

EYE ABNORMALITIES PRESENT AT BIRTH

BASICS

OVERVIEW

- Single or multiple abnormalities that affect the eyeball (known as the “globe”) or the tissues surrounding the eye (known as “adnexa,” such as eyelids, third eyelid, and tear glands) that may be observed in young dogs and cats at birth or within the first 6 to 8 weeks of life
- Congenital refers to “present at birth;” congenital abnormalities may be genetic or may be caused by a problem during development of the puppy or kitten prior to birth or during birth
- The “cornea” is the clear outer layer of the front of the eye; the pupil is the circular or elliptical opening in the center of the iris of the eye; light passes through the pupil to reach the back part of the eye (known as the “retina”); the iris is the colored or pigmented part of the eye—it can be brown, blue, green, or a mixture of colors; the “lens” is the normally clear structure directly behind the iris that focuses light as it moves toward the back part of the eye (retina); the “retina” contains the light-sensitive rods and cones and other cells that convert images into signals and send messages to the brain, to allow for vision

GENETICS

- Suspected genetic background for several congenital (present at birth) eye abnormalities, some with a unknown mode of inheritance
- Remaining strands of iris tissue (the colored or pigmented part of the eye) that may extend from one part of the iris to another or from the iris to the lining of the cornea (the clear outer layer of the front of the eye) or from the iris to the lens (the normally clear structure directly behind the iris that focuses light as it moves toward the back part of the eye [retina]); condition known as “persistent pupillary membranes”—simple autosomal dominant trait in Basenjis
- Inherited embryonic developmental abnormalities of the eye (known as “persistent hyperplastic tunica vasculosa lentis” or “PHTVL” and “persistent hyperplastic primary vitreous” or “PHPV”)—autosomal dominant allele with variable expression in Doberman Pinschers; inherited in Staffordshire bull terriers
- Abnormal development of the back part of the eye (retina; condition known as “retinal dysplasia”) in English springer spaniels—simple autosomal recessive trait
- Inherited abnormal development of the eye, leading to changes in various parts of the eye in collies (known as “collie eye anomaly”)—autosomal recessive trait
- Abnormal development of the back part of the eye (retina; condition known as “retinal dysplasia”) in briards—simple autosomal recessive trait
- Abnormal development of the light-sensitive cells in the back part of the eye (retina; condition known as “photoreceptor dysplasia”) in Collies, Irish setters, and Cardigan Welsh corgis—autosomal recessive trait
- Other types of abnormal development of the light-sensitive cells in the back part of the eye (photoreceptor dysplasia) in dogs—recessively inherited
- Abnormal development of the light-sensitive cells in the back part of the eye (photoreceptor dysplasia) in Abyssinians, domestic shorthairs, and Persians—postulated autosomal dominant trait in Abyssinians and domestic shorthairs, but autosomal recessive trait in Persians

SIGNALMENT/DESCRIPTION of ANIMAL

Species

- Dogs and cats

Breed Predispositions

- See **GENETICS** and **SIGNS** for breeds commonly affected

Mean Age and Range

- Observed in young dogs and cats at birth or within the first 6 to 8 weeks of life

SIGNS/OBSERVED CHANGES in the ANIMAL

- Depend on defect
- May cause no signs of disease and may be an incidental finding in a complete eye examination
- May not affect vision or may cause severe visual loss or blindness
- Small eye (known as “microphthalmos”)—a congenitally small eye (present at birth); size of the eyeball varies, but smaller than normal; easily noted by comparing the size of the eyes; more difficult to detect if both eyes (bilateral) are involved; often associated with other genetic defects, such as opacities of the cornea (the normally clear front part of the eye); persistent strands of iris tissue (persistent pupillary membrane); cataract (opacity in the normally clear lens—the lens is the normally clear structure directly behind the iris that focuses light as it moves toward the back part of the eye [retina]); separation of the back part of the eye (retina) from the underlying, vascular part of the eyeball (retinal detachment), and abnormal development of the back part of the eye (retinal dysplasia)
- Absence of the eyeball (known as “anophthalmos”)—congenital (present at birth) lack of the eyeball or globe; often associated with other genetic defects, such as opacities of the cornea (the normally clear front part of the eye); persistent strands of iris tissue (persistent pupillary membrane); cataract (opacity in the normally clear lens—the lens is the normally

