

Hereditary eye disease

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Revised by Peter G C Bedford,
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Hereditary eye disease

Clinical examination for inherited eye disease

The main purpose of the Canine Health Schemes (CHS) Eye Examination Scheme, run by British Veterinary Association (BVA), The Kennel Club (KC) and International Sheep Dog Society (ISDS), is to ensure that there is no clinical evidence of hereditary eye disease in dogs that are to be used for breeding. Additionally, the Scheme helps to identify breed-related problems which may be inherited, especially if they have welfare implications for the affected dog. An Eye Examination Certificate is issued which records the inherited eye disease status relevant to the breed being examined as either 'clinically unaffected' or 'clinically affected', together with any additional comments about other clinical findings. In breeds in which primary glaucoma is recognised, the basic clinical examination is supplemented by examination of the drainage angle (gonioscopy) for primary closed angle glaucoma, or assessment of the intraocular fluid pressure (tonometry) for primary open angle glaucoma. In addition, the clinical examination of puppies when they are still part of a litter (litter screening) is used to identify signs of congenital, or early onset, hereditary eye disease. When new disease is considered to be potentially inherited, inclusion in the Scheme is based on scientific evidence which includes clinical prevalence of at least 1% over a minimum three-year period and/or the peer-reviewed scientific literature.

Since its inception in the 1960s, the Scheme has been expanded to include assessment of the adjacent (adnexal) structures such as the eyelids. The result of this expansion is that certification has important subsidiary benefits, notably, recording abnormalities of potential or actual clinical significance, whatever their origin. Examples are provided later.

It is clearly sensible for all dogs, both pure-bred and cross-bred, to be examined under the Scheme prior to breeding, as this is the simplest way of identifying breed-related and potentially inherited problems. Advice on the frequency of re-testing is provided each time the dog is examined under the Scheme. Examination and certification of older dogs, usually those no longer used for breeding, should be regarded as essential, as longitudinal information collected over time provides owners and breeders with information needed to make informed breeding decisions. A reduced fee provides a financial incentive for certification of dogs aged eight years and older. Examination of the older dog is recommended for a number of reasons:

- Collecting longitudinal information. A longitudinal study is an observational research method in which data is gathered for the same dogs over a period of time.
- Ensuring that the dog remains free from the hereditary eye diseases listed for the breed. A number of inherited eye diseases may only be detected later in life (for example, various types of hereditary cataract and some forms of progressive retinal atrophy).

- Indicating whether late onset, potentially hereditary diseases, are emerging in older animals.
- Identifying both age-related eye diseases and generalised diseases with ocular manifestations, some of which may need treatment.

Further information is provided in the leaflet *Why should we check the eyes of older dogs?* (available from www.bva.co.uk/chs)

The inherited eye diseases currently certified under the Eye Examination Scheme are reviewed in this article, together with examples of some other conditions of the eye and adnexa which may be inherited, as well as other examples of non-inherited disease. Potentially inherited and non-inherited disease is currently recorded in the middle section of the certificate

Inherited eye disease status is recorded in the bottom section of the Certificate as congenital (present from birth) or neonatal and non-congenital (acquired later in life). This simple classification is not entirely satisfactory as the eyes of puppies cannot be examined until the puppy is at least 5 to 6 weeks old. There is thus a presumption that abnormalities viewed at this stage are of congenital origin, whereas it is possible that some may actually be neonatal rather than congenital. In addition, because the eye is immature at birth, it is possible that some developmental diseases may not always be apparent at litter screening (for example retinal dysplasia) and others may become less obvious because of postnatal maturation

The normal eye

Fig. 1

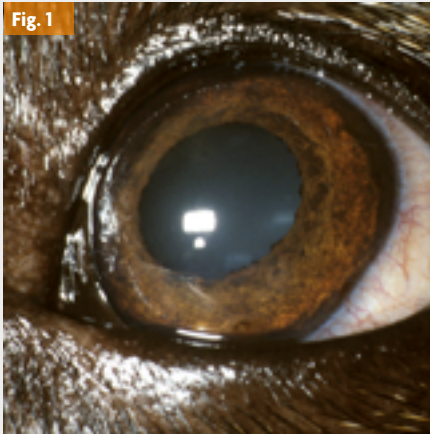


Fig. 1: Normal adult eye of a Border Collie with a pigmented iris. **Fig. 2:** Ocular fundus of that eye, showing the tapetal fundus (yellow) dorsally and heavily pigmented non-tapetal fundus ventrally. The tapetum is a reflective layer of cells beneath the central retina. A variety of colours can be seen.

Fig. 2



Fig. 3



Fig. 3: Normal adult eye of a Crossbred dog with variations of pigmentation (heterochromia) in different sectors of the iris. **Fig. 4:** Ocular fundus of that eye. Note that there is less pigment ventrally, corresponding with the area of reduced pigmentation in the iris.

Fig. 4



Fig. 5

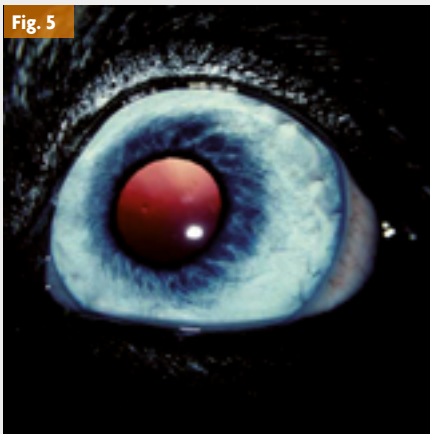


Fig. 5: Subalbinotic eye of a normal adult Border Collie. **Fig. 6:** Ocular fundus of the subalbinotic eye. Both retinal (dark red) and choroidal (pale red) blood vessels are visible. There is no tapetum in this eye.

Fig. 6

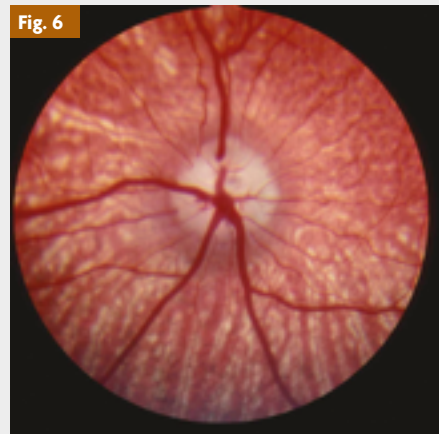


Fig. 7



Fig. 7: The eyes of newborn puppies are not fully developed at birth and the tapetum has not yet formed in this five-week-old puppy. **Fig. 8:** Normal ocular fundus of the eye of the Border Collie puppy pictured on the left.

