

OCULAR DISORDERS

presumed to be inherited in purebred dogs

CARDIGAN WELSH CORGI

SEVENTH EDITION

2014

GENETICS COMMITTEE OF THE
AMERICAN COLLEGE OF VETERINARY OPHTHALMOLOGISTS

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 20th century during the early days of this College's inception, have continued into the 21st 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 most current edition of this document has been prepared in PDF format. The content of this production has originated from several sources. 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 American College of Veterinary Ophthalmologists for Companion Animal Eye Registry (OFA / CAER)) and Canine Eye Registration Foundation (CERF) examinations. The research copies are then conscientiously submitted to the registry 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 data base grows.

This production builds on the basis provided by the diligent efforts of all previous Genetics Committees. Out of collegial respect and for an historical perspective I would like to acknowledge the previous Chairpersons of the ACVO Genetics Committee recognizing that with every chair, a multitude of dedicated committee members were responsible for the accomplishments and contributions of each committee. Dr. David Covitz 1986-1988, Dr. Randall Scagliotti 1988-1992, Dr. Cynthia Cook 1992-1995, Dr. Keith Collins 1995-1997, the late Dr. Cindy Wheeler 1997-1999, Dr. Nancy Bromberg 1999-2003.

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

Melanie Morgan Williams
DVM, Diplomate ACVO,
Chair, ACVO Genetics Committee 2003-2006

7th Edition 2014 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 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) and all Genetics Committee members.

The Genetics Committee members (2014-2015): Dr. Ellen Belknap, Dr. Caroline Betbeze, Dr. Shannon Boveland, Dr. Janet Isherwood, Dr. Ruth Marrion, Dr. Jessica Meekins, Dr. Kenneth Pierce, Dr. Lynn Sandmeyer, Dr. Wendy Townsend, Dr. Kristina Vygantas, and OFA liaison Dr. Katie Diehl.

Eddie Dziuk, OFA Chief Operating Officer.

Fifth Edition 2010 Version Content:

Scientific literature and statistics to December 31st, 2008.
Acceptance of literature and statistics @ Genetics Committee meeting November 2009.
Compilation and proofing of content of 2010 Version, November 2009 – November 2010.
Publication, November 2010.

Sixth Edition 2013 Version Content:

Scientific literature and statistics to January 1 2014
Acceptance of literature and statistics at Genetics Committee meeting November 2013.
Publication October 2013

Seventh Edition 2014 Version Content:

Scientific literature and statistics to January 1 2015
Acceptance of literature and statistics at Genetics Committee meeting October 2014.
Publication Summer 2015

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.

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 greatly assist 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

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) in 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 ten 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.

*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.

1. **Glaucoma** – See above *note.
2. **Keratocconjunctivitis 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 above *note.
3. **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 above *note.
4. **Lens luxation or subluxation** – See above *note.
5. **Persistent hyperplastic primary vitreous/persistent hyperplastic tunica vasculosa lentis** – See above *note.
6. **Retinal detachment** – See above *note.
7. **Retinal atrophy – generalized (PRA)** - Breeding is not advised for any animal demonstrating bilaterally symmetric retinal degeneration (considered to be PRA unless proven otherwise).
8. **Retinal dysplasia, geographic or detached forms** – See above *note
9. **Optic nerve coloboma**
10. **Optic nerve hypoplasia**

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

Mastiff – persistent pupillary membrane

Basenji – persistent pupillary membrane

Pembroke Welsh Corgi – persistent pupillary membrane

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

- 1) Entropion
- 2) Ectropion
- 3) Macropharon

4) 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 untreated, can lead to loss of vision. A responsible breeding program should recognize and select away from these exaggerated facial features.

The information contained in this book is protected by copyright. Duplication of any part is not allowed without express written permission from the American College of Veterinary Ophthalmologists and/or the OFA.

Glossary of Terms

(For more detailed definition the reader is referred to medical and genetic scientific texts)

Achromatopsia: see **Day blindness**

Canine multifocal retinopathy: characterized by numerous distinct (i.e. multi-focal), 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: noninflammatory, 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 and 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 inferiotemporal 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: 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.

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 **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 disease 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 retinal 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 retains maturity.

Retinal dysplasia – geographic: 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 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 *rcd4* 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.

