313             | incipient cataract, equatorial cortex                | 0         |       | 1         | 3.1%  |
| 100.316             | incipient cataract, nucleus                          | 0         |       | 1         | 3.1%  |
| 100.321             | incomplete cataract, anterior cortex                 | 0         |       | 1         | 3.1%  |
| 100.345             | <i>significant cataracts (summary)</i>               | 6         | 3.1%  | 5         | 15.6% |
| 100.375             | <i>subluxation/luxation, unspecified</i>             | 1         | 0.5%  | 0         |       |
| <b>VITREOUS</b>     |  |           |       |           |       |
| 110.120             | persistent hyaloid artery/remnant                    | 1         | 0.5%  | 0         |       |
| 110.320             | vitreal degeneration                                 | 3         | 1.6%  | 1         | 3.1%  |
| <b>RETINA</b>       |  |           |       |           |       |
| 120.170             | retinal dysplasia, folds                             | 7         | 3.6%  | 0         |       |
| 120.310             | generalized progressive retinal atrophy (PRA)        | 2         | 1.0%  | 0         |       |
| <b>OPTIC NERVE</b>  |  |           |       |           |       |
| 130.120             | optic nerve hypoplasia                               | 2         | 1.0%  | 0         |       |
| <b>OTHER</b>        |  |           |       |           |       |
| 900.000             | other, unspecified                                   | 2         | 1.0%  | 0         |       |
| 900.100             | other, not inherited                                 | 3         | 1.6%  | 0         |       |
| 900.110             | other. suspect not inherited/significance unknown    | 4         | 2.1%  | 3         | 9.4%  |
| <b>NORMAL</b>       |  |           |       |           |       |
| 0.000               | normal globe   | 155       | 80.3% | 25        | 78.1% |

# VIZSLA

|    | <b>DISORDER</b>                           | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b>     |
|----|---|--------------------|------------------|----------------------------|
| A. | Glaucoma                                  | Not defined        | 1, 2             | NO                         |
| B. | Corneal dystrophy<br>- epithelial/stromal | Not defined        | 1                | Breeder option             |
| C. | Persistent pupillary<br>membranes         | Not defined        | 1                | Breeder option             |
|    | - iris to iris                            | Not defined        | 1                | Passes with no<br>notation |
|    | - lens pigment foci/no<br>strands         | Not defined        | 1                |                            |
| D. | Cataract                                  | Not defined        | 1                | NO                         |

## Description and Comments

### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated intraocular pressure occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of the IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine screening exam for certification.

### B. Corneal Dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore is not noted on the certificate.

### D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation,

specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Strom AR, Hassig M, Iburg TM, et al. Epidemiology of canine glaucoma presented to University of Zurich from 1995 to 2009. Part 1: Congenital and primary glaucoma (4 and 123 cases). *Vet Ophthalmol.* 2011 Mar;14:121-126.

# OCULAR DISORDERS REPORT

## VIZSLA

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015<br>2,890 |      | 2016-2020<br>1,286 |      |
|---------------------|--|--------------------|------|--------------------|------|
|                     |  | #                  | %    | #                  | %    |
| <b>EYELIDS</b>      |  |                    |      |                    |      |
| 20.140              | ectopic cilia  | 1                  | 0.0% | 0                  |      |
| 21.000              | entropion, unspecified   | 3                  | 0.1% | 0                  |      |
| 22.000              | ectropion, unspecified   | 3                  | 0.1% | 0                  |      |
| 25.110              | distichiasis   | 25                 | 0.9% | 10                 | 0.8% |
| <b>NASOLACRIMAL</b> |  |                    |      |                    |      |
| 32.110              | imperforate lower nasolacrimal punctum                         | 0                  |      | 1                  | 0.1% |
| 40.910              | keratoconjunctivitis sicca                                     | 1                  | 0.0% | 1                  | 0.1% |
| <b>NICTITANS</b>    |  |                    |      |                    |      |
| 51.100              | third eyelid cartilage anomaly                                 | 4                  | 0.1% | 0                  |      |
| 52.110              | prolapsed gland of the third eyelid                            | 7                  | 0.2% | 0                  |      |
| <b>CORNEA</b>       |  |                    |      |                    |      |
| 70.700              | corneal dystrophy  | 38                 | 1.3% | 17                 | 1.3% |
| 70.730              | corneal endothelial degeneration                               | 2                  | 0.1% | 0                  |      |
| <b>UVEA</b>         |  |                    |      |                    |      |
| 93.710              | persistent pupillary membranes, iris to iris                   | 58                 | 2.0% | 27                 | 2.1% |
| 93.720              | persistent pupillary membranes, iris to lens                   | 12                 | 0.4% | 2                  | 0.2% |
| 93.740              | persistent pupillary membranes, iris sheets                    | 1                  | 0.0% | 0                  |      |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 89                 | 3.1% | 100                | 7.8% |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 1                  | 0.0% | 2                  | 0.2% |
| 93.999              | uveal cysts  | 2                  | 0.1% | 0                  |      |
| <b>LENS</b>         |  |                    |      |                    |      |
| 100.200             | cataract, unspecified  | 4                  | 0.1% | 0                  |      |
| 100.210             | cataract. suspect not inherited/significance unknown           | 105                | 3.6% | 34                 | 2.6% |
| 100.301             | punctate cataract, anterior cortex                             | 10                 | 0.3% | 2                  | 0.2% |
| 100.302             | punctate cataract, posterior cortex                            | 15                 | 0.5% | 6                  | 0.5% |
| 100.303             | punctate cataract, equatorial cortex                           | 2                  | 0.1% | 1                  | 0.1% |
| 100.305             | punctate cataract, posterior sutures                           | 4                  | 0.1% | 1                  | 0.1% |
| 100.307             | punctate cataract, capsular                                    | 8                  | 0.3% | 6                  | 0.5% |
| 100.311             | incipient cataract, anterior cortex                            | 16                 | 0.6% | 4                  | 0.3% |
| 100.312             | incipient cataract, posterior cortex                           | 19                 | 0.7% | 16                 | 1.2% |
| 100.313             | incipient cataract, equatorial cortex                          | 19                 | 0.7% | 1                  | 0.1% |
| 100.314             | incipient cataract, anterior sutures                           | 1                  | 0.0% | 1                  | 0.1% |
| 100.315             | incipient cataract, posterior sutures                          | 4                  | 0.1% | 0                  |      |
| 100.316             | incipient cataract, nucleus                                    | 2                  | 0.1% | 1                  | 0.1% |
| 100.317             | incipient cataract, capsular                                   | 6                  | 0.2% | 3                  | 0.2% |
| 100.326             | incomplete cataract, nucleus                                   | 0                  |      | 1                  | 0.1% |
| 100.328             | y-suture tip opacities   | 2                  | 0.1% | 1                  | 0.1% |
| 100.330             | generalized/complete cataract                                  | 2                  | 0.1% | 0                  |      |
| 100.345             | significant cataracts (summary)                                | 114                | 3.9% | 44                 | 3.4% |
| 100.375             | subluxation/luxation, unspecified                              | 2                  | 0.1% | 0                  |      |
| <b>VITREOUS</b>     |  |                    |      |                    |      |
| 110.120             | persistent hyaloid artery/remnant                              | 3                  | 0.1% | 2                  | 0.2% |
| 110.135             | PHPV/PTVL  | 1                  | 0.0% | 0                  |      |

| <b>VITREOUS CONTINUED</b>                                 | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 110.200 vitreous degeneration-anterior chamber            | 1 0.0%           | 6 0.5%           |
| 110.320 vitreal degeneration                              | 13 0.4%          | 5 0.4%           |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 3 0.1%           | 2 0.2%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 5 0.2%           | 0                |
| 120.960 retinopathy                                       | 2 0.1%           | 4 0.3%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.120 optic nerve hypoplasia                            | 1 0.0%           | 1 0.1%           |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 51 1.8%          | 0                |
| 900.100 other, not inherited                              | 73 2.5%          | 1 0.1%           |
| 900.110 other. suspect not inherited/significance unknown | 50 1.7%          | 58 4.5%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 2,520 87.2%      | 1,023 79.5%      |

# VOLPINO ITALIANO

|    | DISORDER      | INHERITANCE            | REFERENCE | BREEDING<br>ADVICE | GENETIC TESTS<br>AVAILABLE              |
|----|---------------|------------------------|-----------|--------------------|---|
| A. | Lens luxation | Autosomal<br>recessive | 1         | NO                 | Mutation in the<br><i>ADAMTS17</i> gene |

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## Description and Comments

### A. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma), causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

There are no breed eye screening examination statistics providing detailed descriptions of hereditary ocular conditions of the Volpino Italiano. The condition listed above is currently noted solely due to the availability of a genetic test for the disease.

1. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011; 14: 378-384.

# OCULAR DISORDERS REPORT VOLPINO ITALIANO

| Diagnostic Name                     | TOTAL DOGS EXAMINED | 1991-2015 |        | 2016-2020 |   |
|-------------------------------------|---------------------|-----------|--------|-----------|---|
|                                     |                     | #         | %      | #         | % |
| <b>NORMAL</b><br>0.000 normal globe |                     | 1         |        | 0         |   |
|                                     |                     | 1         | 100.0% | 0         |   |

# WEIMARANER

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE         | GENETIC TESTS AVAILABLE          |
|----|--|-------------|-----------|-------------------------|----------------------------------|
| A. | Distichiasis   | Not defined | 1         | Breeder option          |                                  |
| B. | Corneal dystrophy<br>- epithelial/stromal                        | Not defined | 1         | Breeder option          |                                  |
| C. | Persistent pupillary membranes<br>- lens pigment foci/no strands | Not defined | 1         | Passes with no notation |                                  |
| D. | Cataract   | Not defined | 1         | NO                      |                                  |
| E. | Retinal atrophy<br>- generalized                                 | X-linked    | 1, 2      | NO                      | Mutation in the <i>RPGR</i> gene |

## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regards to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded. Breeding discretion is advised.