Fig. 8



(for example, the choroidal hypoplasia part of Collie eye anomaly). On the Certificate, the diseases which are identifiable during the neonatal stage are described as congenital/neonatal.

In summary, clinical examination for certification under the Eye Examination Scheme includes:

1. Litters of puppies in breeds with congenital/neonatal inherited eye disease (using the Litter Screening form).
2. All pure-bred or cross-bred dogs before they are used for breeding and when they are subsequently bred from using the Eye Examination Certificate. For dogs that are bred from year on year, annual re-examination is required throughout the dog's breeding life.
3. Breeds in which primary glaucoma is recognised require additional assessment as part of the certification using the Eye Examination Certificate. The basic clinical examination should be supplemented by gonioscopy, to assess the drainage angle in breeds susceptible to primary closed angle/angle closure glaucoma and tonometry, to measure the intraocular fluid pressure in breeds susceptible to primary open angle glaucoma. Both these procedures will need to be repeated in later life.
4. Dogs of eight years of age or older are examined, in order to check ocular and general health, to provide valuable longitudinal data and, to ensure that later onset inherited diseases are recorded accurately. The Eye Examination Certificate is also used for this purpose.

Inherited eye diseases certified under the Eye Examination Scheme

Glaucoma

Glaucoma is the term used to describe the effects of a sustained pathological increase in the intraocular fluid pressure (IOP). Within the eye the cornea and lens are sustained by a fluid secretion known as the aqueous humour (the aqueous). In the normal eye the rate of aqueous formation and the rate of aqueous outflow are in equilibrium, and the normal canine IOP measured with an applanation or rebound electronic tonometer is within a 10–25mmHg range. In glaucoma, the clinical features seen are the result of the structural damage caused by the elevated IOP. In particular, it is damage to the retinal ganglion cells and axons of the optic nerve which is the most significant feature in sight loss. Once the process of retinal ganglion cell and optic nerve degeneration has begun, the most that therapy can achieve is a slowing down in the loss of sight.

Glaucoma is not a single disease entity, but rather a pathological process with a number of possible causes and a final common pathway. Two broad categories of glaucoma are recognised; primary (inherited) and secondary (non-inherited). In primary glaucoma there is no antecedent intraocular disease and, although the aetiology is complex, all are due to the impairment or cessation of aqueous outflow from the anterior chamber of the eye. Aqueous outflow is through the iridocorneal (drainage) angle and, in the dog, the angle is extended posteriorly into the ciliary body as the ciliary cleft. It is within the ciliary cleft that the trabecular meshwork is found and the canine equivalent of the primate canal of Schlemm,

the aqueous plexus, is situated in the scleral tissues which form the outer wall of the cleft. In cases of primary glaucoma, defect of the iridocorneal angle and the structures associated with the ciliary cleft is responsible for the inadequate drainage, leading to an increase in the IOP. The secondary glaucomas are associated with antecedent eye disease such as uveitis, primary lens luxation, trauma and neoplasia.

Classification of primary glaucoma

Currently, two types of primary glaucoma may be distinguished; primary closed angle glaucoma (PCAG) (or primary angle closure glaucoma [PACG]) and primary open-angle glaucoma (POAG) (see the leaflet *Primary glaucoma* — available from www.bva.co.uk/chs). The nomenclature has been 'borrowed' from human medicine and, although acceptable, does not accurately describe the situation. In canine glaucoma, these terms are used to describe the appearance of the entrance to the ciliary cleft; in PCAG the cleft entrance is closed and in POAG the cleft entrance is initially open.

- **Primary closed angle glaucoma (PCAG)/primary angle closure glaucoma (PACG):** In the normal dog, the ciliary cleft entrance is between 1.5 and 2mm in width and spanned by a number of iris root processes or fibres, collectively referred to as the pectinate ligament. Dogs which develop PCAG demonstrate a congenital predisposition to the disease, in that the pectinate

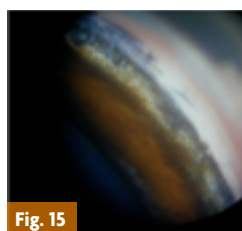
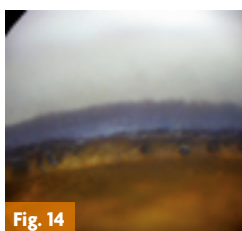
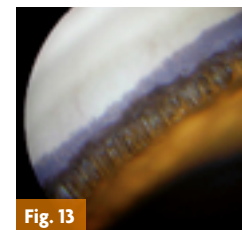
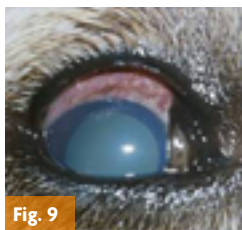


Fig. 9: Acute closed angle glaucoma. **Fig. 10:** Gonioscopy: a Barkan gonioslens in situ, a column of saline within the silicone tubing creates a negative pressure to keep the lens in place. **Fig. 11:** Gonioscopy: a Koeppel gonioslens in situ, coupling gel keeps the lens in place. **Fig. 12:** Normal drainage angle of a Siberian Husky. The drainage angle is of normal width and is spanned by the pectinate ligament. In this poorly pigmented eye, the white band of the scleral shelf is clearly distinguished. There is great variation in the number, width, pigmentation and distribution of the fibres that comprise the pectinate ligament in different breeds, but the width of the normal drainage angle is not subject to such variation. **Fig. 13:** Normal drainage angle of a Flat Coated Retriever. The width of the drainage angle is normal and the fibres of the pectinate ligament are clearly defined. This eye is more heavily pigmented than the one pictured on the left and the scleral shelf is obscured by pigment. **Fig. 14:** Goniodysgenesis in a Flat Coated Retriever. There is extensive pectinate ligament abnormality and sheets of mesenchymal tissue occlude the majority of the drainage angle. Aqueous drainage is via a limited number of 'flow holes'. The drainage angle is slightly narrowed and normal pigment obscures the scleral shelf. The eye was normotensive at the time of examination (intraocular pressure of 18 mmHg), despite the compromised drainage angle. **Fig. 15:** Welsh Springer Spaniel – goniodysgenesis. Here the pectinate ligament is seen as a continuous band of tissue occluding the entrance to the ciliary cleft and the angle is narrowed.

ligament is abnormal, with the fibres being thicker than normal (fibrae latae) or aggregated into sheets of tissue (laminae). Pectinate ligament abnormality (PLA), with or without narrowing of the angle, is referred to as *goniodysgenesis*. To assist owners and breeders and as part of data collection for goniodysgenesis, a simple grading system is used to assess the degree of PLA present and to help monitor any changes to the angle over time (see *Primary glaucoma* — available from www.bva.co.uk/chs).

