5. Definitions

See chapter 3, Introduction, and chapter 8, The Veterinary Ophthalmologists’ Advice, for a presentation in regards to when a disease, throughout this Manual, is considered a known hereditary eye disease or a presumed inherited eye disease. In short, a disease is considered a known hereditary eye disease when there is evidence for inheritance through scientific publication(s) and a DNA-based test is available. It is considered a presumed hereditary eye disease when the lesion has a characteristic age of onset and course of progression, and when the frequency of the problem is greater in a specific breed (Chapter 8).

For more detailed information in regards to definitions (below) the reader is referred to medical and genetic scientific texts.

Agenesis: congenital failure of development (see and use aplasia)

Albinism: localized absence of pigmentation, particularly in the iris and the choroid, may be accompanied by microphthalmia and other ocular defects (coloboma)

Allele: one of two or more alternative forms of a gene occupying corresponding sites (loci) on a pair of homologous chromosomes

Amblyopia: reduced visual acuity, without detectable anatomical defects in the optic media or fundus

Amaurosis: blindness, without detectable anatomical defects in the optic media or the fundus

Angle (iridocorneal): (geometric) angle between the base of the iris and the cornea adjacent to the limbus; the drainage angle. Aqueous humor leaves the anterior chamber via the pectinate ligament and the trabecular meshwork within the iridocorneal angle into the venous circulation. To determine if an iridocorneal angle abnormality (ICAA) exists, the pectinate ligament (PL) and the iridocorneal angle (ICA) width are evaluated by gonioscopy.

Anisocoria: different size of the individual’s pupils

Abnormality: deviation from normal

Anomaly: deviation from normal, especially as a result of congenital/developmental, non-progressive defects

Aniridia: see and use hypoplasia iris. the lesion has a characteristic age of onset and course of progression

Anophthalmos: absence of a true eyeball

Anterior: denote the front portion; e.g. the cornea is anterior to the lens

Anterior chamber: compartment between the cornea and iris, filled with aqueous humor

Anterior segment: compartment of the eye: from the cornea to the posterior lens capsule

Aplasia: congenital failure of development

Aqueous (humor): transparent fluid filling the anterior and posterior chambers

Asteroid hyalosis: White stationary crystalline precipitates suspended in the vitreous. May be seen as a result of vitreous degeneration, old age or systemic disease
Atresia of lacrimal punctum: Use Lacrimal punctum atresia.

**Autosome:** every normal *chromosome* which is differing from the sex *chromosome* in the degree of condensation, the way of motility and orientation and morphology; usually to be found in pairs

**Autosomal mode of inheritance:** mode of hereditary transmission of a characteristic whose *gene* is localized on an autosome

**Axis:** along an imaginary line connecting the center of the *cornea* and the *retina, axial* (adj.)

**Bergmeister’s papilla:** conical shaped remnant of the *hyaloid artery* attached to the *optic disc*

**Bilateral:** concerning both eyes, see and use OU

**Bulbar:** referring to the globe

**Bulbus luxation:** displacement of the globe through the lid fissure (or proptosis [= extensive exophthalmos] of the globe). Frequent in breeds with shallow orbit.

**Buphthalmos:** a secondarily enlarged globe, usually due to glaucoma

**Canine multifocal retinopathy (CMR): known hereditary eye disease:** autosomal mode of inheritance suspected. DNA-tests for specific breeds are available. Recognized as barely progressive, grey to tan bulging areas of circumscribed retinal detachments, generally more or less up to one optic disc diameter

**Canthus:** see eyelid

**Caruncle:** fleshy, haired eminence arising in the nasal canthus, extending into the *conjunctiva*; if hairs are misdirected, may cause conjunctival and/or corneal irritation

**Cataract:** any hereditary or non-hereditary, congenital or acquired, non-physiological opacity of the *lens* and/or its capsule. The defect may result in blindness if complete and bilateral. All bilateral or unilateral cataracts and especially cortical cataracts are *known and presumed hereditary eye diseases* except in cases known to be associated with trauma, other causes of ocular inflammation, metabolic disease, nutritional deficiencies, *persistent pupillary membrane, persistent hyaloid artery* or old age. DNA-tests for specific breeds are available.

**Ceroid lipofuscinosis (CLN): known hereditary 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 ataxia and blindness. DNA-tests for specific breeds are available.

**Cherry eye:** see and use prolapsed gland of the nictitating membrane

**Choroid:** thin vascular layer that lies between the *sclera* and *retina* in the *posterior* part of the eye

**Choroidal (retinal) hypo- (dys-) plasia (CH, CRD): Known hereditary congenital eye disease** which is characterized by inadequate development of the *choroid* present at birth which is nonprogressive. Most commonly identified in the Collie breed where it is a manifestation of “Collie Eye Anomaly”

**Chorioretinitis:** an inflammatory process of the *choroidal* and outer retinal structures, observed in the acute phase as blurring, swollen, oedematous areas and later as chorioretinal scaring as pigmented spots with hyperreflective borders
**Chromosomes**: rod- or hook-shaped structures that can be found as essential part of each cell nucleus in species-specific shape, inner structure and number; carriers of the genetic information

**Chronic superficial keratitis (CSK)/Pannus**: *Presumed hereditary eye disease*: bilateral inflammatory disease of the *cornea* which usually starts as a greyish haze at the *inferior* or *inferiortemporal* *cornea*, followed by the formation of a vascularized subepithelial opacity that begins to spread towards the central *cornea*; pigmentation follows the vascularization. Vision impairment occurs, if severe. The disease can be seen with concurrent plasmoma and/or medial canthus erosion