In the Weimaraner, because there is significant clinical disease associated with the abnormal hairs, breeding should be discouraged.

### B. Corneal Dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

### C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted



on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. PRA is inherited as an autosomal recessive trait in most breeds.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Kropatsch R, Akkad D, Frank M, et al. A large deletion in RPGR causes XLPRA in Weimarener dogs. *Canine Genetics and Epidemiol.* 2016; 3:7.

# OCULAR DISORDERS REPORT WEIMARANER

| Diagnostic Name     | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|---------------------|--|-----------|-------|-----------|-------|
|                     |  | #         | %     | #         | %     |
| <b>EYELIDS</b>      |  |           |       |           |       |
| 21.000              | entropion, unspecified   | 3         | 0.2%  | 0         |       |
| 25.110              | distichiasis   | 492       | 29.7% | 131       | 25.7% |
| <b>NASOLACRIMAL</b> |  |           |       |           |       |
| 32.110              | imperforate lower nasolacrimal punctum                         | 0         |       | 2         | 0.4%  |
| <b>NICTITANS</b>    |  |           |       |           |       |
| 51.100              | third eyelid cartilage anomaly                                 | 13        | 0.8%  | 4         | 0.8%  |
| <b>CORNEA</b>       |  |           |       |           |       |
| 70.700              | corneal dystrophy  | 29        | 1.8%  | 7         | 1.4%  |
| 70.730              | corneal endothelial degeneration                               | 5         | 0.3%  | 1         | 0.2%  |
| <b>UVEA</b>         |  |           |       |           |       |
| 93.150              | iris coloboma  | 2         | 0.1%  | 0         |       |
| 93.710              | persistent pupillary membranes, iris to iris                   | 15        | 0.9%  | 3         | 0.6%  |
| 93.720              | persistent pupillary membranes, iris to lens                   | 3         | 0.2%  | 0         |       |
| 93.730              | persistent pupillary membranes, iris to cornea                 | 5         | 0.3%  | 0         |       |
| 93.750              | persistent pupillary membranes, lens pigment foci/no strands   | 0         |       | 9         | 1.8%  |
| 93.760              | persistent pupillary membranes, endothelial opacity/no strands | 2         | 0.1%  | 0         |       |
| 93.810              | uveal melanoma   | 1         | 0.1%  | 0         |       |
| 93.999              | uveal cysts  | 5         | 0.3%  | 3         | 0.6%  |
| <b>LENS</b>         |  |           |       |           |       |
| 100.200             | cataract, unspecified  | 2         | 0.1%  | 0         |       |
| 100.210             | cataract. suspect not inherited/significance unknown           | 92        | 5.6%  | 39        | 7.7%  |
| 100.301             | punctate cataract, anterior cortex                             | 10        | 0.6%  | 7         | 1.4%  |
| 100.302             | punctate cataract, posterior cortex                            | 5         | 0.3%  | 1         | 0.2%  |
| 100.303             | punctate cataract, equatorial cortex                           | 8         | 0.5%  | 2         | 0.4%  |
| 100.304             | punctate cataract, anterior sutures                            | 1         | 0.1%  | 0         |       |
| 100.305             | punctate cataract, posterior sutures                           | 1         | 0.1%  | 1         | 0.2%  |
| 100.306             | punctate cataract, nucleus                                     | 7         | 0.4%  | 4         | 0.8%  |
| 100.307             | punctate cataract, capsular                                    | 1         | 0.1%  | 2         | 0.4%  |
| 100.311             | incipient cataract, anterior cortex                            | 40        | 2.4%  | 8         | 1.6%  |
| 100.312             | incipient cataract, posterior cortex                           | 11        | 0.7%  | 5         | 1.0%  |
| 100.313             | incipient cataract, equatorial cortex                          | 12        | 0.7%  | 8         | 1.6%  |
| 100.314             | incipient cataract, anterior sutures                           | 2         | 0.1%  | 1         | 0.2%  |
| 100.315             | incipient cataract, posterior sutures                          | 2         | 0.1%  | 0         |       |
| 100.316             | incipient cataract, nucleus                                    | 4         | 0.2%  | 4         | 0.8%  |
| 100.317             | incipient cataract, capsular                                   | 1         | 0.1%  | 4         | 0.8%  |
| 100.321             | incomplete cataract, anterior cortex                           | 1         | 0.1%  | 2         | 0.4%  |
| 100.322             | incomplete cataract, posterior cortex                          | 0         |       | 1         | 0.2%  |
| 100.323             | incomplete cataract, equatorial cortex                         | 0         |       | 1         | 0.2%  |
| 100.326             | incomplete cataract, nucleus                                   | 0         |       | 1         | 0.2%  |
| 100.330             | generalized/complete cataract                                  | 5         | 0.3%  | 0         |       |
| 100.345             | <i>significant cataracts (summary)</i>                         | 113       | 6.8%  | 52        | 10.2% |
| 100.375             | <i>subluxation/luxation, unspecified</i>                       | 1         | 0.1%  | 1         | 0.2%  |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>VITREOUS</b>   |             |           |
| 110.120 persistent hyaloid artery/remnant                 | 4 0.2%      | 1 0.2%    |
| 110.135 PHPV/PTVL   | 0           | 2 0.4%    |
| 110.200 vitreous degeneration-anterior chamber            | 1 0.1%      | 2 0.4%    |
| 110.320 vitreal degeneration                              | 2 0.1%      | 1 0.2%    |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 2 0.1%      | 0         |
| 120.180 retinal dysplasia, geographic                     | 4 0.2%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 6 0.4%      | 0         |
| 120.400 retinal hemorrhage                                | 1 0.1%      | 0         |
| 120.960 retinopathy                                       | 1 0.1%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 12 0.7%     | 0         |
| 900.100 other, not inherited                              | 50 3.0%     | 5 1.0%    |
| 900.110 other. suspect not inherited/significance unknown | 16 1.0%     | 25 4.9%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,066 64.4% | 290 57.0% |

# WELSH SPRINGER SPANIEL

|    | DISORDER  | INHERITANCE                        | REFERENCE | BREEDING ADVICE |
|----|---|------------------------------------|-----------|-----------------|
| A. | Glaucoma  | Presumed<br>autosomal<br>dominant  | 1-4       | NO              |
| B. | Entropion   | Not defined                        | 1         | Breeder option  |
| C. | Distichiasis  | Not defined                        | 1         | Breeder option  |
| D. | Corneal dystrophy<br>- epithelial/stromal           | Not defined                        | 1         | Breeder option  |
| E. | Persistent pupillary<br>membranes<br>- iris to iris | Not defined                        | 1         | Breeder option  |
| F. | Cataract  | Presumed<br>autosomal<br>recessive | 1, 5, 6   | NO              |
| G. | Retinal dysplasia<br>- folds                        | Not defined                        | 1         | Breeder option  |

## Description and Comments

### A. Glaucoma

An elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests is part of a routine breed eye screening exam. Due to the increased incidence of PLD in the breed and the increased progression observed with age, it may be prudent to perform repeated gonioscopy examinations over time.

Primary angle closure glaucoma has been reported in the Welsh Springer Spaniel. Females are affected more than males. Onset ranges from 10 weeks to 10 years. At the iridocorneal angle, the pectinate ligaments appear sparse and wispy in contrast to the sturdy fibers seen in other breeds. A dominant mode of inheritance is reported.

### B. Entropion

A conformational defect resulting in an "in-rolling" of one or both of the eyelids which may cause ocular irritation. It is likely that entropion is influenced by several genes (polygenic),

defining the skin and other structures which make up the eyelids, the amount and weight of the skin covering the head and face, the orbital contents, and the conformation of the skull.

C. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established, although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

D. Corneal Dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral.

E. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

F. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

In the Welsh Springer Spaniel, lesions may be seen as early as 8-12 weeks of age and progress rapidly to complete cataract, impairing vision. A recessive mode of inheritance is reported.

G. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

2. Cottrell B, Barnett K. Primary glaucoma in the Welsh Springer Spaniel. *J Small Anim Pract.* 1988;29:185-199.
3. Gelatt KN, MacKay EO. Prevalence of the breed-related glaucomas in pure-bred dogs in North America. *Vet Ophthalmol.* 2004;7:97-111. Epub 2004/02/26.
4. Oliver JA, Ekiri A, Mellersh. Prevalence and Progression of Pectinate Ligament Dysplasia in the Welsh Springer Spaniel. *J Sm Anim Pract.* 2016;57: 416-421.
5. Barnett KC. Hereditary cataract in the Welsh Springer Spaniel. *J Small Anim Pract.* 1980;21:621-625. Epub 1980/11/01.
6. Barnett KC. The diagnosis and differential diagnosis of cataract in the dog. *J Small Anim Pract.* 1985;26:305.