clear structure directly behind the iris that focuses light as it moves toward the back part of the eye [retina]); separation of the back part of the eye (retina) from the underlying, vascular part of the eyeball (retinal detachment), and abnormal development of the back part of the eye (retinal dysplasia)

- “Hidden” eyeball (known as “cryptophthalmos”)—a small eyeball or globe that is concealed by other defects in the tissues surrounding the eye (adnexa); often associated with other genetic defects, such as opacities of the cornea (the normally clear front part of the eye); persistent strands of iris tissue (persistent pupillary membrane); cataract (opacity in the normally clear lens—the lens is the normally clear structure directly behind the iris that focuses light as it moves toward the back part of the eye [retina]); separation of the back part of the eye (retina) from the underlying, vascular part of the eyeball (retinal detachment), and abnormal development of the back part of the eye (retinal dysplasia)
- Absence of the eyelid (known as “eyelid agenesis”) or failure of the eyelid to form properly (known as a “coloboma”)—often result in congenitally open eyelids; considered hereditary in Burmese cats; usually affect the upper eyelid; may note squinting or spasmodic blinking (known as “blepharospasm”) and tearing (known as “epiphora”)
- Dermoids—congenital (present at birth), tumor-like, islands of displaced skin tissue involving either eyelids, conjunctiva, or cornea; sometimes affect more than one structure; may note squinting or spasmodic blinking (blepharospasm) and tearing epiphora)
- Congenital (present at birth) absence of central canal in the tear drainage system, through which the tears drain (known as “atresia”) and lack of normal openings on the eyelids into the tear drainage system (known as “imperforate puncta”)—affects cats and dogs; imperforate puncta: common in several dog breeds (such as cocker spaniels); results in tear streaking on the face, just below the corner of the eye closest to the nose and on the side of the nose; usually not associated with other eye abnormalities
- Congenital (present at birth) dry eye (known as “keratoconjunctivitis sicca” of “KCS”)—may occur sporadically in any dog or cat breed; may be hereditary in Yorkshire terriers; usually only one eye (unilateral) is affected; affected eye often appears smaller than the normal eye; results in a thick mucoid discharge from a red and irritated eye
- Remaining strands of iris tissue (the colored or pigmented part of the eye) that may extend from one part of the iris to another or from the iris to the lining of the cornea (the clear outer layer of the front of the eye) or from the iris to the lens (the normally clear structure directly behind the iris that focuses light as it moves toward the back part of the eye [retina]); may coexist with a variety of other eye defects (such as iris defects and cataracts); affects any species; recorded in numerous dog breeds; shown to be hereditary in Basenjis
- Iris cysts—circular, pigmented or nonpigmented ball-like structures that float freely in the front part of the eye, between the cornea and the iris (known as the “anterior chamber”) or are attached to the iris or to the lining of the cornea
- Congenital (present at birth) increased pressure within the eye (glaucoma) with secondary enlargement of the eye (known as “buphthalmos”)—affects dogs and cats; rare; often note increased tearing and an enlarged, red, and painful eye
- Congenital (present at birth) abnormalities of the pupil (the center of the iris)—more than one pupil (known as “polycoria”); absence of a pupil (known as “acorea”); lack of the iris (known as “aniridia”); abnormally shaped pupil (known as “dyscoria”)
- Congenital (present at birth) cataract (opacity in the normally clear lens—the lens is the normally clear structure directly behind the iris that focuses light as it moves toward the back part of the eye [retina])—primary, often inherited (such as in the Cavalier King Charles spaniel) or secondary to other developmental defects; often associated with other congenital abnormalities of the lens
- Abnormal development of the back part of the eye (retina; condition known as “retinal dysplasia”)—affects a variety of dog breeds; occurs sporadically in cats; abnormalities seen with an ophthalmoscope range from localized folds of retinal tissue to localized or complete separation of the back part of the eye (retina) from the underlying, vascular part of the eyeball (retinal detachment)
- Abnormal development of the light-sensitive cells (rods and cone) in the back part of the eye (retina; conditions known as “rod and cone dysplasias”) of dogs—rod and cone dysplasia affects Irish Setters and collies; rod dysplasia and early rod degeneration affect the Norwegian elkhound; cone degeneration affects Alaskan malamutes
- Abnormal development of the light-sensitive cells (rods and cone) in the back part of the eye (retina; conditions known as “rod and cone dysplasias”) of cats—affects Persians, Abyssinians, and American mixed-breeds; may show dilated pupils at 2 to 3 weeks of age and short, rapid movements of the eyeball (known as “nystagmus”) at 4 to 5 weeks of age, with signs of deterioration of the back part of the eye (retina) seen with an ophthalmoscope at 8 weeks of age, and night and day blindness some weeks later
- Abnormal development of the back part of the eye (retinal dysplasia) in briards—causes night blindness, short, rapid movements of the eyeball (nystagmus), and abnormally large pupils
- Separation of the back part of the eye (retina) from the underlying, vascular part of the eyeball (retinal detachment”)—seen in conjunction with inherited eye diseases (such as abnormal development of the back part of the eye [retinal dysplasia]); mainly found in Labrador retrievers, Bedlington terriers, and Sealyham terriers and with collie eye anomaly; may note a widely dilated pupil that is unresponsive to light stimuli; retinal detachment may be seen in the pupil as a funnel-shaped “curtain;” complete detachment results in blindness
- Lack of development of the optic nerve (the nerve that runs from the back of the eye to the brain; condition known as “optic nerve hypoplasia”)—occurs sporadically as a congenital (present at birth) eye defect in dogs and cats; believed to have a hereditary background in miniature and toy poodles; often results in blindness