**Ciliated caruncle**: see and use *caruncle*

**Ciliary body**: see and use *corpus ciliare*

**Ciliary body cysts**: pigmented cysts arising from pigmented epithelial cells of the *corpus ciliare* - use *uveal cysts*

**Ciliary cleft**: triangular extension of the anterior chamber into the ciliary body, anteriorly lined by the pectinate ligament and containing wide spaces, interspersed with cell-lined cords of connective tissue, defined as the trabecular meshwork

**Ciliary processes**: 60 to 80 folds of the *corpus ciliare* that produce *aqueous humour*

**Co-dominance**: refers to a set of three *phenotypes* controlled by a pair of *alleles*. The *heterozygote* displays a *phenotype* either intermediate between, or distinctly different from the two *homozygous* *phenotypes*

**Collarette**: see iris collarette

**Collie Eye Anomaly (CEA): *known hereditary congenital eye disease***: a *congenital* syndrome of ocular anomalies mainly in Collie breeds affecting the choroid and sclera and indirectly the retina and optic disc. It is characterized by bilateral and often symmetrical defects including *choroidal hypoplasia (CH, CRD)* with or without *coloboma*, *retinal detachment* and intraocular hemorrhage. Vision varies with the degree to which an individual is affected and may be minimally compromised to having severe visual impairment or blindness. DNA-tests for choroidal hypoplasia in specific breeds are available.

**Coloboma**: congenital defect of a portion of the eye due to a failure in closure of the body halves; most frequently affecting the *iris* or the *optic nerve* at the 6 o’clock position. The latter is a *presumed hereditary congenital eye disease* that if large, may cause retinal detachment resulting in blindness or visual impairment. When there is a congenital absence of iris tissue, see and use *iris hypoplasia*. Consequently, for coloboma in eyelid, retina, choroidea, sclera or optic nerve/papilla use the anatomical name first then the anomaly, e.g. eyelid coloboma, retinal coloboma, choroidal coloboma, scleral coloboma and/or optic nerve coloboma.

**Cone degeneration (CD): *known hereditary eye disease***, characterized by abnormal development of cones causing day blindness with normal fundus appearance. DNA-tests for specific breeds are available.

**Cones**: primary visual cells of the eye functioning in bright light providing sharp visual acuity and colour sensitivity

**Cone rod dystrophy (CRD): *known hereditary retinal disease*** characterized by abnormal development of cones and rods, in which the cones are affected earlier/ more severely than rods. Clinical signs may vary but affected animals become day blind early in life. An *electroretinogram (ERG)* is diagnostic. DNA-tests for specific breeds are available.

**Congenital**: condition present at birth, when the eye lids open, or in the first 6 to 8 weeks of life (dog or cat), which may or may not be hereditary
**Congenital stationary night-blindness (CSNB):** presumed hereditary congenital eye disease that is non-progressive with abnormal or absent rod function. An electroretinogram (ERG) is diagnostic.

**Conjunctiva:** thin vascular membrane which covers the sclera (bulbar conjunctiva), the nictitating membrane and the inner surfaces of the upper and lower eyelids (tarsal conjunctiva)

**Cornea:** transparent structure forming the front of the eye; continuous with the sclera at the limbus

**Corneal degeneration:** cell death in one or more of the layers of the cornea which may be spontaneous or secondary to other ocular conditions. Occurs uni- or bilaterally and can be associated with inflammatory response, i.e. vascularisation or fibrosis as opposed to corneal dystrophy

**Corneal dystrophy:** presumed hereditary eye disease; non-inflammatory corneal opacity in one or more of the corneal layers (epithelium, stroma, endothelium). It is usually bilateral but not always symmetrical. The onset in one eye may precede the other

**Corneal dystrophy, endothelial:** abnormal loss of the inner lining (endothelium) of the cornea causing progressive fluid retention (edema) leading to increased corneal thickness, keratitis, corneal clouding and decreased vision

**Corneal dystrophy, epithelial / stromal:** non-inflammatory corneal opacity (white to grey with crystalline appearance) in one or more of the corneal layers. Often associated with deposits of cholesterol and other lipids (or fats) within the cornea

**Corneal dystrophy, macular:** known hereditary eye disease; there is a bilateral diffuse haziness of the cornea and there are multiple whitish/grey macula like lesions throughout the corneal stroma. The periphery appears slightly less affected. Density and size of the lesions progresses throughout life leading to quite severe visual impairment. DNA-tests for specific breeds are available.

