ECVO Manual: Chapter 5 - Definitions (2023)

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 or presumed hereditary eye disease (KP-HED). 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 iris hypoplasia**. KP-HED; Partial or complete (rare) absence of the iris. Usually it is a bilateral, hereditary anomaly.

Anophthalmos: absence of a true eyeball (rare). This term is used for the ECVO certificate if no ocular structures are visible in the orbit during the KP-HED exam. Not to be confused with surgical removal of the eye. Histopathology must be done to determine if the animal has true anophthalmos or if there are any remnants of ocular structures.

Anterior: denotes 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: see and **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 centre of the cornea and the retina, axial (adj.)

Bergmeister's papilla: a conical shaped remnant of the hyaloid artery attached to the optic nerve head.

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): KP-HED; 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: is an opacity (generally whitish) in different shapes and sizes in the lens nucleus, and/or cortex and/or capsule; it is resulting from pathologic changes in lens protein composition or disruption of lens fiber arrangement.
Clinical significance: The clinical significance is influenced by the extent, density and location of the opacity, as well as its potential to progress, which leads to scattering of incident light, reduced illumination, reduced contrast sensitivity, increased glare, degraded color vision, and loss of visual acuity and visual function.
Classification according to aetiology:

- **Primary cataracts**: all bilateral or unilateral cataracts and especially cortical cataracts are KP-HEDs (except secondary cataracts)

- **Secondary cataracts**: cataracts known to be caused by physical influences (trauma, electric, irradiation), ocular inflammation, metabolic diseases, nutritional deficiencies, age, intoxication or another KP-HED.

DNA-tests for specific breeds are available.

Ceroid lipofuscinosis (CLN): KP-HED 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 nictitating membrane, prolapsed gland

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): KP-HED congenital eye disease which is characterized by inadequate development of the choroid present at birth which is non-progressive. Most commonly identified in the Collie breed where it is a manifestation of "Collie Eye Anomaly" but can also be seen in other breeds.

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

Chorioretinal scar/lesion: a single or a few, typically **non-progressive**, lesions in the tapetum , characterized as a focal hypo- or hyper-reflective lesion often seen with a hyper-pigmented central area; or in the non-tapetum as a focal hypo- or hyper-reflective lesion

Chorioretinopathy, primary retinal disease: KP-HED in the Chinese Crested dog breed. It is characterized as bilateral, **progressive**, circumscribed areas with pigmented or light-coloured center, leading to visual impairment or blindness. Affects the RPE, the choroidal structures and then the photoreceptors secondarily.

Chorioretinopathy, secondary retinal disease with genetic predisposition: Choroidal and retinal disorder seen in several breeds. Clinical presentation varies from single, non-progressive chorioretinal scars to progression to complete retinal atrophy with choroidal fibrosis. The disorder has presumably a genetic predisposition with environmental factors as a triggering factor. The disease entity is also called "working dog retinopathy". Males are more commonly affected than females which has led to a suggestion of genetic modifiers in the X-chromosome. Parasites, mainly Toxocara Canis, are suggested as an environmental trigger. Known affected breeds: Border collie, Flat coated Retriever and Borzoi. See specific breed for details.

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: KP-HED; bilateral inflammatory disease of the cornea which usually starts as a greyish haze at the inferior or inferio-temporal 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 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 corpus ciliare, 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): KP-HED; a congenital syndrome of ocular anomalies originally described in Collie breeds (short hair, long hair, Border Collie, Bearded Collie) and Shetland sheepdog 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 or CRD) with or without coloboma, retinal detachment and intraocular haemorrhage. Vision varies with the degree to which an individual is affected and may be minimally compromised to having severe visual impairment or blindness. Breeds known to have CEA (genetic test available) are: Australian Kelpie, Australian Shepherd, Bearded Collie, Border Collie, Boykin Spaniel, Collie (short hair, long hair), Hokkaido, Lancashire Heeler, Miniature American Shepherd, Nova Scotia Duck Tolling Retriever, Shetland Sheepdog (Sheltie), Silken Windhound, Silken Windsprite.

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.

Cone degeneration (CD): KP-HED; 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): KP-HED; 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): KP-HED congenital eye disease that is non-progressive with abnormal or absent rod function. An electroretinogram (ERG) is diagnostic. Although initially described in the Briard, the term CSNB is now a misnomer for the disease in the Briard as the

condition is not stationary and also reduces cone-mediated vision. In the Briard, see and use **Retinal dystrophy/RPE 65 null mutation**

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: KP-HED; 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: KP-HED; 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: KP-HED; non-inflammatory corneal opacity (white to grey with crystalline appearance) in one or more of the corneal layers. Often it is associated with deposits of cholesterol and other lipids (or fats) within the cornea.

Corneal dystrophy, macular: KP-HED; 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 erosion: destruction or partial loss of the corneal epithelium

Corneal ulceration: local defect or excavation of the corneal stroma by destruction or loss of part of the stromal tissue extending into or through the stroma, which can result in severe scarring and corneal perforation.