# OCULAR DISORDERS REPORT WELSH SPRINGER SPANIEL

| Diagnostic Name   | TOTAL DOGS EXAMINED |       | 1991-2015<br>2,576 |       | 2016-2020<br>644 |   |
|---|---------------------|-------|--------------------|-------|------------------|---|
|   | #                   | %     | #                  | %     | #                | % |
| <b>GLOBE</b>  |                     |       |                    |       |                  |   |
| 10.000 glaucoma   | 1                   | 0.0%  | 0                  |       |                  |   |
| <b>EYELIDS</b>  |                     |       |                    |       |                  |   |
| 21.000 entropion, unspecified                                       | 39                  | 1.5%  | 16                 | 2.5%  |                  |   |
| 22.000 ectropion, unspecified                                       | 3                   | 0.1%  | 0                  |       |                  |   |
| 25.110 distichiasis   | 299                 | 11.6% | 108                | 16.8% |                  |   |
| <b>NASOLACRIMAL</b>   |                     |       |                    |       |                  |   |
| 32.110 imperforate lower nasolacrimal punctum                       | 1                   | 0.0%  | 2                  | 0.3%  |                  |   |
| <b>CORNEA</b>   |                     |       |                    |       |                  |   |
| 70.700 corneal dystrophy  | 43                  | 1.7%  | 18                 | 2.8%  |                  |   |
| 70.730 corneal endothelial degeneration                             | 2                   | 0.1%  | 0                  |       |                  |   |
| <b>UVEA</b>   |                     |       |                    |       |                  |   |
| 93.150 iris coloboma  | 1                   | 0.0%  | 0                  |       |                  |   |
| 93.710 persistent pupillary membranes, iris to iris                 | 579                 | 22.5% | 170                | 26.4% |                  |   |
| 93.720 persistent pupillary membranes, iris to lens                 | 2                   | 0.1%  | 2                  | 0.3%  |                  |   |
| 93.730 persistent pupillary membranes, iris to cornea               | 1                   | 0.0%  | 0                  |       |                  |   |
| 93.740 persistent pupillary membranes, iris sheets                  | 1                   | 0.0%  | 0                  |       |                  |   |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands | 4                   | 0.2%  | 6                  | 0.9%  |                  |   |
| 93.999 uveal cysts  | 2                   | 0.1%  | 2                  | 0.3%  |                  |   |
| 97.150 chorioretinal coloboma, congenital                           | 0                   |       | 1                  | 0.2%  |                  |   |
| <b>LENS</b>   |                     |       |                    |       |                  |   |
| 100.200 cataract, unspecified                                       | 6                   | 0.2%  | 0                  |       |                  |   |
| 100.210 cataract. suspect not inherited/significance unknown        | 130                 | 5.0%  | 28                 | 4.3%  |                  |   |
| 100.301 punctate cataract, anterior cortex                          | 11                  | 0.4%  | 7                  | 1.1%  |                  |   |
| 100.302 punctate cataract, posterior cortex                         | 3                   | 0.1%  | 3                  | 0.5%  |                  |   |
| 100.303 punctate cataract, equatorial cortex                        | 1                   | 0.0%  | 1                  | 0.2%  |                  |   |
| 100.304 punctate cataract, anterior sutures                         | 2                   | 0.1%  | 1                  | 0.2%  |                  |   |
| 100.306 punctate cataract, nucleus                                  | 1                   | 0.0%  | 2                  | 0.3%  |                  |   |
| 100.307 punctate cataract, capsular                                 | 1                   | 0.0%  | 3                  | 0.5%  |                  |   |
| 100.311 incipient cataract, anterior cortex                         | 4                   | 0.2%  | 0                  |       |                  |   |
| 100.312 incipient cataract, posterior cortex                        | 2                   | 0.1%  | 2                  | 0.3%  |                  |   |
| 100.313 incipient cataract, equatorial cortex                       | 2                   | 0.1%  | 2                  | 0.3%  |                  |   |
| 100.316 incipient cataract, nucleus                                 | 2                   | 0.1%  | 0                  |       |                  |   |
| 100.317 incipient cataract, capsular                                | 2                   | 0.1%  | 0                  |       |                  |   |
| 100.321 incomplete cataract, anterior cortex                        | 0                   |       | 1                  | 0.2%  |                  |   |
| 100.330 generalized/complete cataract                               | 1                   | 0.0%  | 0                  |       |                  |   |
| 100.345 <i>significant cataracts (summary)</i>                      | 38                  | 1.5%  | 22                 | 3.4%  |                  |   |
| 100.375 <i>subluxation/luxation, unspecified</i>                    | 1                   | 0.0%  | 0                  |       |                  |   |
| <b>VITREOUS</b>   |                     |       |                    |       |                  |   |
| 110.120 persistent hyaloid artery/remnant                           | 10                  | 0.4%  | 1                  | 0.2%  |                  |   |
| 110.135 PHPV/PTVL   | 1                   | 0.0%  | 0                  |       |                  |   |
| 110.320 vitreal degeneration  | 5                   | 0.2%  | 0                  |       |                  |   |

|   | 1991-2015   | 2016-2020 |
|---|-------------|-----------|
| <b>FUNDUS</b>   |             |           |
| 97.120 coloboma   | 2 0.1%      | 0         |
| <b>RETINA</b>   |             |           |
| 120.170 retinal dysplasia, folds                          | 29 1.1%     | 5 0.8%    |
| 120.180 retinal dysplasia, geographic                     | 4 0.2%      | 0         |
| 120.310 generalized progressive retinal atrophy (PRA)     | 8 0.3%      | 1 0.2%    |
| <b>OPTIC NERVE</b>  |             |           |
| 130.110 micropapilla                                      | 3 0.1%      | 0         |
| 130.120 optic nerve hypoplasia                            | 6 0.2%      | 1 0.2%    |
| 130.150 optic disc coloboma                               | 4 0.2%      | 0         |
| <b>OTHER</b>  |             |           |
| 900.000 other, unspecified                                | 19 0.7%     | 0         |
| 900.100 other, not inherited                              | 50 1.9%     | 4 0.6%    |
| 900.110 other. suspect not inherited/significance unknown | 20 0.8%     | 25 3.9%   |
| <b>NORMAL</b>   |             |           |
| 0.000 normal globe  | 1,718 66.7% | 332 51.6% |



# WELSH TERRIER

| DISORDER  | INHERITANCE            | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE                 |
|---|------------------------|-----------|-----------------|---|
| A. Persistent pupillary membranes<br>- iris to iris | Not defined            | 1         | Breeder option  |   |
| B. Lens luxation                                    | Autosomal<br>recessive | 1, 2      | NO              | Mutation in the<br><i>ADAMTS17</i> gene |

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## Description and Comment

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Gould D, Pettitt L, McLaughlin B, et al. *ADAMTS17* mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011 Nov;14:378-384.

# OCULAR DISORDERS REPORT WELSH TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 10.000 glaucoma   |                     | 1         | 0.3%  | 0         |       |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 20.140 ectopic cilia  |                     | 1         | 0.3%  | 0         |       |
| 25.110 distichiasis   |                     | 12        | 3.3%  | 1         | 2.5%  |
| <b>NASOLACRIMAL</b>   |                     |           |       |           |       |
| 40.910 keratoconjunctivitis sicca                                   |                     | 1         | 0.3%  | 0         |       |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.700 corneal dystrophy  |                     | 4         | 1.1%  | 0         |       |
| 70.730 corneal endothelial degeneration                             |                     | 3         | 0.8%  | 1         | 2.5%  |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 30        | 8.4%  | 1         | 2.5%  |
| 93.720 persistent pupillary membranes, iris to lens                 |                     | 2         | 0.6%  | 0         |       |
| 93.730 persistent pupillary membranes, iris to cornea               |                     | 3         | 0.8%  | 0         |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 4         | 1.1%  | 7         | 17.5% |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.200 cataract, unspecified                                       |                     | 1         | 0.3%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 22        | 6.1%  | 0         |       |
| 100.301 punctate cataract, anterior cortex                          |                     | 2         | 0.6%  | 0         |       |
| 100.302 punctate cataract, posterior cortex                         |                     | 2         | 0.6%  | 0         |       |
| 100.307 punctate cataract, capsular                                 |                     | 1         | 0.3%  | 0         |       |
| 100.311 incipient cataract, anterior cortex                         |                     | 3         | 0.8%  | 0         |       |
| 100.312 incipient cataract, posterior cortex                        |                     | 2         | 0.6%  | 0         |       |
| 100.313 incipient cataract, equatorial cortex                       |                     | 1         | 0.3%  | 0         |       |
| 100.317 incipient cataract, capsular                                |                     | 2         | 0.6%  | 0         |       |
| 100.328 y-suture tip opacities                                      |                     | 0         |       | 3         | 7.5%  |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 14        | 3.9%  | 3         | 7.5%  |
| 100.375 <i>subluxation/luxation, unspecified</i>                    |                     | 3         | 0.8%  | 0         |       |
| <b>RETINA</b>   |                     |           |       |           |       |
| 120.170 retinal dysplasia, folds                                    |                     | 1         | 0.3%  | 0         |       |
| <b>OTHER</b>  |                     |           |       |           |       |
| 900.000 other, unspecified  |                     | 6         | 1.7%  | 0         |       |
| 900.100 other, not inherited  |                     | 13        | 3.6%  | 0         |       |
| 900.110 other. suspect not inherited/significance unknown           |                     | 1         | 0.3%  | 4         | 10.0% |
| <b>NORMAL</b>   |                     |           |       |           |       |
| 0.000 normal globe  |                     | 291       | 81.1% | 28        | 70.0% |

# WEST HIGHLAND WHITE TERRIER

|    | DISORDER   | INHERITANCE                  | REFERENCE | BREEDING<br>ADVICE                        |
|----|--|------------------------------|-----------|---|
| A. | Keratoconjunctivitis sicca   | Not defined                  | 1-4       | NO  |
| B. | Persistent pupillary membranes<br>- iris to iris<br>- lens pigment foci/no strands | Not defined<br>Not defined   | 1, 5<br>1 | Breeder option<br>Passes with no notation |
| C. | Cataract   | Presumed autosomal recessive | 1, 5      | NO  |
| D. | Y-suture tip opacity   | Not defined                  | 1         | Breeder option                            |
| E. | Retinal dysplasia<br>- folds   | Not defined                  | 1         | Breeder option                            |

## Description and Comments

### A. Keratoconjunctivitis sicca

An abnormality of the tear film, most commonly a deficiency of the aqueous portion, although the mucin and/or lipid layers may be affected; results in ocular irritation and/or vision impairment.