**Corneal edema:** fluid accumulation within the cornea resulting in cloudiness

**Chronic corneal erosion**/indolent ulcer/ superficial chronic corneal epithelial defect, SCCED: type of superficial corneal ulcer; defect of the corneal epithelium due to degenerative basal cell layer

**Corpus ciliare:** middle part of the uveal tract, containing the pars plicata (ciliary processes) and pars plana (ciliary muscles)

**Day blindness:** loss of photopic (daylight) vision caused by abnormal cone function

**Dermoid:** presumed hereditary eye disease; a congenital patch of skin in an abnormal location. Most ocular dermoids affect the cornea or adjacent conjunctiva, and its presence usually causes ocular irritation

**Descemet’s membrane:** the basement membrane of the corneal endothelium

**Distichiasis:** presumed hereditary eye disease; single or multiple hairs (cilia) from an abnormally located hair follicle in the eyelid margin, usually growing from or in between the Meibomian glands, and arising from the Meibomian duct openings, which may cause ocular irritation. The defect is due to abnormal differentiation of a tarsal gland. Distichiasis usually occurs at an early age (< 1-2 years), but may occur any time in life

**Dominant:** describes the mode of hereditary transmission such that only one of the two genes of a pair must be affected in order for the individual to demonstrate the characteristic controlled by that gene. A completely dominant phenotype is identical in individuals either heterozygous or homozygous for the responsible allele. Incomplete dominance is used variably to refer to incomplete penetrance, incomplete expressivity, or codominance

**Dry eye:** see and use keratoconjunctivitis sicca
Dysgenesis: see and use dysplasia

Dysplasia: abnormal development or growth

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

Ectopic cilia: presumed hereditary eye disease; single or multiple hairs (cilia) from an abnormally located hair follicle in the eyelid margin, usually growing from or in between the Meibomian glands emerging through the eyelid conjunctiva. Ectopic cilia occur more frequently in younger dogs. They generally cause severe discomfort and corneal disease

Ectropion: presumed hereditary eye disease; a conformational defect resulting in eversion (rolling-out) of the margin of the eyelids, which may cause ocular problems due to exposure. In the hereditary forms, 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 skin covering the head and face, the orbital contents and the conformation of the skull. Secondary, non-hereditary ectropion may also occur, for example due to iatrogenic, trauma or scarring

Ectropion with macroblepharon: presumed hereditary eye disease; ectropion associated with an excessively large lid 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.

Electroretinogram: a graphic record of the electrical response that follows stimulation of the retina by light

Electroretinography (ERG): an electrophysiological test of retinal function

Endothelium (of the cornea): the innermost layer of the cornea

Enophthalmos: abnormal deep positioning of the globe within the orbit (opposite of exophthalmos)

Entropion: presumed hereditary eye disease; a conformational defect resulting in “in-rolling” of one or both of the margins 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. Secondary, non-hereditary entropion may also occur, for example due to trauma, severe enophthalmos, loss of orbital fat, etc.

Epiphora: overflow of tears onto the face; may be caused either by increased tear production or reduced tear drainage through the nasolacrimal duct

Epithelium of the cornea: the outermost layer of the cornea

Esotropia: see and use strabismus (squint) convergens

Eversion of the cartilage of the nictitating membrane: presumed hereditary eye disease; scroll-like curling of the cartilage of the nictitating membrane, usually evertting the margin. The condition may occur in one or both eyes and may cause mild ocular irritation

Exophthalmos: protrusion of the eyeball beyond the bony orbit (opposite of enophthalmos)

Exotropia: see and use strabismus divergens (squint)
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

Expressivity: refers to the phenotypic expression, or clinical appearance, of a given genotype. Variable expressivity refers to a range of different phenotypes, all representing the same genotype at a given locus

Eyelids: the moveable folds of skin and muscle over the superior and inferior portions of the eye.

Lid canthus: the nasal and temporal or junction of the upper and lower eyelids

Lid fissure: slit opening between eyelids

Fibrae latae: presumed hereditary eye disease, pectinate ligament fibres with either a confluent (broad) base and shortened thin insertions at the cornea or formation of thick fibres (< 5 fibres)

Fissure: see eyelids

Fundus: the posterior portion of the interior of the eye as viewed with an ophthalmoscope; observed in most domestic animals with the tapetum lucidum or tapetal area and the non-tapetal area

Gene: information unit for the development of an hereditary characteristic which has been identically reproduced within the body cell and which has been distributed among the daughter cells; genes are lined up in a row in chromosomes

Gene mutation: mutation concerning a single gene which can be detected by a different genetic product (e.g. defect of an enzyme)

Gene test/ genetic testing: identification of animals carrying or not carrying the mutant disease gene by revealing the animal's genotype for the disease in question

Genotype: refers to the allele(s) present at one or more genetic loci. Most commonly refers to the pair of alleles (either identical or different) present at a single chromosome locus; distinct from phenotype

Glaucoma, primary: known or presumed hereditary eye disease in several dog breeds and in a few cat breeds. The disease process has a complex etiology. It is characterized by an elevation of intraocular pressure (IOP) which, when sustained, results in destruction of intraocular structure and function, resulting in blindness. The elevated intraocular pressure occurs mainly with developmental abnormalities or disease processes affecting the intraocular circulation and especially the drainage of aqueous humor from the eye through the irido-corneal angle. Diagnosis and classification of glaucoma requires measurement of the IOP (tonometry) and examination of the iridocorneal angle (gonioscopy). DNA-tests for Primary Open Angle Glaucoma (POAG) in specific breeds are available.

Glaucoma, pigmentary: see and use ocular melanosis

Goniodygenesis/ goniodysplasia: see and use pectinate ligament abnormality (PLA).

Gonioscopy: a procedure which uses a contact lens to examine the iridocorneal angle (ICA) to evaluate the ICA width and the pectinate ligament

“Go normal” (“masked”): A term that is used in the context with the collie eye anomaly syndrome. It describes the insufficient development of the choroid, diagnosed in the 5th to 7th week after birth; camouflaged by choroidal cell material after the 7th to 10th week leading therefore at a later examination to the judgement of “no abnormalities or normal”
Hemeralopia: see and use day blindness. The same term can mean night blindness in Latin based countries. Therefore the wording day or night blindness is preferred in the scheme to prevent misunderstanding.