Corneal epithelial defect: spontaneous chronic corneal epithelial defect (SCCED): SCCEDs in dogs are typically found in middle-aged to older dogs of all breeds. Clinically, the defects are characterized by the presence of epithelial erosion surrounded by a circumferential sheet of loosely adherent or non-adherent epithelial cells (“epithelial lip”). These epithelial defects may be present for weeks to months, particularly if left untreated or if treated inappropriately. Typical histopathological findings include loss of the corneal epithelial basement membrane and formation of a superficial, acellular, hyalinized zone in the stroma. Together, these histological abnormalities lead to delayed wound healing and poor epithelial adhesion.

Corneal edema: fluid accumulation within the cornea resulting in cloudiness

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: KP-HED; 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: KP-HED; 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 co-dominance.

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: KP-HED; 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: KP-HED; 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: KP-HED; 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: KP-HED; 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: medial strabismus (convergens); see and **use strabismus**

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

Exophthalmos due to shallow orbit: KP-HED in some brachycephalic breeds; As part of the facial and skull bone deformities, the bony cavity is not deep enough to accommodate a normal sized globe. In combination with macroblepharon this leads to an anatomically pathological exposure of the globe, recognizable by the visible sclera when the eye is directed straight ahead.

Exotropia: lateral strabismus (divergens); see and **use strabismus**

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: PK-HED; pectinate ligament fibres with either a confluent (broad) base and shortened thin insertions at the cornea or formation of thick fibres (< 5 fibres). It is part of the **Irido Corneal Angle abnormality (ICAA).**

Fissure: see Lid fissure

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: KP-HED in several dog breeds and in a few cat breeds. The disease process has a complex aetiology. 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 iridocorneal 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 Irido Corneal Angle Abnormality**

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

"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 "hemeralopia" is used for 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: KP-HED; characterized by congenital thinning and/or 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: KP-HED; characterized by congenital incomplete formation of the lens equator, sometimes called lens coloboma. See and **use lens hypoplasia**

Hypoplasia-/ optic disc hypoplasia: KP-HED; congenital failure of development of the optic nerve which causes visual deficit or blindness and abnormal pupil response in the affected eye. It 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 lacrimal punctum atresia**

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 corneal epithelial defect (SCCED)

Inferior: lower region, also referred to as ventral

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 (PL).

Iridocorneal angle (ICA) width: the width of the ICA is evaluated (using gonioscopy) by comparison of the length of the pectinate ligament PL (A) and the distance from the origin of the PL 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 PL (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 PL is severely reduced.
b) Or closed (collapsed) and PL is not visible

A = length of PL; B = distance from the origin of the PL to the anterior surface of the cornea at the transection area

Grading of ICA width:
– Open = normal
– Narrow = affected moderate
– Closed = affected severe

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.

Irido Corneal Angle abnormality (ICAA): KP-HED; characterized by an abnormal pectinate ligament that can be divided into 2 predominant types: 1. Fibrae latae, 2. Lamina and/or by a narrow or closed iridocorneal angle; Diagnosis is by gonioscopy.

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**

Iris melanoma: See and **use "uveal melanoma"**

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: KP-HED; inflammation of the cornea accompanied by multiple small areas of corneal ulceration.

Keratoconjunctivitis sicca (KCS): KP-HED 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. KCS is often referred to as "dry eye". Secondary, non-hereditary KCS may also occur, for example due to intoxication, inflammation, 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: KP-HED; 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): KP-HED; pectinate ligament fibres form plates or sheets of continuous tissue (>5 fibres), with or without flow holes. It is part of the **Irido Corneal Angle abnormality (ICAA).**

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): KP-HED; 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; it is usually congenital.

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

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

Limbus: junction between the cornea and the sclera

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

Macroblepharon: KP-HED; an exceptionally large palpebral fissure. Macroblepharon in conjunction with laxity of the lateral canthal structures may lead to lower lid ectropion in combination with lateral and medial entropion and upper lid entropion and trichiasis. This may in severe cases 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.

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 anomalies**").

Microblepharon: KP-HED; an exceptionally short palpebral fissure. Microblepharon may lead to upper lid entropion and trichiasis.

Microcornea: congenital, abnormal small diameter of the cornea

Micropapilla: KP-HED; small optic disc which is not associated with vision impairment. It 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): KP-HED; 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 (MOA): KP-HEDs; congenital/developmental, 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: congenital, abnormally small but anatomically functional globe. See and **use microphthalmos**, as other anomalies are difficult to exclude.

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. Synonyms: third eyelid, nictitans, or haw.

Nictitating membrane, eversion of the cartilage: KP-HED; 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.

Nictitating membrane, prolapsed gland: KP-HED; protrusion of the tear gland associated with the nictitating membrane. The exposed gland may become irritated. Commonly referred to as “cherry eye”.