The age at which glaucoma develops tends to vary with breed but, in most, the disease is one of middle age and usually acute in onset. In addition to raised IOP the common clinical signs of PCAG include pain, episcleral congestion, corneal oedema and a dilated non-responsive pupil. As the canine drainage angle cannot be viewed directly, a special gonioslens is needed to assess the pectinate ligament

and angle width, with the Barkan and Koeppel direct gonioslenses being used most commonly. By facilitating an assessment of the degree of goniodysgenesis, gonioscopy may predict the possibility of glaucoma before it makes its clinical appearance and the grading scheme helps inform breeding decisions. Routine gonioscopy can be performed from five to six months of age in most breeds and should be repeated, as PLA progression over time has been reported in a number of breeds. These include the Flat Coated Retriever, Basset Hound, Dandie Dinmont Terrier and Welsh Springer Spaniel, but gonioscopy should be performed at approximately 1, 4 and 7–8 years of age for any breed at risk.

The PCAG/PACG-affected breeds in the UK

Basset Hound
Border Collie
Dandie Dinmont Terrier
Hungarian Vizsla
Japanese Shiba Inu
Leonberger
Retriever (Flat Coated)
Retriever (Golden)
Siberian Husky
Spaniel (American Cocker)
Spaniel (Cocker)
Spaniel (English Springer)
Spaniel (Welsh Springer)
Spanish Water Dog

The precise mode of inheritance has not been determined for the PCAG affected breeds, but clear breed and line predisposition indicate a genetically determined cause or causes, albeit with a likely complex mode of inheritance.

- **Primary open angle glaucoma:** Primary open angle glaucoma (POAG) presents both dog breeder and clinician alike with real problems in its early diagnosis, for the usual extant clinical features of glaucoma are not present. It is silent in onset, with either vision impairment or globe enlargement usually being the first abnormalities reported by the owner. Goniodysgenesis is not a feature of this type of glaucoma and there are no other predisposing features that can be used to forecast the advent of the disease. It is inherited as a simple recessive trait and has been described extensively in the Beagle and the Petit Basset Griffon Vendeen (PBGV). In the Beagle the causal mutation has been identified in the *ADAMTS 10* gene and for the PBGV the mutation is in the *ADAMTS 17* gene. Fortunately, DNA tests have been developed for both these breeds. POAG has also been reported in other breeds, notably the Norwegian Elkhound, the Basset Hound and the Basset Fauve de Bretagne and DNA tests are also available for these breeds.

In the PBGV the earliest clinical presentation is either lens instability (phacodonesis), lens subluxation in a normotensive eye, or a moderately raised IOP, or actual globe enlargement with possible lens subluxation/

luxation and vision impairment. The disease affects most dogs from 3 to 6 years of age, but a later onset is possible. Only when the IOP appreciably increases, variable degrees of episcleral congestion and mild corneal oedema may be seen. The pupil may be partially dilated and with the passage of time the pupillary light reflex (PLR) is eventually impaired or lost. Signs of ocular pain are subtle and affected dogs may be lethargic and may sleep more. With progression, globe enlargement occurs, but it is only in advanced disease that

The POAG-affected breeds in the UK

Basset Fauve de Bretagne
Basset Hound
Petit Basset Griffon Vendeen
Shar Pei



Fig. 16



Fig. 17

Fig. 16: Tonopen in use. **Fig. 17:** Primary open angle glaucoma – the globe is enlarged and secondary lens luxation has occurred.

deformation (cupping) of the optic disc can be seen. Retinal degeneration characterised by increased tapetal reflectivity and blood vessel attenuation are also late changes in POAG. Lens subluxation may occur even before there is a rise in IOP, but total lens luxation would appear to be unusual unless there is gross globe enlargement. The elevation of IOP is moderate initially, with pressures between 30 and 40 mmHg being routinely recorded.

The POAG-affected breeds in the UK

Basset Hound
Basset Fauve de Bretagne
Petit Basset Griffon Vendeen
Shar Pei

In terms of the CHS Eye Examination Scheme as applied to POAG, gonioscopy is not necessary, for angle closure occurs only in late disease where there is globe enlargement, but tonometry can prove helpful

where early disease is present. Annual examination in dogs of 3 to 9 years of age is advisable. Although lens subluxation is common before and after the rise in IOP, POAG should not be confused with primary lens luxation (see below).

Persistent hyperplastic primary vitreous

The embryonic lens is supplied in part with nutrients by the hyaloid artery (which grows from the optic stalk to reach the posterior lens surface at about day 25 of gestation) and the tunica vasculosa lentis develops. Regression of vascular supply starts at about day 45 and is complete some two to four weeks after birth. A persistence of these vessels and proliferation of associated mesodermal elements of the posterior part of the TVL results in fibrovascular plaque formation on the posterior lens capsule. This plaque appears as a dense yellow/white opacity with multiple pigment foci and blood vessels which may be visible within the plaque and at its periphery. Other features of persistent hyperplastic primary vitreous (PHPV) include persistent capsulopupillary vessels (iridohyaloid vessels), lens colobomas, posterior lenticonus, intralenticular and retrolental haemorrhage, secondary cataract, persistence of the hyaloid artery, Bergmeister's papilla (from remnants of the glial sheath around hyaloid vessels on the optic disc), and retinal dysplasia.

The PHPV-affected breeds in the UK

Dobermann
Staffordshire Bull Terrier

Currently, the Dobermann and Staffordshire Bull Terrier are certified for PHPV under the Eye Examination Scheme. The mode of inheritance is complex and

to date no mutations have been identified that play a role in the development of this disease, although the genetic data available suggest an autosomal dominant gene with variable or incomplete penetrance.

Significance

Severe lesions cause marked visual loss or blindness. Yellow/brown focal dots on the posterior lens capsule, the mildest form of PHPV, have no discernible effect on vision. Small areas of retrolental plaque formation, which may not involve the lens periphery, allow some vision to be present. More extensive plaques and cataract, or other lens abnormalities, usually cause severe visual impairment or blindness. The Staffordshire Bull Terrier suffers less from the posterior lens capsule deformities, but if retinal dysplasia is present, it has more widespread retinal folds and rosettes than the Dobermann. PHPV is not common in either breed in the UK, but represents a serious congenital inherited problem in some affected dogs. Surgical treatment of those cases with visual problems is fraught with difficulty and there is a high risk of postoperative complications.

