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) observed in young dogs and cats at birth or within the first 6–8 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

- Known, suspected, or unknown mode of inheritance for several congenital (present at birth) eye abnormalities
- 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”—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)—dominant trait 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—recessive trait
- Inherited abnormal development of the eye, leading to changes in various parts of the eye in collies (known as “collie eye anomaly”)—recessive trait
- Abnormal development of the back part of the eye (retina; condition known as “retinal dysplasia”) in briards—recessive trait
- Abnormal development of the light-sensitive cells in the back part of the eye (retina; condition known as “rod-
cone dysplasia”) in collies, Irish setters, Cardigan Welsh corgis, and Sloughi dogs—recessive trait
• Deterioration of the light-sensitive cells in the back part of the eye (retina; condition known as “cone-rod dystrophy”) in pit bull terrier and long- and shorthaired dachshunds—recessive trait
• Abnormal development of the light-sensitive cells in the back part of the eye (photoreceptor dysplasia) in Abyssinians, domestic shorthairs, and Persians—dominant trait in Abyssinians and domestic shorthairs, but recessive trait in Persians

**SIGNALMENT/DESCRIPTION OF PET**

**Species**
- Dogs
- Cats

**Breed Predilections**
- See “Genetics” and “Signs/Observed Changes in the Pet” for breeds commonly affected

**Mean Age and Range**
- Observed in young dogs and cats at birth or within the first 6–8 weeks of life

**SIGNS/OBSERVED CHANGES IN THE PET**

- Depend on defect
- May cause no signs of disease; often 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; 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; associated with other genetic defects “Hidden” eyeball (known as “cryptophthalmos”)—a small eyeball or globe concealed by other defects in the tissues surrounding the eye (adnexa) Absence of the eyelid (known as “eyelid agenesis”) or failure of the eyelid to form properly (known as “coloboma”)—may result in congenitally open eyelids; affect the upper eyelid; may cause squinting or spasmodic blinking (known as “blepharospasm”) and tearing (known as “epiphora”)”
- Dermoids—islands of displaced skin tissue involving either eyelids, conjunctiva, or cornea; may cause squinting or spasmodic blinking (blepharospasm) and tearing epiphora
- 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”)—common in dog breeds; results in tear streaking on the face, just below the corner of the eye closest to 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; usually only one eye (unilateral) is affected; affected eye 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 other eye defects; affects any species; recorded in numerous dog breeds, especially basenjis
- Iris cysts—pigmented or nonpigmented ball-like structures that float freely in eye; can be attached to the iris or to the lining of the cornea (known as the “corneal endothelium”)
- Congenital (present at birth) increased pressure within the eye (glaucoma)—affects dogs and cats; rare; note increased tearing and an enlarged, red, and painful eye
- 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 or secondary to other developmental defects; associated with other abnormalities of the lens

Abnormal development of the back part of the eye (retina; condition known as “retinal dysplasia”)—affects several 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 cones) in the back part of the eye (retina; conditions known as “rod-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

Deterioration of the light-sensitive cells (rods and cones) in the back part of the eye (retina; condition is cone-rod dystrophy) in pit bull terriers—enlarged or dilated pupils (the circular or elliptical opening in the centers of the iris of the eye) and visual difficulties at 6–7 weeks of age; in longhaired dachshunds—highly variable signs; from early day blindness to slight visual impairment in daylight at 6–7 weeks of age; in shorthaired dachshunds—visual impairment in bright light at 6–7 weeks of age; many puppies have markedly constricted pupils (known as “miotic pupils”) in the light

Abnormal development of the light-sensitive cells (rods and cone) in the back part of the eye (retina; conditions known as “rod-cone dysplasias”) of cats—affects Persians and Abyssinians; have dilated pupils at 2–3 weeks of age and short, rapid movements of the eyeball (known as “nystagmus”) at 4–5 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 later night and day blindness

Abnormal development of the back part of the eye (retinal dysplasia) in briards—congenital (present at birth) night blindness, short, rapid movements of the eyeball (nystagmus), and enlarged or dilated pupils

Separation of the back part of the eye (retina) from the underlying, vascular part of the eyeball (retinal detachment)—seen in conjunction with other inherited eye diseases (such as abnormal development of the back part of the eye [retinal dysplasia]); found in Labrador retrievers, Bedlington terriers, and Sealyham terriers and with collie eye anomaly; often observed with other eye defects; signs include widely dilated pupils that are unresponsive to light stimuli; 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; has a hereditary background in miniature and toy poodles; may result in blindness

CAUSES
- Genetic
- Spontaneous malformations of the eyeball and/or surrounding tissues
- Infections and inflammations during pregnancy
- Toxicity during pregnancy
- Nutritional deficiencies

RISK FACTORS
- Breeding dogs or cats that are affected by a genetic disorder or have the genetic background for an inherited disease

Treatment

HEALTH CARE
- Pets 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 symptomatic treatment (such as for treatment of congenital “dry eye” [KCS]) or eye surgery
- Congenital (present at birth) “dry eye” (keratoconjunctivitis sicca or KCS)—remove any discharge from around the eye, flush eye with saline intended for use in the eye
- Inhibit self-mutilation after surgical procedures by using an Elizabethan collar or by directly bandaging the paws
or eye

- Blind pets should not be walked outdoors without a leash; they should be supervised carefully

**ACTIVITY**

- Usually no change in activity is necessary; however, visually impaired or blind pets need adequate exercise

**DIET**

- Vitamins, antioxidants, and omega-3 fatty acids should be provided regularly, especially for pets with cone-rod deterioration; ask your pet’s veterinarian for recommendations for providing these nutrients to your pet

**SURGERY**

- Depends on abnormality of the eye or surrounding tissues (adnexa)
- If possible, wait to have surgery until the pet has reached adult size to avoid “overcorrecting” the defect; however, 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—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, possibly in combination with antibiotics; cyclosporine ophthalmic 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)—repeated monitoring of tear production and 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 progression of abnormalities
- Large defects of the back of the eye (retina) and abnormal development of the back part of the eye (retinal dysplasia)—regular checkups to monitor possible 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 pets and of known carriers of documented hereditary defects; DNA-based tests are available for some rod, cone, and retinal defects to identify carriers
POSSIBLE COMPLICATIONS

- Depend on defect
- Cataract surgery can result in increased pressure within the eye (glaucoma), separation of the retina from the underlying tissues (retinal detachment) and scarring of the cornea (clear part of the front of the eye) in puppies and kittens; surgery seldom recommended before 8–12 weeks of age
- Severe abnormalities of the external parts of the eye may make defects difficult to correct completely
- Inflammation of the cornea (known as “keratitis”) and skin (known as “dermatitis”) may occur in pets with congenital (present at birth) “dry eye” (keratoconjunctivitis sicca or KCS), due to minerals being deposited on the cornea or skin from the saliva following surgical treatment for “dry eye” (parotid duct transposition)

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; most abnormalities affecting the adnexa can be corrected in puppies and kittens
- Congenital (present at birth) “dry eye” (KCS)—good prognosis with surgical treatment (parotid duct transposition)
- Glaucoma (increased pressure within the eye)—prognosis is poor

Key Points

- Congenital (present at birth) eye abnormalities can affect vision; they may progress; and complications can develop
- Blind pets should not be walked outdoors without a leash
- Blind pets 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 pet may do fine, especially if some tear production exists; if client cannot medicate on a regular basis, surgery (parotid duct transposition) should be considered