In the West Highland White Terrier, this disease has been reported more commonly in females than males.

### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

In the West Highland White Terrier, these membranes, when present, often bridge from the iris to the lens and may result in cataract with vision impairment.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

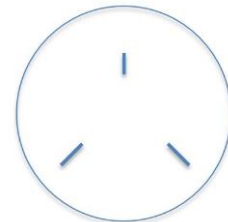
### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

The cataract described in the West Highland White Terrier initially involves the posterior Y sutures and may infrequently progress, resulting in vision impairment. The age of onset is less than 6 months of age. A recessive mode of inheritance is suggested by the pedigrees which have been studied.

### D. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

### E. Retinal dysplasia - folds

Linear, triangular, curved or curvilinear foci of retinal folding that may be single or multiple. When seen in puppies, this condition may partially or completely resolve with maturity. Its significance to vision is unknown. There are two other forms of retinal dysplasia (geographic, detached) which are known to be inherited in other breeds and, in their most severe form, cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Sansom J, Barnett KC, Neumann W, et al. Treatment of keratoconjunctivitis sicca in dogs with cyclosporine ophthalmic ointment: a European clinical field trial. *Vet Rec.* 1995; 137: 504-507.
3. Baker GJ, Formston C. An evaluation of transplantation of the parotid duct in the treatment of kerato-conjunctivitis sicca in the dog. *J Small Anim Pract.* 1968; 9: 261-268.
4. Kaswan RL, Martin CL, Chapman WL, Jr. Keratoconjunctivitis sicca: histopathologic study of nictitating membrane and lacrimal glands from 28 dogs. *Am J Vet Res.* 1984; 45: 112-118.
5. Narfstrom K. Cataract in the West Highland White Terrier. *J Small Anim Pract.* 1981; 22: 467-471.

# OCULAR DISORDERS REPORT WEST HIGHLAND WHITE TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>1,379 |       | 2016-2020<br>392 |       |
|---|---------------------|--------------------|-------|------------------|-------|
|   |                     | #                  | %     | #                | %     |
| <b>GLOBE</b>  |                     |                    |       |                  |       |
| 0.110 microphthalmia  |                     | 5                  | 0.4%  | 0                |       |
| <b>EYELIDS</b>  |                     |                    |       |                  |       |
| 25.110 distichiasis   |                     | 2                  | 0.1%  | 1                | 0.3%  |
| <b>NASOLACRIMAL</b>   |                     |                    |       |                  |       |
| 32.110 imperforate lower nasolacrimal punctum                         |                     | 0                  |       | 2                | 0.5%  |
| 40.910 keratoconjunctivitis sicca                                     |                     | 3                  | 0.2%  | 0                |       |
| <b>CORNEA</b>   |                     |                    |       |                  |       |
| 70.210 corneal pannus   |                     | 1                  | 0.1%  | 0                |       |
| 70.700 corneal dystrophy  |                     | 1                  | 0.1%  | 0                |       |
| 70.730 corneal endothelial degeneration                               |                     | 3                  | 0.2%  | 0                |       |
| <b>UVEA</b>   |                     |                    |       |                  |       |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 117                | 8.5%  | 29               | 7.4%  |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 21                 | 1.5%  | 3                | 0.8%  |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 5                  | 0.4%  | 1                | 0.3%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 16                 | 1.2%  | 7                | 1.8%  |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 4                  | 0.3%  | 0                |       |
| <b>LENS</b>   |                     |                    |       |                  |       |
| 100.200 cataract, unspecified   |                     | 21                 | 1.5%  | 0                |       |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 112                | 8.1%  | 34               | 8.7%  |
| 100.301 punctate cataract, anterior cortex                            |                     | 18                 | 1.3%  | 4                | 1.0%  |
| 100.302 punctate cataract, posterior cortex                           |                     | 9                  | 0.7%  | 2                | 0.5%  |
| 100.303 punctate cataract, equatorial cortex                          |                     | 3                  | 0.2%  | 2                | 0.5%  |
| 100.304 punctate cataract, anterior sutures                           |                     | 1                  | 0.1%  | 0                |       |
| 100.305 punctate cataract, posterior sutures                          |                     | 17                 | 1.2%  | 5                | 1.3%  |
| 100.306 punctate cataract, nucleus                                    |                     | 9                  | 0.7%  | 2                | 0.5%  |
| 100.307 punctate cataract, capsular                                   |                     | 9                  | 0.7%  | 3                | 0.8%  |
| 100.311 incipient cataract, anterior cortex                           |                     | 35                 | 2.5%  | 2                | 0.5%  |
| 100.312 incipient cataract, posterior cortex                          |                     | 22                 | 1.6%  | 4                | 1.0%  |
| 100.313 incipient cataract, equatorial cortex                         |                     | 5                  | 0.4%  | 0                |       |
| 100.314 incipient cataract, anterior sutures                          |                     | 2                  | 0.1%  | 0                |       |
| 100.315 incipient cataract, posterior sutures                         |                     | 5                  | 0.4%  | 0                |       |
| 100.316 incipient cataract, nucleus                                   |                     | 14                 | 1.0%  | 1                | 0.3%  |
| 100.317 incipient cataract, capsular                                  |                     | 9                  | 0.7%  | 4                | 1.0%  |
| 100.321 incomplete cataract, anterior cortex                          |                     | 1                  | 0.1%  | 2                | 0.5%  |
| 100.322 incomplete cataract, posterior cortex                         |                     | 2                  | 0.1%  | 1                | 0.3%  |
| 100.325 incomplete cataract, posterior sutures                        |                     | 4                  | 0.3%  | 0                |       |
| 100.326 incomplete cataract, nucleus                                  |                     | 0                  |       | 1                | 0.3%  |
| 100.328 y-suture tip opacities  |                     | 11                 | 0.8%  | 7                | 1.8%  |
| 100.330 generalized/complete cataract                                 |                     | 30                 | 2.2%  | 0                |       |
| 100.345 <i>significant cataracts (summary)</i>                        |                     | 227                | 16.5% | 40               | 10.2% |
| <b>VITREOUS</b>   |                     |                    |       |                  |       |
| 110.120 persistent hyaloid artery/remnant                             |                     | 2                  | 0.1%  | 0                |       |
| 110.200 vitreous degeneration-anterior chamber                        |                     | 0                  |       | 1                | 0.3%  |

| <b>VITREOUS CONTINUED</b>                                 | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 110.320 vitreal degeneration                              | 11 0.8%          | 1 0.3%           |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 45 3.3%          | 11 2.8%          |
| 120.180 retinal dysplasia, geographic                     | 3 0.2%           | 0                |
| 120.190 retinal dysplasia, detached                       | 1 0.1%           | 0                |
| 120.310 generalized progressive retinal atrophy (PRA)     | 14 1.0%          | 2 0.5%           |
| 120.910 retinal detachment without dialysis               | 1 0.1%           | 0                |
| 120.920 retinal detachment with dialysis                  | 2 0.1%           | 0                |
| 120.960 retinopathy                                       | 1 0.1%           | 0                |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.150 optic disc coloboma                               | 2 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 33 2.4%          | 0                |
| 900.100 other, not inherited                              | 17 1.2%          | 1 0.3%           |
| 900.110 other. suspect not inherited/significance unknown | 31 2.2%          | 17 4.3%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,001 72.6%      | 274 69.9%        |

# WHIPPET

|    | DISORDER   | INHERITANCE         | REFERENCE | BREEDING ADVICE | GENETIC TESTS AVAILABLE           |
|----|--|---------------------|-----------|-----------------|-----------------------------------|
| A. | Persistent pupillary membranes<br>- iris to iris   | Not defined         | 1         | Breeder option  |                                   |
| B. | Cataract   | Not defined         | 1         | NO              |                                   |
| C. | Vitreous degeneration  | Not defined         | 1, 2      | Breeder option  |                                   |
| D. | Choroidal hypoplasia (Collie Eye Anomaly)<br>- staphyloma/coloboma<br>- retinal detachment<br>- retinal hemorrhage<br>- optic nerve coloboma | Autosomal recessive | 3         | NO              | Mutation in the <i>NHEJ1</i> gene |
| E. | Retinal atrophy – generalized  | Not defined         | 4         | NO              |                                   |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### B. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

### C. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment. This is a significant problem in the Whippet.



- D. Choroidal hypoplasia (Collie Eye Anomaly)
- staphyloma/coloboma
  - retinal detachment
  - retinal hemorrhage
  - optic nerve coloboma

A spectrum of malformations present at birth and ranging from inadequate development of the choroid (choroidal hypoplasia) to defects of the choroid, sclera, and/or optic nerve (coloboma/staphyloma) to complete retinal detachment (with or without hemorrhage). Mildly affected animals will have no detectable vision deficit.