Retinal dysplasia

The term retinal dysplasia (RD) embraces a number of congenital or neonatal conditions resulting from atypical differentiation of the retina during embryonic life. In addition to genetically determined hereditary retinal dysplasia, a wide variety of extraneous insults to the developing retina may be acquired, non-inherited RD (for example, irradiation and infectious agents such as canine herpes virus) may cause acquired, non-inherited, retinal dysplasia. Defective retinal development results both in extremely varied clinical and microscopic appearances so that, for example, folds, ridges, rosettes, geographic abnormalities and localised detachments are all possible manifestations of multifocal retinal dysplasia (MRD); whereas total retinal dysplasia (TRD) is most commonly associated with non-attachment or complete detachment of the retina.

Classification

- **Multifocal retinal dysplasia (MRD):** Linear folding of the sensory retina and the formation of rosettes composed of variable numbers of neuroretinal cells are the histological characteristics of MRD. Typically, the lesions range from vermiform grey streaks, dots and circles to multiple focal sites of tapetal hyperreflectivity, which may or may not be associated with hypertrophy of the retinal pigment epithelium. Circular shaped areas of dysplastic retina may also be encountered and the term geographic retinal dysplasia (GRD) is used to describe these lesions. In most cases, the areas of GRD are seen in the tapetal fundus dorsal to the optic disc and, rather surprisingly, are usually unilateral. In the English Springer Spaniel, dysplastic changes occur in the developing sensory retina at 45 to 50 days of gestation. The other breeds currently certified under the Eye Examination Scheme for MRD are the Cavalier King Charles Spaniel, Hungarian Puli, Rottweiler, Golden Retriever and American Cocker Spaniel.



Fig. 18



Fig. 19



Fig. 20

Fig. 18: PHPV with intralenticular haemorrhage ventrolaterally. The intralenticular haemorrhage progressed to involve the whole lens. **Fig. 19:** PHPV. In this dog, the hyaloid vessel has remained patent and there is haemorrhage into the lens. Note the numerous vacuoles within the lens cortex, which are indicative of progressive cataract formation. **Fig. 20:** The same eye as that pictured in Fig. 19 some months later after cataract formation.

Litter screening is useful, although subtle changes are not always clearly defined. In older animals remodelling of some or all multifocal lesions may result in them becoming less obvious, even disappearing, to ophthalmoscopic examination over time. This does not appear to be the case with the geographic form of RD.

- **Total retinal dysplasia (TRD):** A somewhat more complex form of RD, which is associated with non-attachment or complete detachment of the retina. Non-attachment may result from an apparent failure of contact between the inner (retinal) and outer (retinal pigment epithelial) layers of the optic cup during embryogenesis; other ocular abnormalities, such as microphthalmos and nystagmus, are often present in these cases. The Bedlington Terrier, Labrador Retriever and Sealyham Terrier are certified for TRD under the Eye Examination Scheme, although this condition has been recorded in other breeds, including the Yorkshire Terrier and Samoyed. In the Bedlington Terrier most affected dogs have an infundibular retinal detachment. Puppies are blind from birth and may present with leukocoria, a white appearance to the pupil due to the presence of the detached retina immediately behind the posterior lens capsule. Retinal neovascularisation may result in intraocular haemorrhage. In the Sealyham Terrier a total detachment of the retina is similarly present and microphthalmos and nystagmus are common. Two forms of TRD are recognised as inherited in the Labrador Retriever. In one form, the dysplasia is seen as a complete retinal detachment, apparently resulting from an inability of the developing retina to match the rapid growth of the choroid and sclera. The resulting detachment leads to degeneration of the neurosensory retina due to ischaemic anoxia and such dogs are blind. Other ocular defects, such as microphthalmos, nystagmus and cataract, may also be present.

The second form of TRD, dwarfism with retinal dysplasia type 1 — DRD1, which has not been reported in the UK, is an ocular-skeletal dysplasia associated with severe ocular defects and short-limbed dwarfism. The disease has also been reported in the Samoyed (dwarfism with retinal dysplasia type 2 — DRD2). This phenotype is inherited as an autosomal recessive in both breeds and the mutations have been identified as a 1-base insertional mutation in exon 1 of COL9A3 in the Labrador Retriever, and a 1,267-bp deletion mutation in the 5' end of COL9A2 in the Samoyed.

The MRD/TRD-affected breeds in the UK

Certified for MRD

Cavalier King Charles Spaniel
Hungarian Puli
Retriever (Golden)
Rottweiler
Spaniel (American Cocker)
Spaniel (English Springer)

Certified for TRD

Bedlington Terrier
Sealyham Terrier

Certified for TRD and MRD

Retriever (Labrador)

Significance

A simple autosomal recessive gene is responsible for retinal dysplasia in most breeds. Diagnosis is complicated by the fact that retinal dysplasia may be the result of both genetic and non-genetic influences, the ophthalmoscopic changes may be more difficult to detect in puppies of less than six months of age, and a clear distinction between the various ocular manifestations of the multifocal and total types is sometimes difficult. To add yet further complexity, remodelling of dysplastic lesions may occur over time.

While many dogs with MRD will have no obvious sight deficiency, some can be severely visually impaired, as are all dogs affected with TRD.

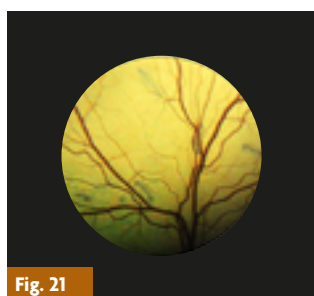


Fig. 21



Fig. 22

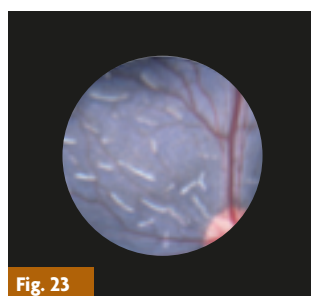


Fig. 23



Fig. 24

Fig. 21: Golden Retriever — multifocal retinal dysplasia (MRD). **Fig. 22:** MRD in an English Springer Spaniel. Some of the larger focal lesions with pigmented centres to the right of the dorsal primary retinal vessels resemble inactive chorioretinopathy, but there are also classical rosettes and vermiform lesions to the left of the vessels. **Fig. 23:** Cavalier King Charles Spaniel puppy — multifocal retinal dysplasia (MRD). **Fig. 24:** Cavalier King Charles Spaniel — geographical retinal dysplasia (GRD).