Hereditary: genetically transmitted from parent to offspring

Heterochromia iridis: difference of colour in the two irides of the same animal or in different areas of the same iris in one eye (the latter: heterochromia iridum)

Heterozygote: an individual in which the members (or alleles) of a given pair of genes are dissimilar; heterozygous, adj.

Homozygote: an individual in which the members (or alleles) of a given pair of genes are alike; homozygous, adj.

Hyaloid artery (HA): embryological artery which nourishes the lens; arising from the optic papilla to the posterior pole of the lens and regresses before birth

Hyperopia: farsightedness

Hypoplasia: defective development of an organ or part resulting in a smaller than normal size or immature state

Hypoplasia iris: presumed hereditary eye disease characterized by congenital absence of iris (sphincter) tissue or colobomatous defects due to failure in closure of the optic fissure. It may be a separate disorder or associated with other ocular malformations. See and use iris hypoplasia

Hypoplasia lens: presumed hereditary eye disease characterized by congenital incomplete formation of the lens equator, sometimes called lens coloboma. See and use lens hypoplasia

Hypoplasia- optic disc hypoplasia: presumed hereditary eye disease: congenital failure of development of the optic nerve which causes blindness and abnormal pupil response in the affected eye. Can often not be differentiated from micropapilla on a routine (dilated) ECVO eye examination

Immune-mediated disease: a state in which the immune responses, which are essential to the protection of the body, act in an enhanced and unregulated fashion resulting in damage or destruction of autogenous (self) bodily tissues

Imperforate lacrimal punctum: see and use atresia of lacrimal punctum

Incidence: rate at which a certain event occurs, e.g., the number of new cases of a specific disease occurring during a certain period

Indolent ulcer: see (chronic) corneal erosion

Inferior: (also referred to as ventral) lower region

Intraocular pressure (IOP): the pressure formed by a balance between intraocular fluid production and outflow, measured with a tonometer (applanation or rebound)

Iridocorneal angle (ICA): (geometric) angle between the base of the iris and the cornea adjacent to the limbus and anterior opening of the ciliary cleft, spanned by the comb-like pectinate ligament.

Iridocorneal angle (ICA) width: the width of the ICA is evaluated (using gonioscopy) by comparison of the length of the pectinate ligament (A) and the distance from the origin of the pectinate ligament to the anterior surface of the cornea at the transection area (B);
The ICA width is judged as open (normal) if the length of the pectinate ligament (A) is equal to or more than 1/3 of B ($A \geq 1/3$ of B);

The ICA is judged as abnormal if:

a) The ICA is narrow: A is smaller than 1/3 of B ($A < 1/3$ of B) and the visible length of the pectinate ligament is severely reduced.

b) Or closed (collapsed) and pectinate ligament not visible

**Grading of the ICAW**

<table>
<thead>
<tr>
<th>Terminology</th>
<th>Ratio A/B:</th>
</tr>
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<tbody>
<tr>
<td>closed</td>
<td>PL not visible</td>
</tr>
<tr>
<td>narrow</td>
<td>$A &lt; 1/3$ of B</td>
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<tr>
<td>open</td>
<td>$A \geq 1/3$ of B</td>
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**Open:** PL length (A) is equal to or more than 1/3 of B; $A \geq 1/3$ of B

**Narrow:** PL length (A) is smaller than 1/3 of B; $A < 1/3$ of B (visible length of PL is severely reduced)

**Closed:** PL not visible = collapsed/closed angle


**Iridodonesis:** quivering of the iris, indication of lens (sub)luxation

**Iris:** the visible, coloured portion of the vascular tunic of the eye, situated in front of the **lens**, with a central opening, the **pupil**

**Iris atrophy:** degenerative loss of iris tissue, to be differentiated from iris **coloboma/hypoplasia**. May occur spontaneously as aging change or be secondary to inflammation or **glaucoma**

**Iris collarette:** area of the annular vessel of the optic cup, where the vascular loops of the **pupillary membrane** (PM) start from and where the vessels of the anterior tunica vasculosa lentis and the PM end. In the case of persistent pupillary membranes, the remnants are attached to the surface of the iris in this area and not to the pupillary margin of the iris

**Iris coloboma:** see and use iris **hypoplasia**

**Keratitis:** non-specific inflammation of the **cornea**; may or may not be associated with infection

**Keratitis, chronic superficial:** see and use **chronic superficial keratitis**

**Keratitis, punctate:** presumed **hereditary eye disease**; inflammation of the **cornea** accompanied by multiple small areas of corneal ulceration

**Keratoconjunctivitis sicca (KCS):** presumed **hereditary eye disease** in some dog breeds. An abnormality of the **tear film**, most commonly a deficiency of the aqueous portion, although the mucin and/or lipid layers may be
affected. Progressive KCS may result in ocular irritation and vision impairment. Often called “dry eye”. Secondary, non-hereditary KCS may also occur, for example due to intoxication, iatrogenic, trauma, neurogenic or infection.

**Lacrimal punctum:** one of the two small openings at the nasal canthal margin of the palpebral conjunctiva which drain the tears away from the eye and into the nasolacrimal drainage system. Abnormalities in the lacrimal puncta may result in epiphora.