This disorder is collectively referred to as "Collie Eye Anomaly" and has been identified in the longhaired Whippet. The choroidal hypoplasia component is caused by a 7799 base pairs deletion with the gene *NHEJ1*. The mutation is a recessive trait. A DNA test is available and is diagnostic only for the choroidal hypoplasia component of CEA. For colobomas to develop, an additional mutation in a second gene has to be present; that gene is still unknown.

- E. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. Except for X-linked PRA in the Siberian Husky, in all breeds studied to date, PRA is inherited as an autosomal recessive trait.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Krishnan, H., et al. (2020). "Vitreous degeneration and associated ocular abnormalities in the dog." *Vet Ophthalmol* **23**(2): 219-224. PMID: 31464365
3. Parker HG, Kukekova AV, Akey DT, et al. Breed relationships facilitate fine-mapping studies: a 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. *Genome research*. 2007;17:1562-1571. Epub 2007/10/06.
4. Somma A, Moreno J, Sato M, et al. Characterization of a novel form of Progressive Retinal Atrophy in Whippet dogs: a clinical, electroretinographic, and breeding study. *Vet Ophth*. 2016: 1-10.

# OCULAR DISORDERS REPORT WHIPPET

| Diagnostic Name  | TOTAL DOGS EXAMINED  | 1991-2015<br>10,890 |      | 2016-2020<br>2,894 |      |
|------------------|--|---------------------|------|--------------------|------|
|                  |  | #                   | %    | #                  | %    |
| <b>GLOBE</b>     |  |                     |      |                    |      |
| 0.110            | microphthalmia   | 1                   | 0.0% | 0                  |      |
| <b>EYELIDS</b>   |  |                     |      |                    |      |
| 20.140           | ectopic cilia  | 2                   | 0.0% | 0                  |      |
| 22.000           | ectropion, unspecified   | 1                   | 0.0% | 0                  |      |
| 25.110           | distichiasis   | 7                   | 0.1% | 6                  | 0.2% |
| <b>NICTITANS</b> |  |                     |      |                    |      |
| 50.210           | pannus of third eyelid   | 1                   | 0.0% | 0                  |      |
| 52.110           | prolapsed gland of the third eyelid                            | 1                   | 0.0% | 0                  |      |
| <b>CORNEA</b>    |  |                     |      |                    |      |
| 70.210           | corneal pannus   | 5                   | 0.0% | 0                  |      |
| 70.700           | corneal dystrophy  | 37                  | 0.3% | 10                 | 0.3% |
| 70.730           | corneal endothelial degeneration                               | 5                   | 0.0% | 1                  | 0.0% |
| <b>UVEA</b>      |  |                     |      |                    |      |
| 93.110           | iris hypoplasia  | 0                   |      | 2                  | 0.1% |
| 93.140           | corneal endothelial pigment without PPM                        | 1                   | 0.0% | 0                  |      |
| 93.180           | iris sphincter dysplasia                                       | 2                   | 0.0% | 0                  |      |
| 93.710           | persistent pupillary membranes, iris to iris                   | 100                 | 0.9% | 44                 | 1.5% |
| 93.720           | persistent pupillary membranes, iris to lens                   | 10                  | 0.1% | 0                  |      |
| 93.730           | persistent pupillary membranes, iris to cornea                 | 11                  | 0.1% | 0                  |      |
| 93.740           | persistent pupillary membranes, iris sheets                    | 16                  | 0.1% | 0                  |      |
| 93.750           | persistent pupillary membranes, lens pigment foci/no strands   | 9                   | 0.1% | 4                  | 0.1% |
| 93.760           | persistent pupillary membranes, endothelial opacity/no strands | 4                   | 0.0% | 4                  | 0.1% |
| 93.810           | uveal melanoma   | 0                   |      | 1                  | 0.0% |
| 93.999           | uveal cysts  | 18                  | 0.2% | 6                  | 0.2% |
| <b>LENS</b>      |  |                     |      |                    |      |
| 100.200          | cataract, unspecified  | 11                  | 0.1% | 0                  |      |
| 100.210          | cataract. suspect not inherited/significance unknown           | 410                 | 3.8% | 106                | 3.7% |
| 100.301          | punctate cataract, anterior cortex                             | 44                  | 0.4% | 16                 | 0.6% |
| 100.302          | punctate cataract, posterior cortex                            | 21                  | 0.2% | 4                  | 0.1% |
| 100.303          | punctate cataract, equatorial cortex                           | 18                  | 0.2% | 9                  | 0.3% |
| 100.304          | punctate cataract, anterior sutures                            | 4                   | 0.0% | 2                  | 0.1% |
| 100.305          | punctate cataract, posterior sutures                           | 5                   | 0.0% | 10                 | 0.3% |
| 100.306          | punctate cataract, nucleus                                     | 16                  | 0.1% | 2                  | 0.1% |
| 100.307          | punctate cataract, capsular                                    | 2                   | 0.0% | 2                  | 0.1% |
| 100.311          | incipient cataract, anterior cortex                            | 51                  | 0.5% | 13                 | 0.4% |
| 100.312          | incipient cataract, posterior cortex                           | 36                  | 0.3% | 4                  | 0.1% |
| 100.313          | incipient cataract, equatorial cortex                          | 53                  | 0.5% | 12                 | 0.4% |
| 100.314          | incipient cataract, anterior sutures                           | 1                   | 0.0% | 2                  | 0.1% |
| 100.315          | incipient cataract, posterior sutures                          | 9                   | 0.1% | 0                  |      |
| 100.316          | incipient cataract, nucleus                                    | 13                  | 0.1% | 2                  | 0.1% |
| 100.317          | incipient cataract, capsular                                   | 17                  | 0.2% | 2                  | 0.1% |
| 100.321          | incomplete cataract, anterior cortex                           | 0                   |      | 4                  | 0.1% |
| 100.322          | incomplete cataract, posterior cortex                          | 1                   | 0.0% | 4                  | 0.1% |
| 100.323          | incomplete cataract, equatorial cortex                         | 0                   |      | 3                  | 0.1% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.326 incomplete cataract, nucleus                      | 0                | 1 0.0%           |
| 100.328 y-suture tip opacities                            | 1 0.0%           | 13 0.4%          |
| 100.330 generalized/complete cataract                     | 16 0.1%          | 2 0.1%           |
| 100.345 <i>significant cataracts (summary)</i>            | 319 2.9%         | 107 3.7%         |
| 100.375 <i>subluxation/luxation, unspecified</i>          | 33 0.3%          | 1 0.0%           |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 16 0.1%          | 16 0.6%          |
| 110.135 PHPV/PTVL   | 11 0.1%          | 1 0.0%           |
| 110.200 vitreous degeneration-anterior chamber            | 30 0.3%          | 43 1.5%          |
| 110.320 vitreal degeneration                              | 588 5.4%         | 68 2.3%          |
| <b>FUNDUS</b>   |                  |                  |
| 97.110 choroidal hypoplasia                               | 19 0.2%          | 0                |
| 97.120 coloboma   | 4 0.0%           | 0                |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 31 0.3%          | 9 0.3%           |
| 120.180 retinal dysplasia, geographic                     | 4 0.0%           | 5 0.2%           |
| 120.190 retinal dysplasia, detached                       | 4 0.0%           | 1 0.0%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 40 0.4%          | 8 0.3%           |
| 120.400 retinal hemorrhage                                | 1 0.0%           | 0                |
| 120.910 retinal detachment without dialysis               | 4 0.0%           | 0                |
| 120.920 retinal detachment with dialysis                  | 0                | 1 0.0%           |
| 120.960 retinopathy                                       | 6 0.1%           | 5 0.2%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 3 0.0%           | 1 0.0%           |
| 130.120 optic nerve hypoplasia                            | 3 0.0%           | 0                |
| 130.150 optic disc coloboma                               | 14 0.1%          | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 114 1.0%         | 0                |
| 900.100 other, not inherited                              | 233 2.1%         | 2 0.1%           |
| 900.110 other. suspect not inherited/significance unknown | 117 1.1%         | 124 4.3%         |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 9,622 88.4%      | 2,442 84.4%      |

# WHITE SHEPHERD

|    | <b>DISORDER</b>                           | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING<br/>ADVICE</b> | <b>GENETIC TESTS<br/>AVAILABLE</b> |
|----|---|--------------------|------------------|----------------------------|------------------------------------|
| A. | Corneal dystrophy<br>- epithelial/stromal | Not defined        | 1                | Breeder option             |                                    |

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## Description and Comments

### A. Corneal dystrophy - epithelial/stromal

A non-inflammatory corneal opacity (white to gray) present in one or more of the corneal layers; usually inherited and bilateral. In the Mi-Ki, lesions are circular or semicircular central crystalline deposits in the anterior corneal stroma that appear between 2 and 5 years of age. It may be associated with exophthalmos and lagophthalmos common in these dogs.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the White Shepherd breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT WHITE SHEPHERD

| Diagnostic Name    | TOTAL DOGS EXAMINED                                  | 1991-2015 |       | 2016-2020 |       |
|--------------------|--|-----------|-------|-----------|-------|
|                    |  | #         | %     | #         | %     |
| <b>CORNEA</b>      |  |           |       |           |       |
| 70.210             | corneal pannus                                       | 0         |       | 2         | 4.3%  |
| 70.700             | corneal dystrophy                                    | 2         | 11.8% | 4         | 8.5%  |
| <b>UVEA</b>        |  |           |       |           |       |
| 93.720             | persistent pupillary membranes, iris to lens         | 1         | 5.9%  | 0         |       |
| <b>LENS</b>        |  |           |       |           |       |
| 100.210            | cataract. suspect not inherited/significance unknown | 1         | 5.9%  | 3         | 6.4%  |
| 100.305            | punctate cataract, posterior sutures                 | 0         |       | 1         | 2.1%  |
| 100.317            | incipient cataract, capsular                         | 1         | 5.9%  | 0         |       |
| 100.328            | y-suture tip opacities                               | 0         |       | 1         | 2.1%  |
| 100.345            | <i>significant cataracts (summary)</i>               | 1         | 5.9%  | 2         | 4.3%  |
| <b>RETINA</b>      |  |           |       |           |       |
| 120.170            | retinal dysplasia, folds                             | 1         | 5.9%  | 0         |       |
| <b>OPTIC NERVE</b> |  |           |       |           |       |
| 130.110            | micropapilla   | 1         | 5.9%  | 1         | 2.1%  |
| 130.120            | optic nerve hypoplasia                               | 1         | 5.9%  | 0         |       |
| <b>OTHER</b>       |  |           |       |           |       |
| 900.100            | other, not inherited                                 | 0         |       | 1         | 2.1%  |
| 900.110            | other. suspect not inherited/significance unknown    | 0         |       | 5         | 10.6% |
| <b>NORMAL</b>      |  |           |       |           |       |
| 0.000              | normal globe   | 10        | 58.8% | 32        | 68.1% |