Collie eye anomaly

The prevalence of Collie eye anomaly (CEA) in the UK remains high in the Rough Collie, Smooth Collie and Shetland Sheepdog, despite years of clinical testing and the relatively recent introduction of a DNA test for one of the defects in the anomaly. CEA is also seen in the Lancashire Heeler and Border Collie, but to a much lesser extent. The condition has a worldwide distribution and ocular lesions of identical ophthalmoscopic appearance have been described in a number of other collie and non-collie breeds, such as the Bearded Collie, the Australian Shepherd and the Kelpie. The disease is a complex disorder affecting retinal, choroidal and scleral development. The classical lesion is a patch of choroidal hypoplasia (CH) localised to the lateral or dorsolateral region of the fundus, near the optic disc. In some animals the CH may be more extensive and it is not uncommon for the two eyes to be dissimilar. The lesion is apparent as a 'pale patch' and is due to a localised lack of some, or all, retinal and choroidal pigment and tapetum. The choroidal vessels in the affected region are also abnormal, usually in size, number and disposition. In merle dogs, with little pigmentation in the fundus and no tapetum, CH will be less obvious and the appearance of the choroidal vessels then becomes the important diagnostic feature. The term 'go normal' has been applied to cases where postnatal pigmentation and tapetal development obscure the CH lesion to ophthalmoscopic examination, giving adult dogs a 'normal' appearance to the fundus. However, this common description is somewhat inappropriate because such dogs are genotypically affected. The phenomenon is common enough to call into question the relevance of examining dogs as adults rather than as puppies. Norwegian data has indicated that the diagnosis of CH in dogs older than three months of age was almost half that for puppies of seven weeks to three months of age. Furthermore, when puppies which had been diagnosed as having CEA with mild CH at between seven weeks and three months of age were re-examined at about one year of age, 68 per cent had normal appearance to the fundus.

In addition to CH, colobomas and staphylomas of the optic nerve head and/or adjacent tissues may be part of the extended phenotype. Such lesions, seen as depressions or holes in the affected tissue, may be present in approximately 30% of dogs affected with CH, but can sometimes be the only visible abnormality where the 'go normal' effect has taken place. Whereas CH has no demonstrable effect on sight, colobomas may be responsible for vision impairment and retinal detachment. However, complications such as retinal detachment and intraocular haemorrhage are fortunately rare, and as such, the majority of dogs with CEA show no apparent visual defect.

Tortuosity of the retinal vessels and retinal folds, the latter usually in the form of vermiform streaks, are not now regarded as part of the syndrome, but may relate to the smallness of the eye in the Collie types.

Significance

A variety of fundamental issues combine to make this a difficult clinical diagnosis on occasion. However it is worth emphasising that CEA is a congenital condition which can be diagnosed as soon as eye examination is possible at five to six weeks of age and, that it is diagnosed clinically with greatest accuracy in the puppy. The 'go normal' effect will complicate diagnosis in dogs older than 12 weeks of age. In aiming to eliminate CEA from a breed, litter screening, combined with DNA testing, is the best approach. CEA mutation has arisen as a single disease allele in a common ancestor of herding breeds and that all affected dogs share a homozygous deletion of 7.8 kb in the *NHEJ1* gene. The availability of a DNA test has proved of great value in devising comprehensive breeding strategies.

The CEA-affected breeds

Border Collie
Collie (Rough)
Collie (Smooth)
Kelpie
Lancashire Heeler
Shetland Sheepdog

Hereditary cataract

The canine lens is an asymmetrical, transparent, biconvex sphere, with more convexity posteriorly. During foetal life it has a blood supply, but shortly after birth all the blood vessels regress and the lens becomes dependent on the aqueous for its nutritional needs and oxygen supply. An adult lens consists of a central nucleus surrounded by cortical fibres which are continuously produced throughout life. The nucleus itself is divisible into various regions according to age: the oldest central portion of the lens, which is the embryonic nucleus surrounded by the foetal nuclear fibres and the outermost portion being the adult nucleus. The whole lens is contained within an acellular capsule derived from epithelial cells, with the anterior part being thicker than the posterior part. A single layer of epithelial cells lies immediately beneath the anterior capsule, and it is these cells which form the germinal cell layer that produces new lens fibres throughout life. These epithelial cells migrate peripherally within the capsule and elongate to form the cortical fibres at the equatorial parts of the lens. Each fibre extends both anteriorly and posteriorly to meet opposing fibres at junction lines known as the suture lines. The suture lines form an upright 'Y' anteriorly and an inverted 'Y' posteriorly.



Fig. 25

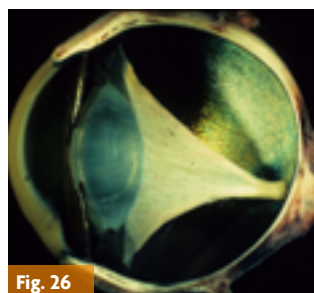


Fig. 26

Fig. 25: Total retinal dysplasia (TRD) in a Labrador Retriever puppy. The retina can be seen lying behind the lens as a greyish membrane. **Fig. 26:** Gross globe – TRD with infundibular retinal detachment.