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 | Grand Basset Griffon Vendeen |
| Alaskan Noble Companion Dog | Hovawart |
| American Blue Lacy | Kai Ken |
| American English Coonhound | Kooikerhondje |
| American Foxhound | Kyi Leo |
| Anatolian Shepherd | Lamalese |
| Azawakh | Large Munsterlander |
| Barbet | Manchester Terrier |
| Basset Fauve de Bretagne | Mudi |
| Beauceron | Otterhound |
| Bergamasco | Perro de Presa Canario |
| Biewer | Peruvian Inca Orchid |
| Bluetick Coonhound | Plott |
| Bolonka Zwetna | Portuguese Pointer |
| Braque du Bourbonnais | Pudelpointer |
| Braque Francais | Pumi |
| Canadian Eskimo Dog | Redbone Coonhound |
| Cane Corso | Scottish Deerhound |
| Caucasian Ovcharka | Silken Windhound |
| Chart Polski | Shikoku |
| Cirneco Dell'Etna | Skye Terrier |
| Drever | Small Munsterlander |
| Deutscher Wachtelhund | Swedish Lapphund |
| Dutch Shepherd | Treeing Walker Coonhound |
| English Foxhound | Tibetan Mastiff |
| Fila Brasileiro | White Shepherd |
| French Spaniel | Xoloitzcuintli |
| German Longhaired Pointer | |

Genetic Testing For Canine Ocular Disorders

A. Contact Information For Genetic Testing Laboratories (as of October 8, 2014)

OptiGen, LLC

Cornell Business & Technology Park
767 Warren Road, Suite 300
Ithaca, NY 14850
Tel: 607-257-0301
Fax: 607-257-0353
E-mail: genetest@optigen.com
www.optigen.com

Animal Health Trust

Lanwades Park
Kentford
Newmarket
Suffolk
United Kingdom
CB8 7UU
Tel: 01638 55 56 21 (UK)
Fax: 01638 55 56 66 (UK)
Email: dnatesting@aht.org.uk
www.aht.org.uk

Animal Genetics Inc.

1336 Timberlane Road
Tallahassee, FL 32312-1766
Tel: 1-866-922-6436
Email: contact@animalgenetics.us
www.animalgenetics.us/

Animal Genetic Testing and Research Lab

Department of Veterinary Science 108
Gluck Equine Research Center
University of Kentucky
Lexington, KY 40546-0099
Tel: 859-257-4757 Ext: 81212
Fax: 859-2575169

Dogenes

161 Sherin Ave
Peterborough, ON K9J 7V5
Canada
Tel: 705-748-0089

GenSol Diagnostics

PO Box 701492

Saint Cloud, FL 34770
Tel: (321) 482-8347
Email: info@gensoldx.com
www.gensoldx.com

Goldstein Molecular and Genetics Laboratory

Cornell University
Richard E. Goldstein
Tel: 607-253-4490
Fax: 607-253-3534
Email: phpt@cornell.edu

HealthGene

2175 Keele Street
Toronto, ON M6M 3Z4
Canada
Tel: 1-877-371-1551 (toll free)
Email: info@healthgene.com

MSU - Laboratory of Comparative Medical Genetics

Michigan State University
2209 Biomedical Physical Sciences
East Lansing, MI, 48824
Tel: 517-355-6463 Ext: 1552
Email: fyfe@cvm.msu.edu

MU - Animal Molecular Genetics Lab

University of Missouri College of Veterinary Medicine
320 Connaway Hall
Columbia, MO 65211
Email: hansenl@missouri.edu
www.caninegeneticdiseases.net/

Orthopedic Foundation for Animals

2300 E Nifong Boulevard
Columbia, MO 65201-3806
Tel: 573-442-0418
Fax: 573-875-5073
Email: ofa@offa.org

Paw Prints Genetics

850 E Spokane Falls Blvd, Suite 200
Spokane, WA 99202
Tel: (855) 202-4889
Email: askus@pawprintgenetics.com
www.pawprintgenetics.com