## WIRE FOX TERRIER\*

\*The Wire Fox Terrier and the Smooth Fox Terrier were originally considered two varieties of the same breed. They became separate breeds in 1985. It is likely that the same genetic diseases exist in both breeds.

|    | <b>DISORDER</b>                                  | <b>INHERITANCE</b>     | <b>REFERENCE</b> | <b>BREEDING<br/>ADVICE</b> | <b>GENETIC TESTS<br/>AVAILABLE</b>      |
|----|--|------------------------|------------------|----------------------------|---|
| A. | Glaucoma   | Not defined            | 1, 2             | NO                         |   |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined            | 1                | Breeder option             |   |
| C. | Cataract   | Not defined            | 1                | NO                         |   |
| D. | Lens luxation                                    | Autosomal<br>recessive | 3                | NO                         | Mutation in the<br><i>ADAMTS17</i> gene |

### Description and Comments

#### A. Glaucoma

Glaucoma is characterized by an elevation of intraocular pressure (IOP) which, when sustained, causes intraocular damage resulting in blindness. The elevated IOP occurs because the fluid cannot leave through the iridocorneal angle. Diagnosis and classification of glaucoma requires measurement of the intraocular pressure (tonometry) and examination of the iridocorneal angle (gonioscopy). Neither of these tests are part of a routine breed eye screening exam.

#### B. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

#### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region. The cataracts observed in Wire Fox Terrier begin in the posterior subcapsular region and are progressive.

D. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

## References

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.
2. Martin CL, Wyman M. Primary glaucoma in the dog. *Vet Clin North Am.* 1978;8:257-286.
3. Gould D, Pettitt L, McLaughlin B, et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Vet Ophthalmol.* 2011;14:378-384.

# OCULAR DISORDERS REPORT

## WIRE FOX TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>GLOBE</b>  |                     |           |       |           |       |
| 0.110 microphthalmia  |                     | 1         | 0.3%  | 0         |       |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 25.110 distichiasis   |                     | 8         | 2.7%  | 1         | 2.4%  |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.700 corneal dystrophy  |                     | 3         | 1.0%  | 0         |       |
| 70.730 corneal endothelial degeneration                             |                     | 1         | 0.3%  | 0         |       |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 92        | 31.0% | 26        | 63.4% |
| 93.720 persistent pupillary membranes, iris to lens                 |                     | 3         | 1.0%  | 2         | 4.9%  |
| 93.730 persistent pupillary membranes, iris to cornea               |                     | 5         | 1.7%  | 0         |       |
| 93.740 persistent pupillary membranes, iris sheets                  |                     | 1         | 0.3%  | 0         |       |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 0         |       | 1         | 2.4%  |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.200 cataract, unspecified                                       |                     | 4         | 1.3%  | 0         |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 2         | 0.7%  | 0         |       |
| 100.301 punctate cataract, anterior cortex                          |                     | 3         | 1.0%  | 0         |       |
| 100.311 incipient cataract, anterior cortex                         |                     | 5         | 1.7%  | 0         |       |
| 100.312 incipient cataract, posterior cortex                        |                     | 5         | 1.7%  | 0         |       |
| 100.313 incipient cataract, equatorial cortex                       |                     | 2         | 0.7%  | 0         |       |
| 100.314 incipient cataract, anterior sutures                        |                     | 1         | 0.3%  | 0         |       |
| 100.321 incomplete cataract, anterior cortex                        |                     | 1         | 0.3%  | 0         |       |
| 100.322 incomplete cataract, posterior cortex                       |                     | 1         | 0.3%  | 0         |       |
| 100.326 incomplete cataract, nucleus                                |                     | 1         | 0.3%  | 0         |       |
| 100.330 generalized/complete cataract                               |                     | 8         | 2.7%  | 0         |       |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 31        | 10.4% | 0         |       |
| <b>VITREOUS</b>   |                     |           |       |           |       |
| 110.120 persistent hyaloid artery/remnant                           |                     | 1         | 0.3%  | 0         |       |
| 110.320 vitreal degeneration  |                     | 1         | 0.3%  | 0         |       |
| <b>RETINA</b>   |                     |           |       |           |       |
| 120.170 retinal dysplasia, folds                                    |                     | 1         | 0.3%  | 0         |       |
| 120.310 generalized progressive retinal atrophy (PRA)               |                     | 4         | 1.3%  | 0         |       |
| <b>OTHER</b>  |                     |           |       |           |       |
| 900.000 other, unspecified  |                     | 3         | 1.0%  | 0         |       |
| 900.100 other, not inherited  |                     | 12        | 4.0%  | 0         |       |
| 900.110 other. suspect not inherited/significance unknown           |                     | 1         | 0.3%  | 0         |       |
| <b>NORMAL</b>   |                     |           |       |           |       |
| 0.000 normal globe  |                     | 179       | 60.3% | 14        | 34.1% |



# WIREHAired POINTING GRIFFON

|    | DISORDER   | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|-------------|-----------|-----------------|
| A. | Distichiasis                                     | Not defined | 1         | Breeder option  |
| B. | Persistent pupillary membranes<br>- iris to iris | Not defined | 1         | Breeder option  |
| C. | Cataract   | Not defined | 1         | NO              |
| D. | Y-suture tip opacity                             | Not defined | 1         | Breeder option  |

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## Description and Comments

### A. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regard to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded; breeding discretion is advised.

### B. Persistent pupillary membranes (PPMs)

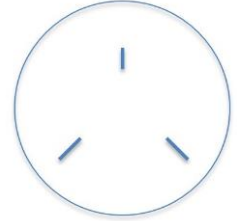
Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

### C. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

#### D. Y-suture tip opacity

These are prominent (or “highlighted” or “more dense”) distal portions of the posterior sutures that may occur in the posterior cortex to occasionally on the posterior lens capsule. This is not a true cataract, so there is no lens fiber disruption (no feathering or bulbous tips). It may be in the shape of a “peace sign” as diagrammed here, but occasionally a patient may have 4-5 suture lines and therefore more suture tip opacities. They may be present only at one suture tip of one eye or up to all three (or more, as stated above) suture tips in both eyes in a given dog. They are more commonly found in multiples or at least bilaterally symmetrical. They may be visible only with biomicroscopy or sometimes with retroillumination. They do not appear to progress (unless mis-diagnosed) and are considered essentially a variation of normal or possibly familial, as they are seen more commonly in certain breeds.



These should be marked under the “Lens” section of the CAER form. The newest version of the form (3/16/21) has boxes that say, “posterior Y-suture tip opacities” which should be marked. If working with an older version of the form, there are 2 places to mark within the lens section as cataract bubbles: “punctate posterior sutures” AND ALSO MARK “suspect not inherited/significance unknown” (without which they technically fail or at least require further information before coding). This diagnosis should ALSO be accompanied by drawings (like below) and/or have comments such as: “E2” or “posterior suture tip opacities.” This helps differentiate them from 1) prominent but otherwise normal full suture lines – which should just be commented on and are treated as normal, and 2) true sutural cataracts - which would either be breeder option or failing.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Wirehaired Pointing Griffon breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT WIREHAired POINTING GRIFFON