**Lacrimal punctum, atresia:** presumed hereditary eye disease; 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

**Lagophthalmos:** failure to close the eyelids completely; results in exposure of the cornea and conjunctiva

**Lamina (LA):** presumed hereditary eye disease; pectinate ligament fibres form plates or sheets of continuous tissue (>5 fibres), with or without flow holes

**Lateral:** see temporal

**Lens:** biconvex refractive structure within the eye suspended between the iris and the retina that focuses sharp images on the retina for acute vision. The axial anterior and posterior parts of the lens are referred to as the poles. The outermost membrane surrounding the lens is referred to as the lens capsule. The center of the lens is the nucleus. The remainder of the lens is the cortex. The zonules are attached to the periphery of the lens (equator) and give support to the lens

**Lens nuclear sclerosis:** normal bilateral aging change of the lens nucleus, which is characterized by a hardening and dehydration (sclerosis) and which does not cause distinct visual impairment

**Lens suture lines:** junction of the lens fibers at the poles. The anterior lens suture lines in dogs and cats are generally in the pattern of an upright Y and posterior in an inverted Y

**Lens luxation (primary):** known hereditary eye disease; partial (subluxation) or complete displacement of the lens from the normal anatomic site, in the fossa patellaris, behind the pupil. Lens luxation may result in elevated intraocular pressure (glaucoma) causing vision impairment or blindness. DNA-tests for specific breeds are available.

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

**Lentiglobus:** sphere-shaped deformity of the lens (anterior or posterior)

**Ligamentum pectinatum abnormality:** see and use Pectinate ligament abnormality (PLA)

**Limbus:** junction between the cornea and the sclera

**Macular corneal dystrophy (MCD):** see and use Corneal dystrophy

**Macroblepharon:** presumed hereditary eye disease; an exceptionally large palpebral fissure. Macroblepharon in conjunction with laxity of the lateralcanthal structures may lead to lower lid ectropion in combination with lateral entropion and upper lid entropion and trichiasis. This may in severe cases sometimes result in diamond-shaped eyes. Either of these conditions may lead to conditions associated with corneal exposure

**Macrophthalmos:** congenital enlarged globe
Medial: see nasal

Medial canthus erosion: Localised erosive dermatitis in the medial canthus, can be seen in conjunction with chronic superficial keratitis or plasmoma, as part of CSK/pannus

Meibomian glands: secretory glands located in the eyelid margin which produce the oily portion of the tear film.

Melanoma iris: presumed hereditary eye disease locally invasive cancer of melanocyte (pigment) cell origin within the iris. Occurs with a higher than normal incidence in the Labrador retriever. Left untreated it may result in secondary glaucoma.

Merle: refers to an incompletely dominant phenotype present in several breeds. Heterozygous individuals (M/m) have a coat colour phenotypically characterized by dilute patches (i.e. blue, grey, cream or white) that vary irregularly in size, extent and intensity of colour. Deafness and ocular defects are sometimes seen in heterozygous individuals. Homozygosity (M/M) is sublethal. Homozygous individuals surviving to birth exhibit marked hypopigmentation, ocular defects including microphthalmia, blindness and colobomas, and deafness (sometimes referred to as “multiple ocular syndrome”)

Microblepharon: presumed hereditary eye disease an exceptionally short palpebral fissure. Microblepharon may lead to upper lid entropion and trichiasis.

Microcornea: congenital, abnormal small diameter of the cornea

Micropapilla: small optic disc which is not associated with vision impairment. May not be differentiated from hypoplastic papilla/ optic disc on a routine, dilated ECVO-eye examination

Microphakia: congenital developmental anomaly in which there is an abnormally small lens

Microphthalmos (microphthalmia): presumed hereditary eye disease, congenital/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

Micropunctum: abnormally small lacrimal punctum

Miosis: constricted pupil

Mittendorf’s dot: conical remnant of the hyaloid artery attached to the posterior capsule of the lens just below the juncture of the posterior lens suture lines

Multiple ocular anomalies (two or more): presumed hereditary eye disease, congenital/developmental, mostly non-progressive anomalies found in the same animal (to be specified in the certificate descriptive comment field). The anomalies found can be e.g. microphthalmia, iris hypoplasia, persistent pupillary membranes, lens anomalies, posterior segment colobomata or other developmental defects. The syndrome is also recognized in relation to the merle gene, especially as a result of merle to merle matings

Mydriasis: dilated pupil

Myopia: near-sightedness

Nanophthalmos: see and use microphthalmos, as other anomalies are difficult to exclude. Congenital, abnormally small but anatomically functional globe.
Nasal: the region of the eye located towards the nose (see medial)

Nictitating membrane: a triangular-shaped structure that consists of a T-shaped cartilage (to provide form and support) and a tear gland, which are covered on the anterior and posterior side by conjunctiva. It is situated in the nasal canthus. It serves as a protective function for the eye and occasionally protrudes across the eye. Also called the third eyelid, nictitans, or haw

Night blindness: loss of scotopic (night) vision caused by a loss of rod function

Non-tapetal fundus (non-tapetum): refers to that area of the fundus where there are no clinically visible reflective cells

Nuclear lens sclerosis: see and use lens nuclear sclerosis

Nyctalopia: see and use night blindness. The same term can mean day blindness in Latin based countries. Therefore the wording day or night blindness is preferred in the scheme to prevent misunderstanding

Ocular melanosis: presumed hereditary eye disease: an abnormal proliferation of melanocytes within the uveal tract that may cause an elevation of the intraocular pressure/glaucoma when an obstruction of the aqueous outflow pathways occurs, occurs with a higher than normal incidence in the Cairn terrier.

