

8. The Veterinary ophthalmologists' advice relating to hereditary eye disease control

In this manual, we choose the term « Veterinary Ophthalmologists' advice relating to HED control » and intentionally avoid the words “pass”, “fail”, « certifiable » and « registerable ».

The ECVO does not prescribe breeding rules, nor does it serve as a registry organization. Breed clubs and registry organizations operate independently of the ECVO and set their own standards for registration. Any registry organization may use the information in this manual and the results of examinations performed by ECVO diplomates and panelists (Eye Scheme Examiners under the ECVO Eye Scheme) for the registration of animals with respect to their suitability for breeding.

It is important to recognize that the sensitivity of genetic disorder detection is greater when larger numbers of dogs are examined. The large number of disorders listed in this manual 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 the paucity of examinations reported for each breed. This may vary from country to country.

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

An ocular disorder is defined as **known to be hereditary** when:

- there are published reports in peer reviewed scientific literature regarding a condition in a particular breed with evidence of inheritance
- there are DNA-based tests available for the eye disease

An ocular disorder is defined as **presumed to be hereditary** when:

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

And more specifically, also when

- the incidence of affected animals (from a National ECVO Panel database, ECVO database and ACVO database which is the base of the OFA Eye Certification Registry (ECR) in the USA) is greater 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
- there is an overwhelming opinion by a majority of the HED committee members that clinical experience could indicate that a particular condition should be listed for a breed in spite of the absence of direct evidence of affected animals on the ECVO database, ECVO Panel database or the OFA ERC reports.
- there is a specific request from a breed club, that a condition be included for their breed. Such requests are reviewed critically and must have received agreement by a majority of the HED committee

The veterinary ophthalmologists' advice is determined by the significance of the condition to vision and/or wellbeing of the animal, such as a condition that is either painful or necessitating surgical intervention or lifelong medication and/or there is a very strong evidence of heritability. Note: in instances that the causal gene mutation is known and a DNA-based test is available the advice may be different. Further breeding advice may be necessary from a geneticist. A DNA-based test may never replace the ophthalmic examination.

Three categories of advice regarding breeding have been established:

- **OPTIONAL (low priority)**
- **NO BREEDING from the affected animal**

- **NO BREEDING from the affected animal, its parents, or its offspring**

1- OPTIONAL (low priority):

The defect is presumed to be hereditary (but there is no scientific evidence for its mode of inheritance) and the entity does not represent a prevalent or potential threat to vision or cause any significant reduced ocular function, pain or distress to the animal. The breeder may decide whether to breed the animal or not, preferably after consultation and discussion with the kennel club and/ or the breed club.

2- NO BREEDING from the affected animal:

Substantial evidence exists to support the heritability of this entity AND/OR the entity represents a major potential threat to vision or other reduced ocular function, pain or distress to the animal.

3- NO BREEDING from the affected animal, its parents, or offspring:

Substantial evidence exists to support the heritability of this entity AND/OR the entity represents a major threat to vision or other ocular function, pain or distress to the animal.

This category is to be used when the mode of inheritance of the entity is likely or known to be recessive and therefore the affected animal's parents and offspring are likely to be carriers of the gene mutation. Use of the relatives of the affected animal may be considered when a DNA-based test is available for the mutation.

In the presence of the DNA-based tests in certain circumstances, the use of affected animals and carriers may be wanted. All such matings should be carefully controlled and all offspring subjected to DNA-based tests.

The veterinary ophthalmologists' advice hereby given is based on the diagnoses obtained during the ophthalmic examination conducted by an ECVO examiner (diplomate or ESE) undersigning the eye examination certificate.

The diagnoses refer therefore 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 one or more genetic abnormalities.

The veterinary ophthalmologists' advice does not take into consideration the results of any genetic testing that may have been performed on the examined animal. This means that an animal may be for example DNA tested unaffected but is displaying clinical evidence of that inherited eye disease since that same disease may be caused by a different mutation in the same breed.