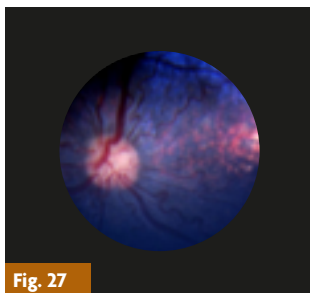


Fig. 27

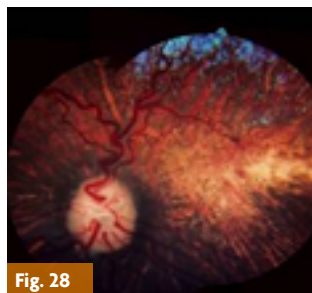


Fig. 28

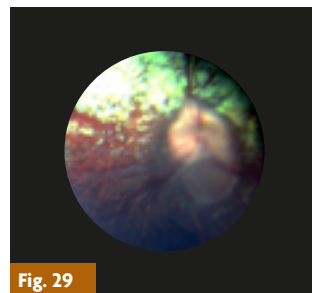


Fig. 29

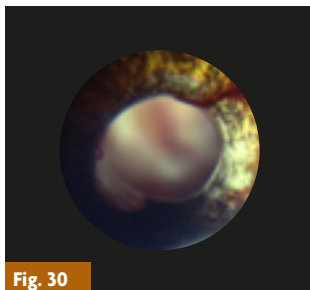


Fig. 30



Fig. 31

Fig. 27: Collie eye anomaly (CEA) in a six-week-old Border Collie puppy. A region of chorioretinal hypoplasia (pale patch) is obvious lateral and slightly dorsal to the optic nerve head—left eye. **Fig. 28:** Border Collie—CEA left eye. **Fig. 29:** CEA in an adult Rough Collie. The most striking feature is the peripapillary coloboma ventral to the optic nerve head—right eye. **Fig. 30:** CEA in an adult Border Collie. This image is dominated by a large colobomatous defect in the centre of the picture and there is also extensive chorioretinal hypoplasia lateral to the coloboma. **Fig. 31:** Rough Collie—CEA with retinal detachment.

Cataract is defined as any opacity of the lens or its capsule. There are many reasons for cataract formation besides inheritance: congenital, due to *in utero* insult; traumatic, as a result of blunt or penetrating injury to the eye; metabolic, as a consequence of, for example, diabetes mellitus; toxic, caused by some drugs; nutritional, produced by inappropriate diets; or a complication of other primary ocular diseases such as uveitis and neoplasia. Cataracts may also develop as a secondary feature in dogs with generalised progressive retinal atrophy (PRA).

Hereditary cataracts (HC) have been reported in several breeds, and breeders should attempt to effect disease control by utilising the Eye Examination Scheme. Fortunately, the age of onset, the appearance, and the evolution of hereditary cataracts (HC) are usually quite specific within the affected breeds, enabling inherited cataracts to be distinguished from other non-inherited types of cataract. Currently, the only congenital example of HC occurs in the Miniature Schnauzer and all other HC develop postnatally. There are variable ages of onset and it is important to examine dogs of over eight years of age to ensure that animals that have been used for breeding remain free of this disease.

Significance

Quite apart from the undesirable perpetuation of abnormality within breeding lines, a proportion of HC progress to produce visual impairment and blindness. The only treatment for cataract is surgical and, although modern techniques give good results, the procedure is still subject to risk and is expensive. Prevention is always preferable.

Primary lens luxation

Primary lens luxation (PLL) is a condition in which an inherited defect in the suspensory ligament (the zonule) of the lens leads to partial or complete dislocation of the lens from its normal position. The clinical signs are not usually observed

before three years of age or later than seven years of age. An acute onset secondary glaucoma is the usual complication, which causes pain and visual loss. Primary lens luxation is recognised as a familial problem in certain terrier breeds (Miniature Bull Terrier, Smooth Fox Terrier, Wire Fox Terrier, Parson Jack Russell Terrier and Sealyham Terrier), the Tibetan Terrier (which is not a true terrier breed), the Lancashire Heeler and the Border Collie. A single nucleotide substitution in the *ADAMTS17* gene has been shown to be the cause of PLL in 17 breeds including the Australian Cattle Dog, the Chinese Crested Dog, the Jagdterrier (also known as the German Hunt Terrier), the Lancashire Heeler, the Miniature Bull Terrier, the Jack Russell Terrier, the Parson Russell Terrier, the Patterdale Terrier, the Rat Terrier, the Sealyham Terrier, the Tenterfield Terrier, the Tibetan Terrier, the Toy Fox Terrier, the Volpino Italiano, the Welsh Terrier, the Wire-haired Fox Terrier and the Yorkshire Terrier. This mutation is not the cause of primary lens luxation in the Border Collie and the Shar Pei, indicating other mutations being responsible in these breeds.

The condition is essentially bilateral, but almost invariably presents unilaterally, as one eye may be affected weeks or

The HC-affected breeds

Alaskan Malamute
Australian Shepherd
Belgian Shepherd Dog
Bichon Frise
Boston Terrier*
Cavalier King Charles Spaniel
French Bulldog
German Shepherd Dog
Giant Schnauzer
Irish Red and White Setter
Large Munsterlander
Leonberger
Miniature Schnauzer*
Norwegian Buhund
Old English Sheepdog
Poodle (Standard)
Retriever (Chesapeake Bay)
Retriever (Golden)
Retriever (Labrador)
Siberian Husky
Spaniel (American Cocker)
Spaniel (Welsh Springer)
Staffordshire Bull Terrier*

*More than one type of hereditary cataract can occur within an individual breed

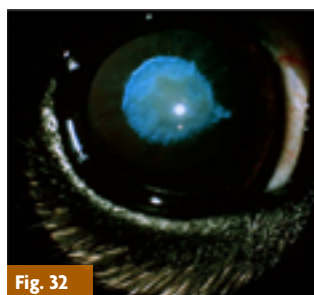


Fig. 32

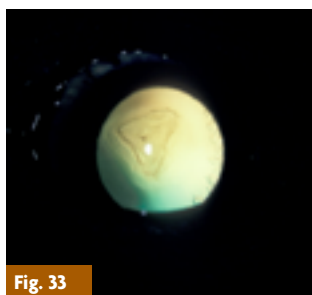


Fig. 33

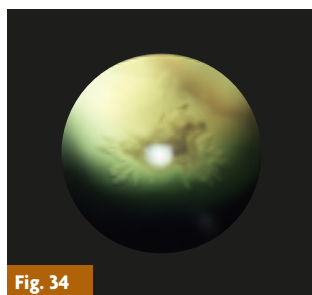


Fig. 34



Fig. 35

Fig. 32: Congenital hereditary cataract (HC) in a Miniature Schnauzer. The nuclear portion of the lens is affected and there is a pyramid-shaped extension medially. Picture: Dr Keith Barnett. **Fig. 33:** HC in a Norwegian Buhund. There is an obvious opacity, located posteriorly, involving the posterior pole and posterior suture lines. In this breed pulverulent nuclear cataracts have also been reported as inherited. **Fig. 34:** HC in a Golden Retriever. The characteristic cataract is located in a posterior polar subcapsular position. **Fig. 35:** Total hereditary cataract in a Labrador Retriever.