CAUSES

- Genetic
- Spontaneous malformations of the eyeball and/or surrounding tissues

- Infections and inflammations during pregnancy—congenital (present at birth) cataract (opacity in the normally clear lens—the lens is the normally clear structure directly behind the iris that focuses light as it moves toward the back part of the eye [retina]); medical syndromes with multiple birth defects
- Toxicity during pregnancy
- Nutritional deficiencies

TREATMENT

HEALTH CARE

- Patients may be referred to a veterinary eye doctor (ophthalmologist) for a complete evaluation, especially if the eye abnormality affects the internal structures of the eye or vision is impaired
- No medical treatment for most congenital (present at birth) abnormalities of the eye, except possibly symptomatic treatment (such as for treatment of congenital “dry eye” [KCS])
- Inhibit self-mutilation after surgical procedures by using an Elizabethan collar or by directly bandaging the paws or eye

ACTIVITY

- Usually no change in activity is necessary

SURGERY

- Depends on specific abnormality of the eye or surrounding tissues (adnexa)
- If possible, wait to have surgery until the patient has reached adult size to avoid “overcorrecting” the defect; however, the surgery may be necessary prior to adulthood, if the abnormalities are too severe and/or may lead to complications
- Abnormalities of the tissues surrounding the eye (adnexa), such as dermoids or severe malformations of the eyelids—surgery as soon as possible
- Imperforate puncta (lack of normal openings on the eyelids into the tear drainage system)—surgically correct as soon as anesthesia is safe
- Congenital (present at birth) “dry eye” (KCS)—surgically move the duct from the parotid salivary gland to the eye (procedure known as a “parotid duct transposition”); the saliva then acts as “tears” in the eye
- Surgical removal of cataracts—congenital (present at birth) opacities in the lens (cataracts) may be associated with other eye abnormalities that may cause surgical complications
- Congenital (present at birth) increased pressure within the eye (glaucoma)—surgical removal of the eye (known as “enucleation”) or surgical removal of the contents of the eyeball with insertion of a silicone ball or prosthesis (known as an “intrascleral prosthesis”) to allow the eye to have a fairly normal appearance; technique relieves pain in the eye, but the eye is still blind—usually treatments of choice; consider euthanasia if both eyes are involved (bilateral glaucoma)