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |      | 2016-2020 |      |
|---|---------------------|-----------|------|-----------|------|
|   |                     | #         | %    | #         | %    |
| <b>GLOBE</b>  |                     |           |      |           |      |
| 0.110 microphthalmia  |                     | 1         | 0.2% | 0         |      |
| <b>EYELIDS</b>  |                     |           |      |           |      |
| 21.000 entropion, unspecified                                       |                     | 3         | 0.6% | 1         | 0.3% |
| 25.110 distichiasis   |                     | 3         | 0.6% | 6         | 1.7% |
| <b>NICTITANS</b>  |                     |           |      |           |      |
| 51.100 third eyelid cartilage anomaly                               |                     | 0         |      | 1         | 0.3% |
| 52.110 prolapsed gland of the third eyelid                          |                     | 1         | 0.2% | 0         |      |
| <b>CORNEA</b>   |                     |           |      |           |      |
| 70.210 corneal pannus   |                     | 0         |      | 1         | 0.3% |
| 70.700 corneal dystrophy  |                     | 1         | 0.2% | 0         |      |
| 70.730 corneal endothelial degeneration                             |                     | 3         | 0.6% | 0         |      |
| <b>UVEA</b>   |                     |           |      |           |      |
| 93.110 iris hypoplasia  |                     | 0         |      | 2         | 0.6% |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 7         | 1.5% | 7         | 1.9% |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 1         | 0.2% | 2         | 0.6% |
| <b>LENS</b>   |                     |           |      |           |      |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 34        | 7.2% | 29        | 8.1% |
| 100.301 punctate cataract, anterior cortex                          |                     | 0         |      | 1         | 0.3% |
| 100.302 punctate cataract, posterior cortex                         |                     | 1         | 0.2% | 2         | 0.6% |
| 100.303 punctate cataract, equatorial cortex                        |                     | 0         |      | 1         | 0.3% |
| 100.305 punctate cataract, posterior sutures                        |                     | 0         |      | 1         | 0.3% |
| 100.306 punctate cataract, nucleus                                  |                     | 2         | 0.4% | 1         | 0.3% |
| 100.307 punctate cataract, capsular                                 |                     | 0         |      | 1         | 0.3% |
| 100.311 incipient cataract, anterior cortex                         |                     | 2         | 0.4% | 1         | 0.3% |
| 100.313 incipient cataract, equatorial cortex                       |                     | 1         | 0.2% | 0         |      |
| 100.316 incipient cataract, nucleus                                 |                     | 2         | 0.4% | 1         | 0.3% |
| 100.328 y-suture tip opacities                                      |                     | 1         | 0.2% | 2         | 0.6% |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 9         | 1.9% | 11        | 3.1% |
| <b>VITREOUS</b>   |                     |           |      |           |      |
| 110.120 persistent hyaloid artery/remnant                           |                     | 0         |      | 2         | 0.6% |
| 110.200 vitreous degeneration-anterior chamber                      |                     | 0         |      | 1         | 0.3% |
| 110.320 vitreal degeneration  |                     | 7         | 1.5% | 1         | 0.3% |
| <b>RETINA</b>   |                     |           |      |           |      |
| 120.170 retinal dysplasia, folds                                    |                     | 4         | 0.8% | 1         | 0.3% |
| 120.180 retinal dysplasia, geographic                               |                     | 1         | 0.2% | 0         |      |
| 120.400 retinal hemorrhage  |                     | 1         | 0.2% | 0         |      |
| 120.960 retinopathy   |                     | 0         |      | 1         | 0.3% |
| <b>OTHER</b>  |                     |           |      |           |      |
| 900.000 other, unspecified  |                     | 6         | 1.3% | 0         |      |
| 900.100 other, not inherited  |                     | 2         | 0.4% | 1         | 0.3% |
| 900.110 other. suspect not inherited/significance unknown           |                     | 8         | 1.7% | 12        | 3.3% |

|                                     | 1991-2015 | 2016-2020 |
|-------------------------------------|-----------|-----------|
| <b>NORMAL</b><br>0.000 normal globe | 411 87.1% | 295 81.9% |

# WIREHAISED VIZSLA

| DISORDER                          | INHERITANCE | REFERENCE | BREEDING ADVICE         |
|-----------------------------------|-------------|-----------|-------------------------|
| A. Persistent pupillary membranes |             |           |                         |
| - iris to iris                    | Not defined | 1         | Breeder option          |
| - lens pigment foci/no strands    | Not defined | 1         | Passes with no notation |

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## Description and Comments

### A. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

## References

There are no references providing detailed descriptions of hereditary conditions of the Wirehaired Vizsla breed. The conditions listed above are generally recognized to exist in this breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT WIREHAIED VIZSLA

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015 |       | 2016-2020 |       |
|---|---------------------|-----------|-------|-----------|-------|
|   |                     | #         | %     | #         | %     |
| <b>EYELIDS</b>  |                     |           |       |           |       |
| 25.110 distichiasis   |                     | 0         |       | 1         | 1.0%  |
| <b>NICTITANS</b>  |                     |           |       |           |       |
| 52.110 prolapsed gland of the third eyelid                          |                     | 3         | 2.5%  | 0         |       |
| <b>CORNEA</b>   |                     |           |       |           |       |
| 70.700 corneal dystrophy  |                     | 0         |       | 1         | 1.0%  |
| <b>UVEA</b>   |                     |           |       |           |       |
| 93.710 persistent pupillary membranes, iris to iris                 |                     | 10        | 8.4%  | 4         | 3.8%  |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands |                     | 11        | 9.2%  | 5         | 4.8%  |
| <b>LENS</b>   |                     |           |       |           |       |
| 100.210 cataract. suspect not inherited/significance unknown        |                     | 18        | 15.1% | 7         | 6.7%  |
| 100.312 incipient cataract, posterior cortex                        |                     | 0         |       | 1         | 1.0%  |
| 100.322 incomplete cataract, posterior cortex                       |                     | 0         |       | 1         | 1.0%  |
| 100.345 <i>significant cataracts (summary)</i>                      |                     | 0         |       | 2         | 1.9%  |
| <b>VITREOUS</b>   |                     |           |       |           |       |
| 110.320 vitreal degeneration  |                     | 2         | 1.7%  | 0         |       |
| <b>RETINA</b>   |                     |           |       |           |       |
| 120.910 retinal detachment without dialysis                         |                     | 1         | 0.8%  | 0         |       |
| <b>OTHER</b>  |                     |           |       |           |       |
| 900.000 other, unspecified  |                     | 4         | 3.4%  | 0         |       |
| 900.100 other, not inherited  |                     | 0         |       | 1         | 1.0%  |
| 900.110 other. suspect not inherited/significance unknown           |                     | 3         | 2.5%  | 6         | 5.7%  |
| <b>NORMAL</b>   |                     |           |       |           |       |
| 0.000 normal globe  |                     | 90        | 75.6% | 82        | 78.1% |

# XOLOITZCUINTLI

|    | <b>DISORDER</b> | <b>INHERITANCE</b> | <b>REFERENCE</b> | <b>BREEDING ADVICE</b> |
|----|-----------------|--------------------|------------------|------------------------|
| A. | Cataract        | Not defined        | 1                | NO                     |

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## Description and Comments

### A. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

## References

There are no references providing detailed descriptions of hereditary ocular conditions of the Xoloitzcuintli breed. The conditions listed above are generally recognized to exist in the breed, as evidenced by identification on breed eye screening examinations and/or clinical experience of veterinary ophthalmologists.

1. ACVO Genetics Committee and/or Data from OFA All-Breeds Report.

# OCULAR DISORDERS REPORT XOLOITZCUINTLI

| Diagnostic Name | TOTAL DOGS EXAMINED  | 1991-2015 |       | 2016-2020 |       |
|-----------------|--|-----------|-------|-----------|-------|
|                 |  | #         | %     | #         | %     |
| <b>EYELIDS</b>  |  |           |       |           |       |
| 25.110          | distichiasis   | 1         | 1.9%  | 0         |       |
| <b>UVEA</b>     |  |           |       |           |       |
| 93.710          | persistent pupillary membranes, iris to iris                 | 1         | 1.9%  | 2         | 2.0%  |
| 93.750          | persistent pupillary membranes, lens pigment foci/no strands | 0         |       | 3         | 2.9%  |
| <b>LENS</b>     |  |           |       |           |       |
| 100.210         | cataract. suspect not inherited/significance unknown         | 0         |       | 3         | 2.9%  |
| 100.305         | punctate cataract, posterior sutures                         | 0         |       | 1         | 1.0%  |
| 100.311         | incipient cataract, anterior cortex                          | 2         | 3.8%  | 1         | 1.0%  |
| 100.312         | incipient cataract, posterior cortex                         | 5         | 9.4%  | 3         | 2.9%  |
| 100.313         | incipient cataract, equatorial cortex                        | 2         | 3.8%  | 1         | 1.0%  |
| 100.317         | incipient cataract, capsular                                 | 2         | 3.8%  | 1         | 1.0%  |
| 100.345         | <i>significant cataracts (summary)</i>                       | 11        | 20.8% | 7         | 6.9%  |
| <b>VITREOUS</b> |  |           |       |           |       |
| 110.320         | vitreal degeneration   | 0         |       | 1         | 1.0%  |
| <b>RETINA</b>   |  |           |       |           |       |
| 120.180         | retinal dysplasia, geographic                                | 1         | 1.9%  | 0         |       |
| <b>OTHER</b>    |  |           |       |           |       |
| 900.100         | other, not inherited   | 0         |       | 1         | 1.0%  |
| 900.110         | other. suspect not inherited/significance unknown            | 0         |       | 4         | 3.9%  |
| <b>NORMAL</b>   |  |           |       |           |       |
| 0.000           | normal globe   | 46        | 86.8% | 86        | 84.3% |



# YORKSHIRE TERRIER

|    | DISORDER                       | INHERITANCE         | REFERENCE | BREEDING ADVICE         | GENETIC TESTS AVAILABLE              |
|----|--------------------------------|---------------------|-----------|-------------------------|--------------------------------------|
| A. | Keratoconjunctivitis sicca     | Not defined         | 1, 2      | NO                      |                                      |
| B. | Distichiasis                   | Not defined         | 1         | Breeder option          |                                      |
| C. | Persistent pupillary membranes | Not defined         | 1         | Breeder option          |                                      |
|    | - iris to iris                 | Not defined         | 3         | Passes with no notation |                                      |
|    | - lens pigment foci/no strands | Not defined         |           |                         |                                      |
| D. | Cataract                       | Not defined         | 1         | NO                      |                                      |
| E. | Lens luxation                  | Autosomal recessive | 3-5       | NO                      | Mutation in the <i>ADAMTS17</i> gene |
| F. | Vitreous degeneration          | Not defined         | 1         | Breeder option          |                                      |
| G. | Retinal atrophy - generalized  | Not defined         | 1         | NO                      |                                      |

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## Description and Comment

### A. Keratoconjunctivitis sicca

An abnormality of the tear film, most commonly a deficiency of the aqueous portion, although the mucin and/or lipid layers may be affected; results in ocular irritation and/or vision impairment. There is evidence that Yorkshire Terriers sometimes present with severe, congenital, unilateral keratoconjunctivitis sicca (KCS) and it is suspected this is due to hypoplasia or aplasia of the gland.