The PLL-affected breeds

Border Collie
Bull Terrier (Miniature)
Fox Terrier (Smooth)
Fox Terrier (Wire)
Lancashire Heeler
Parson Russell Terrier
Sealyham Terrier
Tibetan Terrier

months in advance of the other. Observant owners may notice a change in the appearance of the affected eye which correlates with the lens moving out of its normal position. When the lens moves anteriorly, secondary glaucoma develops rapidly and pain, blepharospasm, photophobia, excessive

lacrimation, an increase in intraocular pressure, together with a widely dilated non-responsive pupil, episcleral and conjunctival congestion, and visual loss are the most obvious clinical features. With posterior lens luxation, secondary glaucoma is less likely unless there is an accompanying movement of vitreous material, although there is always the possibility that the lens may move forward at some stage. As the lens becomes unstable, careful observation will reveal a slight trembling of the iris (iridodonesis) with head and eye movement. The unstable lens may be seen to wobble (phacodonesis) and as it moves into the pupil there can be the internal reflection of light highlighting its equatorial regions. A crescent shaped gap (called the aphakic crescent) will appear between the pupillary margin and the lens equator, a sure sign that the breakdown of the lens zonule is well underway.

Significance

Primary lens luxation is an inherited problem which can cause persistent pain and blindness without prompt surgical intervention. It is a severe disease, and to avoid breeding from any affected stock, DNA testing and examination under the Eye Examination Scheme are necessary for at risk breeds.

Progressive retinal atrophy

Progressive retinal atrophy (PRA) is a generic term for a range of genetically heterogeneous inherited retinal diseases affecting many canine breeds. It is a disease of the retinal photoreceptors (the rods and cones) and both developmental (dystrophies) and degenerative types are seen. The developmental types are of early onset and involve the rod or cone photoreceptors, or both. The loss of photoreceptors and rate of progression is usually rapid as the affected photoreceptors do not develop normally. By contrast, the degenerative types involve photoreceptors that have differentiated normally, and the age of onset is later and progression slower.

The clinical findings in PRA are strikingly similar whatever the underlying pathogenesis. Owners may notice an initial loss of night vision, especially when the dog is in unfamiliar surroundings. As the disease progresses there is vision loss under all lighting conditions and poor pupillary light reflexes with dilated pupils. Ophthalmoscopic examination highlights a generalised, bilaterally symmetrical increase in reflectivity as a result of progressive retinal thinning. This is because there is a cellular reflective layer known as the tapetum in the choroidal tissue beneath the retina, which allows the dog to use reflected light to build an enhanced response to a light signal. Thus as the retina thins in degeneration, there is an increase in the level of the tapetal reflectivity. As the retina continues to degenerate there is a progressive narrowing of the superficial retinal vessels, especially the small peripapillary arterioles. This attenuation renders the blood vessels barely visible, being seen as 'ghost vessels' or invisible on ophthalmoscopy. In those dogs with a poorly developed tapetum, this attenuation of the retinal vessels may be the only

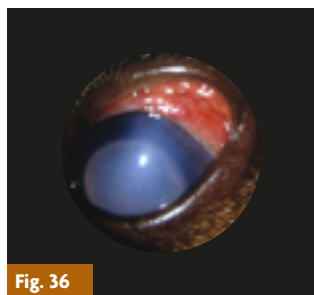


Fig. 36



Fig. 37

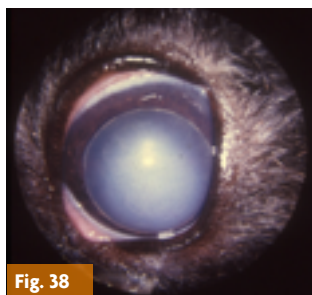


Fig. 38

Fig. 36: Primary lens luxation in a Miniature Bull Terrier. The changes were acute, and the eye painful and red (episcleral and conjunctival congestion), indicative of glaucoma. The companion eye was normotensive (with an IOP of 22 mmHg). The lens has luxated anteriorly and an area of corneal oedema is apparent as a result of endothelial damage from contact with the lens. **Fig. 37:** Tibetan Terrier – primary lens luxation (anterior). The lens equator is highlighted by simple penlight illumination. **Fig. 38:** Tibetan Terrier – primary lens luxation, the anteriorly dislocated lens has developed cataract.

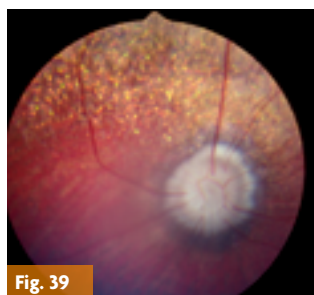


Fig. 39

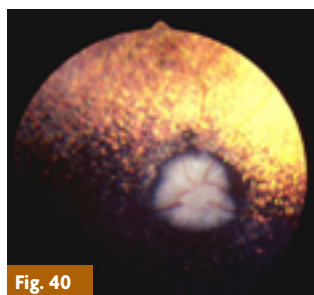


Fig. 40

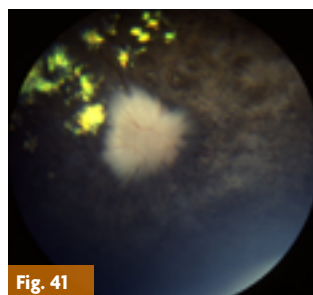


Fig. 41

Fig. 39: Progressive retinal atrophy in a Cocker Spaniel. Attenuation of the retinal vessels and pallor of the optic nerve head are the most obvious features as tapetal islets (a normal variant) do not produce the striking hyperreflectivity seen with a more extensive tapetum. Vision was seriously compromised in this dog. **Fig. 40:** Progressive retinal atrophy in a Miniature Poodle. Tapetal hyperreflectivity is obvious, the optic nerve head is pale and the retinal vessels scarcely visible. The animal was almost totally blind. **Fig. 41:** End stage PRA in a Miniature Poodle. The blood vessels are grossly attenuated and the peripheral optic papilla is crenated as a result of atrophy. Pigmentary changes are visible in the non-tapetal fundus.