MEDICATIONS

Medications presented in this section are intended to provide general information about possible treatment. The treatment for a particular condition may evolve as medical advances are made; therefore, the medications should not be considered as all inclusive.

- Congenital (present at birth) “dry eye” (KCS)—tear substitutes (Tears Naturale® and Viscotears®), possibly in combination with antibiotics (drops or gel); cyclosporine ophthalmic (Optimmune®) ointment
- Congenital (present at birth) opacities in the lens (cataracts)—medications to dilate the pupil may be used to increase visual capability, if the cataract only involves the center of the lens

FOLLOW-UP CARE

PATIENT MONITORING

- Depends on defect
- Congenital (present at birth) “dry eye” (KCS)—requires frequent monitoring of tear production and the status of the external eye structures
- Congenital (present at birth) opacities in the lens (cataracts) and severe persistent hyperplastic tunica vasculosa lentis (PHTVL) and persistent hyperplastic primary vitreous (PHPV)—regular checkups, usually on a 6-month basis, to monitor possible progression of abnormalities
- Large defects of the back of the eye (retina) and abnormal development of the back part of the eye (retinal dysplasia)—yearly checkups to monitor possible complete separation of the back part of the eye (retina) from the underlying, vascular part of the eyeball (retinal detachment)

PREVENTIONS AND AVOIDANCE

- Depend on type and severity of defect

- Restrict breeding of affected animals and of known carriers of documented hereditary defects; DNA-based tests are available for many inherited defects to identify carriers

POSSIBLE COMPLICATIONS

- Depend on defect
- Untreated eyelid abnormalities (such as lack of formation of the eyelid [agenesis] or masses composed of “displaced” skin, frequently with long hair [dermoids]) and congenital (present at birth) “dry eye” (KCS)—recurrent problems with inflammation of the moist tissues surrounding the eye (known as “conjunctivitis”) and inflammation of the cornea (known as “keratitis”)
- Congenital (present at birth) increased pressure within the eye (glaucoma)—painful, blind eye in conjunction with enlargement of the eyeball (buphthalmos); often a dry, pigmented cornea
- Abnormalities of the optic nerve on the back of the eye (retina)—may cause separation of the back part of the eye (retina) from the underlying, vascular part of the eyeball (retinal detachment)
- Separation of the back part of the eye (retina) from the underlying, vascular part of the eyeball (retinal detachment)—may cause bleeding within the eye, especially within the vitreous; the “vitreous” is the clear, gel-like material that fills the back part of the eye (between the lens and the retina)

EXPECTED COURSE AND PROGNOSIS

- Depend on defect and type of medical and/or surgical treatment provided
- Abnormalities of the tissues around the eye (adnexa), such as the eyelids—good prognosis with surgical treatment
- Congenital (present at birth) “dry eye” (KCS)—rather poor prognosis with medical treatment only; somewhat better prognosis with surgical treatment (parotid duct transposition)
- Congenital (present at birth) opacities in the lens (cataracts)—usually good prognosis with surgical treatment, if other structures within the eye are normal

KEY POINTS

- Congenital (present at birth) eye abnormalities can affect vision; they may progress; and complications can develop
- Blind animals may need direct supervision when exposed to a potentially hazardous environment
- Congenital (present at birth) “dry eye” (KCS)—consider medical treatment versus surgical intervention; if the eye is medicated on a regular basis, the patient may do fine, especially if some tear production exists; if client cannot medicate on a regular basis, surgery (parotid duct transposition) should be considered