### B. Distichiasis

Eyelashes abnormally located on the eyelid margin which may cause ocular irritation. Distichiasis may occur at any time in the life of a dog. It is difficult to make a strong recommendation with regards to breeding dogs with this entity. The hereditary basis has not been established although it seems probable due to the high incidence in some breeds. Reducing the incidence is a logical goal. When diagnosed, distichiasis should be recorded. Breeding discretion is advised.

C. Persistent pupillary membranes (PPMs)

Persistent blood vessel remnants in the anterior chamber of the eye which fail to regress normally by 3 months of age. These strands may bridge from iris to iris, iris to cornea, iris to lens, or form sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and when severe, vision impairment or blindness may occur.

Lens pigment foci/no strands is considered an insignificant finding and therefore not noted on the certificate.

D. Cataract

A partial or complete opacity of the lens and/or its capsule. In cases where cataracts are complete and affect both eyes, blindness results. The prudent approach is to assume cataracts to be hereditary except in cases known to be associated with trauma, other causes of ocular inflammation, specific metabolic diseases, persistent pupillary membrane, persistent hyaloid, or nutritional deficiencies. Cataracts may involve the lens completely (diffuse) or in a localized region.

E. Lens luxation

Partial (subluxation) or complete displacement of the lens from the normal anatomic site behind the pupil. Lens luxation not associated with trauma or inflammation is presumed to be inherited. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. A mutation in *ADAMTS17* has been associated with primary lens luxation. A DNA test is available.

F. Vitreous degeneration

A liquefaction of the vitreous gel which may predispose to retinal detachment.

G. Retinal atrophy - generalized

A degenerative disease of the retinal visual cells which progresses to blindness. This abnormality, also known as progressive retinal atrophy or PRA, may be detected by electroretinogram (not part of a routine eye screening examination) before it is apparent clinically. PRA is inherited as an autosomal recessive trait in most breeds.

## References

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4. Farias FH, Johnson GS, Taylor JF, et al. An ADAMTS17 splice donor site mutation in dogs with primary lens luxation. *Invest Ophthalmol Vis Sci.* 2010;51:4716-4721.
5. Walde I. Retinal and corneal dysplasias in the Yorkshire Terrier and other breeds in Austria. *Tierärztliche Praxis.* 1997;25:62.

# OCULAR DISORDERS REPORT YORKSHIRE TERRIER

| Diagnostic Name   | TOTAL DOGS EXAMINED | 1991-2015<br>1,573 |      | 2016-2020<br>641 |      |
|---|---------------------|--------------------|------|------------------|------|
|   |                     | #                  | %    | #                | %    |
| <b>GLOBE</b>  |                     |                    |      |                  |      |
| 0.110 microphthalmia  |                     | 3                  | 0.2% | 3                | 0.5% |
| 10.000 glaucoma   |                     | 1                  | 0.1% | 0                |      |
| <b>EYELIDS</b>  |                     |                    |      |                  |      |
| 21.000 entropion, unspecified   |                     | 0                  |      | 1                | 0.2% |
| 25.110 distichiasis   |                     | 33                 | 2.1% | 6                | 0.9% |
| <b>NASOLACRIMAL</b>   |                     |                    |      |                  |      |
| 40.910 keratoconjunctivitis sicca                                     |                     | 5                  | 0.3% | 2                | 0.3% |
| <b>NICTITANS</b>  |                     |                    |      |                  |      |
| 52.110 prolapsed gland of the third eyelid                            |                     | 1                  | 0.1% | 0                |      |
| <b>CORNEA</b>   |                     |                    |      |                  |      |
| 70.210 corneal pannus   |                     | 4                  | 0.3% | 0                |      |
| 70.220 pigmentary keratitis   |                     | 4                  | 0.3% | 0                |      |
| 70.700 corneal dystrophy  |                     | 12                 | 0.8% | 4                | 0.6% |
| 70.730 corneal endothelial degeneration                               |                     | 1                  | 0.1% | 0                |      |
| <b>UVEA</b>   |                     |                    |      |                  |      |
| 93.110 iris hypoplasia  |                     | 1                  | 0.1% | 0                |      |
| 93.710 persistent pupillary membranes, iris to iris                   |                     | 154                | 9.8% | 49               | 7.6% |
| 93.720 persistent pupillary membranes, iris to lens                   |                     | 4                  | 0.3% | 0                |      |
| 93.730 persistent pupillary membranes, iris to cornea                 |                     | 4                  | 0.3% | 2                | 0.3% |
| 93.750 persistent pupillary membranes, lens pigment foci/no strands   |                     | 12                 | 0.8% | 14               | 2.2% |
| 93.760 persistent pupillary membranes, endothelial opacity/no strands |                     | 2                  | 0.1% | 0                |      |
| <b>LENS</b>   |                     |                    |      |                  |      |
| 100.200 cataract, unspecified   |                     | 23                 | 1.5% | 0                |      |
| 100.210 cataract. suspect not inherited/significance unknown          |                     | 51                 | 3.2% | 10               | 1.6% |
| 100.301 punctate cataract, anterior cortex                            |                     | 27                 | 1.7% | 9                | 1.4% |
| 100.302 punctate cataract, posterior cortex                           |                     | 11                 | 0.7% | 2                | 0.3% |
| 100.303 punctate cataract, equatorial cortex                          |                     | 6                  | 0.4% | 0                |      |
| 100.304 punctate cataract, anterior sutures                           |                     | 3                  | 0.2% | 0                |      |
| 100.305 punctate cataract, posterior sutures                          |                     | 4                  | 0.3% | 2                | 0.3% |
| 100.306 punctate cataract, nucleus                                    |                     | 1                  | 0.1% | 2                | 0.3% |
| 100.307 punctate cataract, capsular                                   |                     | 0                  |      | 3                | 0.5% |
| 100.311 incipient cataract, anterior cortex                           |                     | 24                 | 1.5% | 12               | 1.9% |
| 100.312 incipient cataract, posterior cortex                          |                     | 16                 | 1.0% | 7                | 1.1% |
| 100.313 incipient cataract, equatorial cortex                         |                     | 15                 | 1.0% | 4                | 0.6% |
| 100.314 incipient cataract, anterior sutures                          |                     | 2                  | 0.1% | 1                | 0.2% |
| 100.315 incipient cataract, posterior sutures                         |                     | 3                  | 0.2% | 1                | 0.2% |
| 100.316 incipient cataract, nucleus                                   |                     | 3                  | 0.2% | 0                |      |
| 100.317 incipient cataract, capsular                                  |                     | 1                  | 0.1% | 1                | 0.2% |
| 100.321 incomplete cataract, anterior cortex                          |                     | 2                  | 0.1% | 5                | 0.8% |
| 100.322 incomplete cataract, posterior cortex                         |                     | 0                  |      | 3                | 0.5% |
| 100.323 incomplete cataract, equatorial cortex                        |                     | 0                  |      | 1                | 0.2% |
| 100.326 incomplete cataract, nucleus                                  |                     | 1                  | 0.1% | 1                | 0.2% |
| 100.328 y-suture tip opacities  |                     | 1                  | 0.1% | 1                | 0.2% |

| <b>LENS CONTINUED</b>                                     | <b>1991-2015</b> | <b>2016-2020</b> |
|---|------------------|------------------|
| 100.330 generalized/complete cataract                     | 28 1.8%          | 1 0.2%           |
| 100.345 significant cataracts (summary)                   | 171 10.9%        | 56 8.7%          |
| 100.375 subluxation/luxation, unspecified                 | 1 0.1%           | 0                |
| <b>VITREOUS</b>   |                  |                  |
| 110.120 persistent hyaloid artery/remnant                 | 2 0.1%           | 3 0.5%           |
| 110.135 PHPV/PTVL   | 4 0.3%           | 1 0.2%           |
| 110.200 vitreous degeneration-anterior chamber            | 1 0.1%           | 5 0.8%           |
| 110.320 vitreal degeneration                              | 19 1.2%          | 7 1.1%           |
| <b>RETINA</b>   |                  |                  |
| 120.170 retinal dysplasia, folds                          | 7 0.4%           | 1 0.2%           |
| 120.310 generalized progressive retinal atrophy (PRA)     | 53 3.4%          | 4 0.6%           |
| 120.920 retinal detachment with dialysis                  | 0                | 2 0.3%           |
| 120.960 retinopathy                                       | 4 0.3%           | 1 0.2%           |
| <b>OPTIC NERVE</b>  |                  |                  |
| 130.110 micropapilla                                      | 0                | 1 0.2%           |
| 130.120 optic nerve hypoplasia                            | 3 0.2%           | 0                |
| 130.150 optic disc coloboma                               | 1 0.1%           | 0                |
| <b>OTHER</b>  |                  |                  |
| 900.000 other, unspecified                                | 19 1.2%          | 0                |
| 900.100 other, not inherited                              | 26 1.7%          | 2 0.3%           |
| 900.110 other. suspect not inherited/significance unknown | 22 1.4%          | 24 3.7%          |
| <b>NORMAL</b>   |                  |                  |
| 0.000 normal globe  | 1,195 76.0%      | 476 74.3%        |