

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 panellists (ESE = 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 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.

Three categories of advice regarding breeding have been established:

1- OPTIONAL (low priority)

2- NO BREEDING from the affected animal

3- 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. If the affected dog is used, it is recommended to mate the dog with a dog that is « unaffected » for the same entity.

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 presumed or known to be recessive or (incomplete) dominant and therefore the affected animal's parents and offspring are at the least carriers of the gene mutation.

Only when a DNA-based genetic test for the mutation is available can the use of relatives of the affected animal be considered. Genetic testing has to be done from parents and offspring of the affected animal.

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

Note :

- The advice "NO BREEDING" (category 2 or 3) also applies to minor ("**suspicious**") or **mild** expressions of the entity on the assumption that the extent of the defect (minor, mild, moderate, severe) is based on the same genotype. If minor but specific clinical signs of the mentioned KP-HED (ticked as "suspicious") do not progress and are ticked as "unaffected" on re-examination (see requirements Chapter 3, paragraph 9.2.), the advice for breeding is: **OPTIONAL (low-priority)**
- For **each disorder**: if an animal shows a **severe form of this entity** (e.g. blinding or causing severe pain), the advice for breeding is **NO BREEDING** (category 2 or 3).
- If an animal displays clinical features that possibly fit the KP-HED, but changes are inconclusive, "**undetermined**" is ticked for the relevant disease; re-examination is recommended after 12 months (after 3 months in puppies); if clinical features remain inconclusive ("undetermined") the advice for breeding is: **OPTIONAL (low-priority)**

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.

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

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examination certificate. The diagnoses refer therefore only to the phenotype (clinical appearance) of an animal at the time of examination and may differ from a given genetic test result. Justifications: a) Animals homozygous for a recessive condition may develop the disease later in life; b) animals heterozygous for a recessive condition are carrier of one or more genetic abnormalities (abnormal genotype) but appear clinically normal (normal phenotype); c) an animal does not carry the tested mutation, but has clinical evidence of this inherited eye disease, as the same disease can be caused by a different mutation in the same breed. Therefore, a DNA-based test is not a substitute, but a useful additional diagnosis to the ophthalmologic examination. If relevant, name and result of genetic test can be mentioned in the comments field using the drop-down menu (open text).

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.

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, is 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.**

Eye Disease Number:

1- Persistent Pupillary Membrane (PPM):

- Strands iris to iris: OPTIONAL
- Strands iris to cornea: NO BREEDING from the affected animal
- Retrocorneal remnants without strands: OPTIONAL, *only if substantial*: NO BREEDING from the affected animal
- Strands iris to lens: NO BREEDING from the affected animal
- Fibrotic more or less pigmented tissue remnants on the anterior capsule of the lens, without strands: OPTIONAL, *only if substantial*: NO BREEDING from the affected animal
- Laminae: NO BREEDING from the affected animal

2- Persistent hyperplastic tunica vasculosa lentis/persistent hyperplastic primary vitreus (PHTVL/PHPV):

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

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

4- Retinal Dysplasia (RD):

- (Multi-)focal form in any breed: OPTIONAL. Note: different advice may be given for specific breeds by the breeding clubs

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- Geographic form: OPTIONAL. Note: different advice may be given for specific breeds by the breeding clubs.
- Total: NO BREEDING from the affected animal, its parents and offspring (exceptions see page 1, category 3)

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

6- Collie Eye Anomaly (CEA):

- Choroidal hypoplasia (CH)/chorioretinal dysplasia (CRD): OPTIONAL
- Coloboma and other defects (retinal detachment, haemorrhage): NO BREEDING from the affected animal

7- Other:

- Anophthalmos: NO BREEDING from the affected animal
- Coloboma:
 - Eyelid: NO BREEDING from the affected animal
 - Optic disc (Papilla): NO BREEDING from the affected animal
 - Retina: NO BREEDING from the affected animal
 - Choroida: NO BREEDING from the affected animal
 - Sclera: NO BREEDING from the affected animal
- Dermoid: OPTIONAL
- Exophthalmos due to shallow orbit: NO BREEDING from the affected animal
- Hypoplasia:
 - Iris: OPTIONAL, Note: In severe cases: NO BREEDING from the affected animal
 - Lens: NO BREEDING from the affected animal
 - Choroida: OPTIONAL
- Lacrimal punctum atresia/micropunctum: OPTIONAL
- Lenticonus: NO BREEDING from the affected animal
- Lentiglobus: NO BREEDING from the affected animal
- Macrophthalmos: NO BREEDING from the affected animal
- Microphthalmos: NO BREEDING from the affected animal
- Microblepharon: OPTIONAL, Note: In severe cases: NO BREEDING from the affected animal
- Microphakia: NO BREEDING from the affected animal
- Nictitating membrane, eversion of the cartilage: OPTIONAL
- Nictitating membrane, prolapse of the gland: OPTIONAL
- Multiple other KP-HED (3 or more anomalies): NO BREEDING from the affected animal
- Persistent hyaloid artery (PHA): OPTIONAL, Note: In severe cases: NO BREEDING from the affected animal
- Posterior segment exam not possible: NO BREEDING from the affected animal
- Retinal Dystrophy/RPE65 null mutation: NO BREEDING from the affected animal, its parents or offspring. (exceptions see page 1, category 3)

8- Iridocorneal angle abnormality (ICAA):

Clinical significance: ICAA is a predisposing factor - severe ICAA may lead to primary glaucoma.

- 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.

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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.

11- Entropion/Trichiasis: OPTIONAL; Note: In severe cases: NO BREEDING from the affected animal.

12- Ectropion/Macroblepharon: OPTIONAL; Note: In severe cases: NO BREEDING from the affected animal

13- Distichiasis/Ectopic Cilia: OPTIONAL; Note: In severe cases: NO BREEDING from the affected animal.

14- Corneal Dystrophy:

- Epithelial and/or stromal: OPTIONAL; Note: 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, later onset):

The breeding recommendation differentiates between clinically important and optional, low priority forms of cataracts:

- Cataract „cortical“: NO BREEDING from the affected animal
- Cataract „post. pol“: NO BREEDING from the affected animal
- Cataract „nucleus“: NO BREEDING from the affected animal
- Other lens opacity : OPTIONAL, low priority

OPTIONAL, low priority is valid for the following other lens opacities: Punctate, Suture line tips, Suture line, Nuclear ring, Nuclear fiberglass-like/pulverulent

16- Lens Luxation, primary (PLL):

NO BREEDING from the affected animal, its parents or offspring (e.g. Small Terrier breeds, Chinese Crested Dog, Lancashire Heeler); (exceptions see page 1, category 3)

17- Retinal Degeneration/Progressive Retinal Atrophy (PRA):

NO BREEDING from the affected animal, its parents or offspring; (exceptions see page 1, category 3)

18- Other:

- Canine multifocal retinopathy (CMR): NO BREEDING from the affected animal
- Ceroid lipofuscinosis (CLN): NO BREEDING from the affected animal, Note: for specific breeds there exists a DNA-based genetic test
- Chorioretinopathy, primary retinal disease (In Chinese Crested Dog, pigmentary chorioretinopathy). NO BREEDING from the affected animal.
- Chorioretinopathy, secondary retinal disease with genetic predisposition. (In specific breeds: Border Collie, Flat Coated Retriever and Borzoi, working dog retinopathy). 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

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- Keratoconjunctivitis sicca (KCS): NO BREEDING from the affected animal
- Multiple other KP-HED (3 or more acquired KP-HED): NO BREEDING from the affected animal
- Ocular melanosis: NO BREEDING from the affected animal (e.g. Cairn Terrier)
- Posterior segment exam not possible: NO BREEDING from the affected animal
- Retinal Pigment Epithelial Dystrophy (RPED) (in specific breed: English Cocker Spaniel): NO BREEDING from the affected animal
- Retinopathy, primary retinal disease. (In specific breeds: Shetland Sheepdog, Swedish Vallhund, Basenji, Labrador Retriever). NO BREEDING from the affected animal
- Uveal Cysts: OPTIONAL, Note: In severe cases the advice may be: NO BREEDING from the affected animal
- Uveal melanoma: NO BREEDING from the affected animal
- Uveitis, pigmentary: NO BREEDING from the affected animal
- Uveodermatologic Syndrome (UDS): 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

(Chorio-) retinopathy, inheritance under investigation: used in any breed in which a (chorio-) retinopathy is not described as specific; to be mentioned in descriptive comments: OPTIONAL

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