Finally, it is the opinion of the ECVO HED committee that it is up to the Kennel Clubs and/or the breed clubs to provide the actual Breeding Policy for each breed, which is often done together with geneticists. Other factors would then also be considered, such as frequency of a specific defect in the population and other defects affecting that specific breed of dog or cat, facilitating the establishment of an overall breeding policy.

The information contained in this manual can be used freely but the source (ECVO Hereditary Eye Disease committee) should be mentioned.

The veterinary ophthalmologists' advice relating to HED control, listed on the ECVO certificate, are given below for canine and feline breeds. The list is given in order to promote a general consensus among eye examiners in regards to evaluation of each disorder no matter breed (unless specified).

However, the advice might be different for a given breed, depending for example on the priority in a breed or the size of the actual breeding stock (the gene pool) in a certain breed. Specific advice is then given on the information sheet for each breed that follows in this manual.

Please note:

A - When the veterinary ophthalmologists' advice is **NO BREEDING** (either category 2 or 3), even a minor clinical form of the entity will make this animal (2), or this animal, its parents and its offspring (3) unsuitable

for breeding.

B -For each disorder, if an animal shows **a severe form of this entity (e.g. blinding or causing severe pain)**, the veterinary ophthalmologist's advice should be a category 2: **NO BREEDING** from the affected animal.

- Persistent Pupillary Membrane (PPM):
 - Iris to iris: OPTIONAL
 - Iris to lens: NO BREEDING from the affected animal
 - Iris to cornea: NO BREEDING from the affected animal

- PHTVL/PHPV:
 - Grade 1 OPTIONAL
 - Grade 2-6 NO BREEDING from the affected animal

- Cataract (congenital):
NO BREEDING from the affected animal

- Retinal Dysplasia (RD):
 - (Multi)focal form in any breed: OPTIONAL. Note: different advice may be given for specific breeds (e.g. English SpringerSpaniel, Golden Retriever, LabradorRetriever, Samoyed).
 - Geographic form: OPTIONAL. Note: different advice may be given for specific breeds (e.g. English Springer Spaniel, Labrador Retriever, Samoyed).
 - Total: NO BREEDING from the affected animal, its parents and offspring

- Micropapilla/Optic Nerve Hypoplasia
NO BREEDING from the affected animal

- Collie Eye Anomaly (CEA):
 - Choroidal hypoplasia (CH)/chorioretinal dysplasia (CRD): OPTIONAL
 - Coloboma and other defects (Retinal Detachment, Haemorrhage): NO BREEDING from the affected animal

- Other:
 - Anophthalmos: NO BREEDING from the affected animal

 - Atresia lacrimal punctum/micropunctum: OPTIONAL

 - Coloboma:
 - Eyelid: NO BREEDING from the affected animal
 - Papilla: NO BREEDING from the affected animal
 - Retina: NO BREEDING from the affected animal
 - Choroidea: NO BREEDING from the affected animal
 - Sclera: NO BREEDING from the affected animal
 - Dermoid: OPTIONAL

 - Eversion cartilage nictitating membrane : OPTIONAL

 - Hypoplasia:
 - Iris: OPTIONAL, Note: In severe cases: NO BREEDING from the affected animal (see under specific breeds e.g. Australian Shepherd, Rottweiler, Dalmatian)
 - Lens: NO BREEDING from the affected animal
 - Choroidea: OPTIONAL

 - Lenticonus: NO BREEDING from the affected animal

Lentiglobus: NO BREEDING from the affected animal
Microblepharon: OPTIONAL, Note: In severe cases: NO BREEDING from the affected animal

Macrophthalmos: NO BREEDING from the affected animal

Microphthalmos: NO BREEDING from the affected animal

Microphakia: NO BREEDING from the affected animal
Multiple ocular anomalies (2 or more): NO BREEDING from the affected animal

Persistent hyaloid artery: In severe cases: NO BREEDING from the affected animal

Prolapsed gland of the nictitating membrane: OPTIONAL
Retinal Dystrophy/RPE65 Mutation: NO BREEDING from the affected animal, its parents or offspring.
Breeders may utilize the DNA-based test to use carrier animals that have exceptional characteristics while avoiding the production of affected offspring. All such matings should be carefully controlled and all offspring subjected to DNA-based tests.