Oculus dexter (OD): right eye

Oculus sinister (OS): left eye

Oculi uterque (OU): both eyes

Optic papilla/ optic disc/ optic nerve head: the part of the optic nerve which is visible, by ophthalmoscopic examination, in the fundus

Optic nerve hypoplasia: see and use hypoplastic papilla/ optic disc

Palpebral: associated with the eyelids

Pannus: see and use chronic superficial keratitis (CSK)/pannus

Pars plana/ ora serrata: the peripheral margin of the fundus where the neuroretina ends and is attached. Usually it is attached here in bullous retinal detachment

Papilla, Bergmesiter: see Bergmeister papilla

Pectinate ligament: thin, filamentous fibres radiating from the base of the iris and inserting into the inner surface of the cornea as the entrance of the aqueous drainage system

Pectinate ligament abnormality (PLA): presumed hereditary eye disease: characterized by an abnormal pectinate ligament that can be divided into 2 predominant types:
1. Fibrae latae
2. Lamina
Diagnosis is by gonioscopy.

Penetrance: refers to the proportion of heterozygous individuals expressing the (relatively dominant) phenotype characteristic of the homozygotes. Incomplete penetrance means that less than 100% of the heterozygous individuals express the (relatively) dominant phenotype
Peri-: a prefix meaning around. E.g. peri-nuclear is around the nucleus, which means in the lens cortex

**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 grey-white (persistent hyaloid remnant)

**Persistent hyperplastic tunica vasculosa lentis/ persistent hyperplastic primary vitreous (PHTVL/PHPV):** **known or presumed hereditary, congenital eye disease** which results from failure of regression of the embryologic vascular network, surrounding the developing lens and primary vitreous. The latter fails to regress within the first 2-3 weeks after birth. The defect is currently graded in 6 levels of severity, in which grade 1 is characterized by uni- or bilateral small, yellow to brown dots mainly centrally, retro lentally on the posterior capsule of the lens. These are stationary and do not affect vision. The more severe forms (2-6) usually occur bilaterally and cause visual impairment or blindness. Known hereditary e.g. in the Dobermann and the Staffordshire Bull terrier

**Persistent pupillary membrane (PPM):** **presumed hereditary congenital eye disease** in which blood vessel remnants of the embryological vascular network in the anterior chamber of the eye fail to regress which normally occurs during the first 4 to 5 weeks of life. These remnants may be found on the surface of the iris at the colarette, the lens capsule or against the corneal endothelium or strands may bridge from iris to iris, iris to cornea, iris to lens, with or without sheets of tissue in the anterior chamber. The last three forms pose the greatest threat to vision and, when severe, vision impairment may occur.

**Phenotype:** physical appearance. Distinct from genotype

**Photopic vision:** daylight vision, vision in high light intensities

**Photoreceptors:** see rods and cones

**Pigmentary chorioretinopathy:** **presumed hereditary eye disease** occurs with a higher than normal incidence in the Chinese Crested dog breed. Recognized as bilateral, progressive, circumscribed areas with pigmented or light-colored center, leading to visual impairment or blindness.

**Pigmentary glaucoma:** see and use ocular melanosis

**Pigmentary uveitis:** see uveitis, pigmentary

**Plasmoma:** hyperplastic and hypo-(de-)pigmented margins of the membrana nictitans due to accumulation of inflammatory (plasma) cells. Part of the CSK/pannus syndrome

**Pole:** either extreme of the axis; usually applied to the anterior or posterior axial surfaces of the lens; polar, adj.

**Posterior:** denotes the back portion; e.g. the lens is posterior to the cornea

**Posterior chamber:** compartment between the iris and the lens, zonules and vitreous face, filled with aqueous humor

**Posterior segment:** compartment of the eye: from the vitreous face to the sclera

**Prevalence:** The percentage of a population that is affected with a particular disease at a given time
Prolapsed gland of the nictitating membrane: presumed hereditary eye disease: protrusion of the tear gland associated with the nictitating membrane. The exposed gland may become irritated. Commonly referred to as “cherry eye”

Progressive rod-cone degeneration (PRCD): known hereditary eye disease; progressive rod-cone degeneration, see progressive retinal atrophy (PRA)

Progressive Retinal Atrophy (PRA): see Retinal degeneration

Ptosis: drooping of the upper eyelid

Pupil: central opening of the iris

Pupillary membrane: embryological vascular network nourishing the anterior surface of the lens which is formed during gestation and regresses up to 4-6 weeks after birth. Failure of complete regression results in persistent pupillary membrane (PPM)

Rod-cone dysplasia (RCD): presumed hereditary congenital eye disease. See rod-cone dysplasia

Recessive: mode of inheritance in which both genes must be alike in order for the characteristic to be expressed in an individual. For a recessively hereditary condition, both genes must be abnormal for the disease to be present

Retina: A bilayered structure consisting of the retinal pigment epithelium and the neurosensory retina, the latter layer including the photoreceptor cells (rods and cones)