Night blindness: KP-HED; 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: KP-HED; 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 remains attached to the pars plana in bullous retinal detachment

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): KP-HED; characterized by an abnormal pectinate ligament that can be divided into 2 predominant types: 1. Fibrae latae, 2. Lamina; Diagnosis is by gonioscopy. It is part of the **Irido Corneal Angle abnormality (ICAA).**

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): KP-HED; 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): KP-HED; 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 of fibrous tissue 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): KP-HED; in which blood vessel remnants of the embryological vascular network (pupillary membrane) in the central anterior part of the lens 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 anterior 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: see and **use Chorioretinopathy, pigmentary**

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

Pigmentary uveitis: see uveitis, pigmentary

Plasmoma/plasmacellular conjunctivitis: KP-HED; hyperplastic and hypo-(de-)pigmented margins of the nictitating membrane s due to accumulation of inflammatory (plasma) cells. It is 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

Progressive rod-cone degeneration (PRCD): KP-HED; progressive rod-cone degeneration, see Retinal degeneration

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 normally regresses up to 4-6 weeks after birth. Failure of complete regression results in persistent pupillary membrane (PPM)

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 bi-layered 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): KP-HEDs; a group of bilateral, hereditary dysplastic and/or degenerative diseases of the photoreceptors primarily, with bilaterally similar lesions which show a progressive retinal thinning over all the fundus, 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 mid-periphery 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 (rcd 1-4) 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: KP-HED; 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. Its significance to vision is unknown. When seen in puppies this condition may partially or completely resolve with maturity. 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. Although it is a congenital disease, its manifestation may not be visible until after 8 weeks of age.

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: KP-HED; 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 older publications. A DNA-test for the RPE65 null mutation in 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 in the English Cocker Spaniel.

Retinopathy: any non-specific presumed hereditary or non-hereditary **progressive** disease of the retina, seen as multi-focal pattern or representing various stages of retinal disease, usually detected by ophthalmoscopic examination.

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

Retinopathy, primary retinal disease: Disorder limited to the retina. Known affected breeds: slowly **progressive** retinopathy (SPR) in Shetland Sheepdog, Swedish Vallhund retinopathy, Basenji retinopathy, Stargardt disease in the Labrador Retriever. See specific breed for details.

Retinoscopy: an objective method to measure the error of refraction of the eye. It is 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 neurones providing for detection of shapes and motion.

Rod-cone dysplasia (rcd): KP-HED; 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 have been described. An ERG is diagnostic.

Rod dysplasia: KP-HED; 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: is 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. It is usually an acquired condition in contrast to coloboma.

Strabismus: abnormal alignment of the eyes; the condition of having a squint. Examples of strabismus include convergens (esotropia) or divergens (exotropia) of the eye

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, this means in the lens epithelium (ant.) or cortex (post.).

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

Spontaneous chronic corneal epithelial defect (SCCED): SCCEDs in dogs are typically found in middle-aged dogs of all breeds. Clinically, the defects are characterized by the presence of epithelial erosion surrounded by a circumferential sheet of loosely adherent or non-adherent epithelial cells (“epithelial lip”). These epithelial defects may be present for weeks to months, particularly if left untreated or if treated inappropriately. Typical histopathological findings include loss of the corneal epithelial basement membrane and formation of a superficial, acellular, hyalinized zone in the

stroma. Together, these histological abnormalities lead to delayed wound healing and poor epithelial adhesion.

Superior: the upper region, also referred to as dorsal; 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: KP-HED 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. It is predominantly due to facial folds or due to misalignment of the eyelids.

Tunica vasculosa lentis (TVL): embryonic vascular network which surrounds the lens; The TVL, vasa hyaloidea propria and the pupillary membrane normally fully regress between 2 to 4 weeks after birth, starting from post to anterior, except a minor pig-tail-like remnant attached just below the center of the posterior lens capsule, extending into the vitreous. Persistence of one or more portions is seen. The most common is persistence of parts of the anterior portion, the pupillary membrane, which is referred to as persistent pupillary membrane (PPM). The lack of regression of the posterior part of the TVL and/or the primary vitreous is known as persistent hyperplastic tunica vasculosa lentis (PHTVL) and/or persistent hyperplastic primary vitreous (PHPV).

Uveal cyst: KP-HED; 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 more or less pigmented spheres in the anterior or posterior chamber or occasionally in the vitreous. 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 and may lead to secondary glaucoma.

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

Uveal melanoma: KP-HED; a neoplasm caused by malignant transformation of melanocytes in the uveal tissue (iris, ciliary body, choroid). Occurs with a higher-than-normal incidence in the Labrador Retriever. Left untreated it may result in secondary glaucoma and/or metastasis. Without a histological examination, an uveal melanoma cannot be differentiated from a benign melanocytoma in every case. A distinction may become possible depending on the further development.

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: KP-HED; a form of intraocular inflammation recognized in the Golden Retriever, may or may not be associated with other ocular or systemic disorders

Uveodermatologic syndrome: KP-HED; 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 (vitreal) degeneration: KP-HED; 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

Wieger'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-HED are found on the ECVO website at:
https://www.ecvo.eu/hereditary-eye-diseases/images-for-certified-examiners-panellists.html