- Iridocorneal angle abnormality (ICAA)
Mild-moderate forms: OPTIONAL (according to present scientific information available: if these dogs are used, it is recommended to breed these dogs to unaffected graded dogs).
-Severe form: NO BREEDING from the affected animal.

Iridocorneal angle formation may progressively change with age from normal/unaffected to abnormal/affected (mild/moderate/severe) regarding PLA and ICA-width. Therefore, gonioscopy should be started before breeding and repeated every 3 years

- Entropion/Trichiasis: OPTIONAL
Note: In severe cases: NO BREEDING from the affected animal.
 - Ectropion / Microblepharon: OPTIONAL
Note: In severe cases: NO BREEDING from the affected animal
 - Distichiasis/ Ectopic Cilia: OPTIONAL.
Note: In severe cases: NO BREEDING from the affected animal.
 - Corneal Dystrophy:
Epithelial and/or stromal: OPTIONAL except in severe cases that cause visual problems and/or pain for the dog, e.g. in Siberian Husky or Shetland Sheepdog: NO BREEDING from the affected animal.
Macular dystrophy (e.g. Labrador Retriever): NO BREEDING from the affected animal
Endothelial dystrophy (e.g. Chihuahua, Boston Terrier, Dachshund). NO BREEDING from the affected animal
- 15- Cataract (hereditary, non congenital): NO BREEDING from the affected animal. : Note: different advice may be given for specific breeds (e.g. anterior punctate cataract in retrievers, pulverulent nuclear cataract in the Norwegian buhund).
- Lens Luxation (primary) (PLL): NO BREEDING from the affected animal, its parents or offspring (e.g. Small Terrier breeds, Chinese Crested Dog, Lancashire Heeler)

In instances where a DNA-based genetic test for recessive PLL is available breeders may choose to breed from carrier animals that have outstanding characteristics while still minimizing the risk of producing affected offspring. All such matings should be carefully controlled and all offspring subjected to DNA-based tests.

- Retinal Degeneration/Progressive Retinal Atrophy (PRA):
NO BREEDING from the affected animal, its parents or offspring
In instances where a DNA-based genetic test for recessive PRA is available breeders may choose to breed from carrier animals that have outstanding characteristics while still avoiding production of affected offspring. All such matings should be carefully controlled and all offspring subjected to DNA-based tests.

- Other:

Ceroid lipofuscinosis(CLN): NO BREEDING from the affected animal, Note: for specific breeds there exists a DNA-based genetic test

Chorioretinopathy, pigmentary: NO BREEDING from the affected animal
Glaucoma – primary: NO BREEDING from the affected animal

Keratitis, chronic superficial (CSK)/Pannus: NO BREEDING from the affected animal

Keratitis, punctate: NO BREEDING from the affected animal

Keratokonjunktivitis sicca (KCS): NO BREEDING from the affected animal(e.g. Cavalier King Charles Spaniel, Chinese Crested Dog, English Bulldog, Lhasa Apso, Long-haired Dachshund, Pug, Shih Tzu, West Highland White Terrier)

Melanoma iris: NO BREEDING from the affected animal

Ocular melanosis: NO BREEDING from the affected animal (e.g. Caim Terrier)

Other types of bilateral breed-related retinopathies: NO BREEDING from the affected animal

Retinal Pigment Epithelial Dystrophy (RPED): NO BREEDING from the affected animal

Retinopathy, Canine multifocal (CMR): NO BREEDING from the affected animal, Note: for specific breeds there exists a DNA-based genetic test

Uveal Cysts: OPTIONAL, Note: In severe cases the advice may be: NO BREEDING from the affected animal

Uveitis, pigmentary: NO BREEDING from the affected animal

Vitreous degeneration(*without any sign of lens luxation*): OPTIONAL

Vitreous strands/vitreous prolapsed (*without any sign of lens luxation*): OPTIONAL

Uveodermatologic Syndrome (UDS): NO BREEDING from the affected animal

Figures of the KP-HEDs are found on the ECVO website at <http://ecvo.org/hereditary-eye-diseases/images-for-panellists>