Retinal degeneration/Progressive Retinal Atrophy (PRA): known hereditary eye disease; a group of bilateral, hereditary dystrophic and/or degenerative diseases of the photoreceptors primarily, progressing to blindness in both eyes simultaneously. The onset of the blindness depends on the affected breed and the type of process (dysplasia and/or degeneration). The photoreceptor abnormalities can be detected by an electroretinogram (not part of a routine ECVO Scheme eye examination) before there are detectable fundus changes observed by ophthalmoscopy. These funduscopic changes consist in the early disease of a change in reflectivity with greyish discoloration mainly in the periphery and midperiphery in the tapetal area of the fundus accompanied by slight vascular attenuation. With progression of the disease there are more generalized changes with hyperreflectivity of the tapetal fundus, degeneration and uneven pigment distribution in the non-tapetal fundus, severe vascular attenuation and a pale optic disc. There are multiple genetic types of PRA including different forms of rod-cone dysplasia and degeneration (rcd1-4) and progressive rod cone degeneration (prcd). DNA-tests for specific forms and breeds are available.

Retinal degeneration can also be due to non-hereditary causes, e.g. inflammation and/or infection, toxicity, etc., affecting retinal structures with degeneration of cells or entire cellular layers. The end-stage is often complete retinal atrophy, which may appear ophthalmoscopically similar to (hereditary) PRA

Retinal detachment: separation of the neuroretina from the underlying tissue (the retinal pigment epithelium). It results in blindness when complete. Presumed hereditary eye disease if part of the retinal diseases e.g.: collie eye anomaly (CEA) or retinal dysplasia (RD)

Retinal detachment- Bullous: In the bullous type, there is a fluid filled space under the neuroretina, which is attached to the pars plana/ora serrata and the papilla.

Retinal detachment – Rhegmatogenous: In the rhegmatogenous type, tears in the neuroretina are seen and the neuroretina may be detached from the pars plana/ora serrata.
Retinal dysplasia: presumed hereditary eye disease: abnormal development of the retina with ophthalmoscopically observed early in life, characterized by neuroretinal folding(s), rosettes and partial or total retinal detachment; non-progressive and generally recognized to have three forms: (multi)focal, geographic and total.

Retinal dysplasia- (multi)focal: seen ophthalmoscopically as linear (vermiform), 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. The two other forms of retinal dysplasia (geographic and complete) which are known to be hereditary in some breeds and, in their most severe form, may cause blindness. The genetic relationship between folds and more severe forms of retinal dysplasia is undetermined.

Retinal dysplasia- geographical: any irregularly, horseshoe- or bladder-like shaped area of abnormal retinal development, most often in the central part of the tapetal area of the fundus, in close association with the dorsal retinal vasculature, containing both areas of thinning and areas of elevation representing focal retinal detachment and areas of retinal disorganization. This form may be associated with vision impairment.

Retinal dysplasia- total: severe retinal disorganization associated with total separation (detachment) of the retina. The geographic and total forms of retinal dysplasia are associated with partial or complete vision impairment or blindness and can be diagnosed already in puppies. Retinal dysplasia is known to be hereditary in many breeds. The genetic relationship between the three forms of the disease is not known for all breeds.

Retinal dystrophy/ RPE 65 null mutation: known hereditary eye disease, usually with bilateral, concomitant deterioration of retinal structure and function. In the Briard dog the retinal dystrophy (due to lack of the RPE65 protein) causes congenital night blindness and partial or complete day blindness. Disease also called Congenital Stationary Nightblindness (CSNB) in some publications. A DNA-test for the RPE65 null mutation of Briard dogs is available.

Retinal folds: hereditary or nonhereditary changes in the retina, can be neuroretinal folding due to hereditary factors or as sequelae post inflammation.

Retinal pigment epithelial dystrophy (RPED): accumulation of lipid pigments in the retinal pigment epithelium. There is strong evidence that vitamin E and taurine are involved in the etiology of RPED. Hereditary factors may be involved with the disease.

Retinopathy: any non-specific hereditary or non-hereditary disease condition of the retina, usually detected by ophthalmoscopic examination.

Retinopathy, multifocal bullous: see and use canine multifocal retinopathy (CMR).

Retinoscopy: an objective method to measure the error of refraction of the eye. Used to determine the degree of nearsightedness (myopia) or far-sightedness (hyperopia).

Retro: a prefix meaning: behind a structure or positioned posterior to a structure.

Rods: primary visual cells of the eye functioning in dim or reduced illumination, and with the 2nd and 3rd order retinal neurones providing for detection of shapes and motion.

Rod-cone dysplasia (rcd): presumed hereditary retinal disease: characterized by abortive or abnormal development of rods and cones, in which the rods are affected earlier / more severely than cones. Affected animals become blind early in life, usually within the first 6 months. Different types of rcd’s have been described. An ERG is diagnostic.

Rod dysplasia: presumed hereditary retinal disease: abnormal development of the rod visual cells resulting in vision impairment in dim light usually within the first 6 months of life and total blindness at 3-5 years.
**Sclera**: white, opaque, outer layer of the eyeball, covered by tenons capsule and conjunctiva in the anterior part of the globe, extending to limbus

**Scotopic vision**: night vision, vision in low light intensities

**Semi-dominance**: used variably to refer to either co-dominance, incomplete penetrance or variable expressivity

**Staphyloma**: localized weakness of tissue (usually sclera or cornea) resulting in a bulging of the affected area. Usually an acquired condition in contrast to coloboma

**Strabismus**: non-parallel eye axis or squint convergens (esotropia) or squint divergens (exotropia)

**Stroma (corneal)**: layer of the cornea located between the epithelium and Descemet’s membrane; comprises 90% of the corneal thickness.