PennGen

3850 Spruce Street
Philadelphia, PA 19104-6010
Tel: 215-898-3375

UC-Davis, Veterinary Genetics Lab

PO Box 1102
Davis, CA 95617
Tel: (530) 752-2211
www.vgl.ucdavis.edu

VetGen

3728 Plaza Drive, Suite 1
Ann Arbor, MI 48108
Tel: 1-800-483-8436 (toll free)
www.vetgen.com

OCULAR DISORDERS REPORT

CARDIGAN WELSH CORGI - 1

CARDIGAN WELSH CORGI

| | DISORDER | INHERITANCE | REFERENCE | BREEDING ADVICE |
|----|--|------------------------------------|-----------|-----------------|
| A. | Distichiasis | Not defined | 1 | Breeder option |
| B. | Persistent pupillary membranes - iris to iris | Not defined | 2, 3 | Breeder option |
| C. | Cataract | Not defined | 1 | NO |
| D. | Retinal atrophy - rod-cone dysplasia type 3 (<i>rcd3</i>) * a DNA test is available | Presumed autosomal recessive | 1, 4-6 | NO |
| E. | Central progressive retinal atrophy | Not defined | 1, 7 | NO |

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 (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.

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,

OCULAR DISORDERS REPORT

CARDIGAN WELSH CORGI - 2

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 lack rod ERG responses seen at 6-8 weeks of age. Dogs are completely blind by 2-3 years of age when ophthalmoscopic signs are first visible. A DNA test is available. Current carrier rate from samples submitted is approx. 8.5%.

E. Central Progressive Retinal Atrophy (CPRA)

A progressive retinal degeneration in which photoreceptor death occurs secondary to disease of the underlying pigment epithelium. Progression is slow and some animals never lose vision. CPRA occurs in the United Kingdom, but is uncommon elsewhere.

References

1. ACVO Genetics Committee, 1999 and/or Data from CERF All Breeds Report, 1991-1998.
2. ACVO Genetics Committee, 2002-2003 and/or Data from CERF All-Breeds Report, 2002-2003.
3. ACVO Genetics Committee, 2005 and/or Data from CERF All-Breeds Report 2003-2004.
4. Petersen-Jones SM, Entz DD and Sargan DR. cGMP phosphodiesterase-alpha mutation causes progressive retinal atrophy in the Cardigan Welsh corgi dog. *Invest Ophthalmol Vis Sci.* 1999 Jul;40:1637-1644.
5. Petersen-Jones SM and 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 Jun;5:103-106.
6. Keep JM. Clinical aspects of progressive retinal atrophy in the Cardigan Welsh Corgi. *Aust Vet J.* 1972 Apr;48:197-199.
7. Barnett KC. Comparative aspects of canine hereditary eye disease. *Adv Vet Sci Comp Med.* 1976;20:39-67.

