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: denotesthe 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 **opticdisc**

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 characterizedby 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 **inferiotemporal 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 **homozygousphenotypes**

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 ccongenital 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 isnon-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:resumed 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: seeand 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: abnormaldeep 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 everting 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**

Lidfissure: 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 presumedhereditary eye diseasein 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**

Goniodysgenesis/ 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

Hypolasia 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 dischypoplasia: presumed hereditary eye disease; congenitalfailure 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≥ 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**

Terminology:	closed	narrow	open
Ratio A/B:	PL not visible	A < 1/3 of B	A ≥ 1/3 of B

Open: PL length (A) is equal to or more than 1/3 of B; **A ≥ 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

Modified from publication: «Correlation of morphologic features of the iridocorneal angle to intraocular pressure in Samoyeds» Ekesten B, Narfström K. Am J Vet Res, vol 52, no. 11, November 1991, p 1875-1878.

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:<u>**presumed hereditary eye disease**</u>a 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: <u>**presumed hereditary eye disease**</u> 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):<u>**presumed hereditary eye disease**</u>, 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): <u>**presumed hereditary eye disease, congenital/developmental,**</u> mostly non-progressive anomalies found in the same animal (to be specified in the certificate descriptive comment field). The anomalities 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 reflectile 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:<u>**presumed hereditary eye disease**</u>; an abnormal proliferation of melanocytes within the **uveal tract** that may cause an elevation of the intraocular pressure/**glaucoma** when an obstruction ofthe **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): <u>**presumed hereditary eye disease**</u>; 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 orpresumed 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, retrolentally 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 diseasein 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 ofthe **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 **progressiveretinal atrophy (PRA)**

Progressive Retinal Atrophy (PRA): see Retinal degeneration

Proptosis: extensive exophthalmos; See also bulbus luxation

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 dysplastic 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**, depigmentation 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 ophthalmoscopic changes 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 near-sightedness (**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 neuronesproviding 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. Differenttypes 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`smembrane**; 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 hyperplasticprimary 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: liquefied **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 thelens** 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