**Subcapsular (lens)**: directly behind the lens capsule, which means in the lens epithelium (ant.) or cortex (post.)

**Subepithelial (corneal)**: directly under the epithelial layer, which means in the stroma of the cornea

**Superficial chronic corneal epithelial defect (SCCED)**: see (chronic) corneal erosion

**Superior**: (also referred to as dorsal) upper region; e.g. the upper eyelid is superior to (above) the lower eyelid

**Symblepharon**: adhesions between the bulbar and tarsal conjunctiva, usually the result after a severe inflammation. Not to be mistaken for microphthalmia

**Synchysis scintillans**: liquified vitreous (syneresis) with floating white crystalline precipitates; an expression of vitreous degeneration. See also asteroid hyalosis

**Synechia**: acquired attachment between the iris and the cornea (anterior synechia) and/or lens (posterior synechia). Distinct from congenital persistent pupillary membranes

**Syneresis**: liquefaction of the vitreous and/or fluid filled cavities

**Tapetum lucidum or tapetal area**: area with reflective cell layer in the superior half of the fundus of most domestic animals, located in the choroid, but may be normally absent in some animals. Its function is to enhance light stimulation of the retina, thereby improving the animal’s ability to see in dim light conditions

**Tapetum nigrum**: see and use non-tapetal fundus or non-tapetum

**Tear film**: fluid covering the surfaces of the conjunctiva and cornea as a triple-layered film (outer oily layer, middle aqueous tear fluid layer and inner mucin layer)

**Temporal**: region of the eye located towards the ear (lateral)

**Third eyelid**: see and use nictitating membrane

**Tonometer**: instrument to estimate the intraocular pressure (IOP)

**Tonometry**: measurement of the intraocular pressure (IOP)

**Trabecular meshwork**: the part of the aqueous drainage pathway found within the ciliary cleft
Trichiasis: presumed hereditary eye disease or acquired abnormality of deviated hairs on a normal place around the lid fissure, irritating the conjunctiva, the free lid margin of the opposite lid and/or the conjunctiva and/or the globe. Predominantly on the nasal folds or on the lateral part of the superior eyelid edge

Tunica vasculosa lentis: embryonic vascular network which surrounds the lens as a continuation of the hyaloid vasculature (see persistent hyperplastic tunica vasculosa lentis (PHTVL) and hyperplastic primary vitreous (PHPV). The hyaloid system normally fully regresses between 2 to 4 weeks after birth, except a minor swine-tail-like remnant attached just below the center of the posterior lens capsule, extending into the vitreous

Uveal cyst: presumed hereditary eye disease; usually pigmented membrane spheres of various sizes, arising from posterior pigmented epithelial cells of the iris/ciliary body and which remain attached, or break free floating as pigmented spheres in the anterior chamber. When reaching maximal size, cysts tend to adhere to the endothelial surface in the center of the cornea, thus causing visual impairment. Severe cases which occur with a higher than normal incidence in the Great Dane (Deutsche Dogge) and in the Golden Retriever may lead to secondary glaucoma.

Uveal tract (uvea): pigmented, vascular and muscular layer of the eye comprising of the iris, ciliary body, and choroid

Uveitis: inflammation of the uveal tract (iris, corpus ciliare, choroid). May be caused by infectious agents or may be immune-mediated. There are syndromes of immune-mediated uveitis associated with facial skin depigmentation. With any form of uveitis, adhesions (synechia) may develop between the iris and the lens (posterior synechia) and the peripheral iris and cornea (peripheral anterior synechia). Other complications include secondary cataract and glaucoma

Uveitis pigmentary: presumed hereditary eye disease; a form of intraocular inflammation recognized in the Golden Retriever, may or may not be associated with other ocular or systemic disorders

Uveodermatologic syndrome: an immune-mediated syndrome of severe uveitis combined with dermal depigmentation (vitiligo) and hair depigmentation (poliosis). Secondary glaucoma and/or retinal detachment are frequent complications of this disease. Seen most commonly in the Akita Inu, Samoyed, Siberian Husky breeds. A similar syndrome is recognized in people and is called Vogt-Koyanagi-Harada syndrome (VKH)

Vitreous (corpus vitreum): a transparent gel-like fluid located between the lens and the retina

Vitreous (-eal) degeneration: presumed hereditary eye disease; strands of vitreous or liquefaction of the vitreous gel which may predispose to retinal detachment

Vitreous prolapse: displacement of vitreous anterior to the lens

Vitreal strands: liquified vitreous that may be observed in the vitreous and/or in the anterior chamber

Vogt-Koyanagi-Harada (VKH) syndrome: see uveodermatologic syndrome

Wiegert’s ligament: The attachment of the vitreous on the posterior aspect of the lens. This area occasionally presents as a weak circular condensed area on the posterior lens capsule, not to be confused with persistent tunica vasculosa lentis (PHTVL) and hyperplastic primary vitreous (PHPV)

Zonules: supporting fibers which attach the equator of the lens to the ciliary body
Figures of the KP-HEDs are found on the ECVO website at http://ecvo.org/hereditary-eye-diseases/images-for-panellists