OCULAR DISORDERS REPORT

CARDIGAN WELSH CORGI

| Diagnostic Name | TOTAL DOGS EXAMINED | 1991-1999 1571 | | 2000-2009 1370 | | 2010-2013 547 | | 2014 122 | |
|---|---------------------|-------------------|------|-------------------|------|------------------|------|-------------|------|
| | | # | % | # | % | # | % | # | % |
| GLOBE | | | | | | | | | |
| 0.110 microphthalmia | | 1 | 0.1% | 1 | 0.1% | 0 | | 0 | |
| EYELIDS | | | | | | | | | |
| 25.110 distichiasis | | 51 | 3.2% | 60 | 4.4% | 18 | 3.3% | 3 | 2.5% |
| CORNEA | | | | | | | | | |
| 70.700 corneal dystrophy | | 8 | 0.5% | 5 | 0.4% | 1 | 0.2% | 1 | 0.8% |
| 70.730 corneal endothelial degeneration | | 0 | | 1 | 0.1% | 1 | 0.2% | 0 | |
| UVEA | | | | | | | | | |
| 93.110 iris hypoplasia | | 0 | | 0 | | 0 | | 1 | 0.8% |
| 93.150 iris coloboma | | 0 | | 1 | 0.1% | 0 | | 0 | |
| 93.710 persistent pupillary membranes, iris to iris | | 38 | 2.4% | 49 | 3.6% | 15 | 2.7% | 4 | 3.3% |
| 93.720 persistent pupillary membranes, iris to lens | | 1 | 0.1% | 2 | 0.1% | 0 | | 0 | |
| 93.730 persistent pupillary membranes, iris to cornea | | 3 | 0.2% | 5 | 0.4% | 1 | 0.2% | 0 | |
| 93.740 persistent pupillary membranes, iris sheets | | 0 | | 1 | 0.1% | 0 | | 0 | |
| LENS | | | | | | | | | |
| 100.200 cataract, unspecified | | 15 | 1.0% | 0 | | 0 | | 0 | |
| 100.210 cataract, significance unknown | | 47 | 3.0% | 45 | 3.3% | 8 | 1.5% | 5 | 4.1% |
| 100.301 punctate cataract, anterior cortex | | 5 | 0.3% | 4 | 0.3% | 1 | 0.2% | 0 | |
| 100.302 punctate cataract, posterior cortex | | 7 | 0.4% | 2 | 0.1% | 2 | 0.4% | 0 | |
| 100.303 punctate cataract, equatorial cortex | | 4 | 0.3% | 4 | 0.3% | 3 | 0.5% | 0 | |
| 100.304 punctate cataract, anterior sutures | | 2 | 0.1% | 0 | | 0 | | 0 | |
| 100.305 punctate cataract, posterior sutures | | 0 | | 1 | 0.1% | 1 | 0.2% | 0 | |
| 100.306 punctate cataract, nucleus | | 1 | 0.1% | 1 | 0.1% | 0 | | 0 | |
| 100.311 incipient cataract, anterior cortex | | 19 | 1.2% | 10 | 0.7% | 3 | 0.5% | 1 | 0.8% |
| 100.312 incipient cataract, posterior cortex | | 8 | 0.5% | 8 | 0.6% | 1 | 0.2% | 0 | |
| 100.313 incipient cataract, equatorial cortex | | 7 | 0.4% | 5 | 0.4% | 2 | 0.4% | 1 | 0.8% |
| 100.314 incipient cataract, anterior sutures | | 1 | 0.1% | 1 | 0.1% | 1 | 0.2% | 0 | |
| 100.315 incipient cataract, posterior sutures | | 1 | 0.1% | 0 | | 1 | 0.2% | 0 | |
| 100.316 incipient cataract, nucleus | | 3 | 0.2% | 4 | 0.3% | 0 | | 0 | |
| 100.317 incipient cataract, capsular | | 0 | | 2 | 0.1% | 0 | | 0 | |
| 100.330 generalized/complete cataract | | 6 | 0.4% | 1 | 0.1% | 1 | 0.2% | 0 | |
| VITREOUS | | | | | | | | | |
| 110.120 persistent hyaloid artery/remnant | | 4 | 0.3% | 0 | | 0 | | 0 | |
| 110.320 vitreous degeneration syneresis | | 3 | 0.2% | 0 | | 1 | 0.2% | 0 | |
| 110.330 vitreous degeneration anterior chamber | | 0 | | 2 | 0.1% | 1 | 0.2% | 0 | |
| FUNDUS | | | | | | | | | |
| 97.110 choroidal hypoplasia | | 0 | | 2 | 0.1% | 1 | 0.2% | 0 | |
| 97.120 coloboma | | 0 | | 2 | 0.1% | 0 | | 0 | |
| RETINA | | | | | | | | | |
| 120.170 retinal dysplasia, folds | | 13 | 0.8% | 10 | 0.7% | 1 | 0.2% | 0 | |
| 120.180 retinal dysplasia, geographic | | 4 | 0.3% | 1 | 0.1% | 1 | 0.2% | 0 | |
| 120.310 generalized progressive retinal atrophy (PRA) | | 8 | 0.5% | 1 | 0.1% | 0 | | 0 | |
| 120.400 retinal hemorrhage | | 1 | 0.1% | 0 | | 0 | | 0 | |
| 120.910 retinal detachment without dialysis | | 2 | 0.1% | 0 | | 0 | | 0 | |

OCULAR DISORDERS REPORT

CARDIGAN WELSH CORGI

| | 1991-1999 | 2000-2009 | 2010-2013 | 2014 |
|---------------------------------------|------------|------------|-----------|-----------|
| OPTIC NERVE | | | | |
| 130.120 optic nerve hypoplasia | 3 0.2% | 0 | 0 | 0 |
| OTHER | | | | |
| 900.000 other, unspecified | 0 | 8 0.6% | 8 1.5% | 0 |
| 900.100 other, not inherited | 3 0.2% | 35 2.6% | 3 0.5% | 3 2.5% |
| 900.110 other, suspected as inherited | 4 0.3% | 4 0.3% | 3 0.5% | 0 |
| NORMAL | | | | |
| 0.000 normal globe | 1357 86.4% | 1236 90.2% | 509 93.1% | 115 94.3% |