The PRA-affected breeds

Australian Cattle Dog
Collie (Rough)
Dachshund (Miniature Long-Haired)
Finnish Lapphund
Glen of Imaal Terrier
Gordon Setter
Irish Setter
Irish Wolfhound
Lhasa Apso
Miniature Schnauzer
Norwegian Elkhound
Papillon
Poodle (Miniature)
Poodle (Toy)
Retriever (Chesapeake Bay)
Retriever (Golden)
Retriever (Labrador)
Retriever (Nova Scotia Duck Tolling)
Spaniel (American Cocker)
Spaniel (Cocker)
Spaniel (English Springer)
Swedish Vallhund
Tibetan Terrier
Welsh Corgi (Cardigan)

obvious ophthalmoscopic sign of early PRA. Later in the course of the disease the optic disc becomes paler due to atrophy of its capillaries and nerve fibres, with changes in shape and actual shrinkage becoming obvious. The non-tapetal pigmented part of the fundus also shows extensive areas of depigmentation as the disease progresses. Retinal detachment is possible and, with time, secondary cataract formation can occur, manifesting as opacities in the posterior cortex, or as radial opacities, before progressing to total cataract formation.

In those types of PRA in which the cones or rods are preferentially affected (for example, cone degeneration in the Alaskan Malamute, or rod dysplasia in the Norwegian

Elkhound) the visual defect which develops will reflect the type of photoreceptor involved. Thus, day blindness occurs when the cone photoreceptors are abnormal and night blindness when rod photoreceptors are abnormal. It is possible for more than one type of PRA to be present in the same breed and the same individual. Electroretinography (ERG) is a valuable diagnostic tool enabling the clinician to detect and differentially diagnose the type of disease with accuracy.

Significance

Most types of PRA are inherited as autosomal recessive traits but, less commonly, autosomal dominant and X-linked types of PRA have also been reported.

There is currently no effective treatment for any of the various types of PRA, so it is DNA testing that underpins efforts to eliminate PRA in many breeds. PRA awareness should be a factor in breeding within the many at risk breeds, and regular examination under the Eye Examination Scheme is necessary to maintain awareness at a high level.

Retinal pigment epithelial dystrophy (formerly Central Progressive Retinal Atrophy)

Retinal pigment epithelial dystrophy (RPED), originally referred to as Central Progressive Retinal Atrophy (CPRA), is a disease of the retinal pigment epithelium, the outermost layer of the retina. The breeds at present certified under the Eye Examination Scheme are the Border Collie, Briard, Rough Collie, Smooth Collie, Golden Retriever, Labrador Retriever, Shetland Sheepdog, Cocker Spaniel, English Springer Spaniel and Cardigan Welsh Corgi. Ophthalmoscopic signs may be detected as early as 15 months old, but the disease may not present until much later in life. Unlike its use in PRA, electroretinography (ERG) evaluation is not of value in the early diagnosis of the disease.

The disease is caused by the inability of the retinal pigment epithelial (RPE) cells to degrade spent photoreceptor material used in the sight process. Work completed in the Briard and English Cocker Spaniel breeds has identified a defect in the metabolism of alpha tocopherol (vitamin E) as the cause of the inability of nonpigmented RPE cells to degrade lipopigment. This results in an accumulation of lipopigment within their cytoplasm which destroys their cellular function, so the retinal pigment epithelium can no longer continue to support the photoreceptors. Only the tapetal fundus is affected because the presence of melanin pigment in the non-tapetal part of the fundus protects the retinal pigment epithelium, therefore some peripheral vision is spared.

In a working breed dog affected with RPED, the owner may initially notice an inability to work in bright light, whilst vision in dim light may be adequate until the disease is advanced. In pet dogs, defective vision may not be noticed as early, but as the disease progresses there is a noticeable loss of the central field of vision. Pupillary light responses are often reasonable and complete blindness is most unusual.

Ophthalmoscopic examination in early disease reveals light brown foci of lipopigment accumulation bilaterally appearing initially in the lateral part of the tapetal fundi. These foci become more numerous, spreading across the tapetal fundus and eventually coalescing into large patches, with hyperreflective areas in-between. In advanced cases the pigment may become less obvious as hyperreflectivity increases. The retinal blood vessels may become attenuated late in the disease, but the non-tapetal fundus and optic disc remain ophthalmoscopically normal.

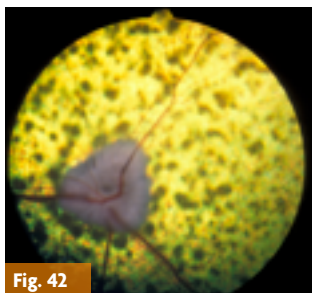


Fig. 42

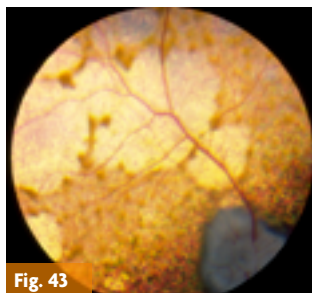


Fig. 43

Fig. 42: Retinal pigment epithelial dystrophy (RPED) in a Cocker Spaniel. At this relatively early stage, multiple focal accumulations of lipopigment are the most obvious feature, together with vascular attenuation and a slightly pale optic nerve head.

Fig. 43: RPED in another Cocker Spaniel at a later stage of disease. The lipopigment has migrated to produce a more cobweb-like appearance.

The Briard is of specific interest in that additionally a defect in retinal polyunsaturated fatty acid metabolism may underlie a form of congenital night blindness in which the appearance of the ocular fundus is initially normal, but by two to three years of age subtle hyper-reflectivity is apparent. Sparse greyish spots appear across the tapetal fundus, increasing in number as the disease progresses and moderate attenuation of the retinal vessels can ensue.

The REPD-affected breeds

Border Collie
Briard
Collie (Rough)
Collie (Smooth)
Retriever (Golden)
Retriever (Labrador)
Shetland Sheepdog
Spaniel (Cocker)
Spaniel (English Springer)
Welsh Corgi (Cardigan)

Significance

Unlike PRA, RPED rarely causes blindness and secondary cataract formation is most unusual. However, when the disease develops in a working dog, the effects are predictably serious due to the loss of central field vision. Inheritance of the disease appears complex and dietary factors influence the phenotypic expression. In the absence of a DNA test, examination under the Eye Examination Scheme is essential to establishing disease control.

CHS Eye Examination Scheme 2022

Alphabetical list of breeds and their eye conditions for certification under the Inherited Eye Diseases Status section of the Certificate of Examination, for which “Clinically Unaffected” or “Clinically Affected” boxes should be ticked:

Alaskan Malamute – HC	Giant Schnauzer – HC	Poodle (Toy) – PRA
Australian Cattle Dog – PRA	Glen of Imaal Terrier – PRA	Retriever (Chesapeake Bay) – PRA, HC
Australian Shepherd – HC	Gordon Setter – PRA	Retriever (Flat Coated) – G
Basset Hound – G, POAG	Hungarian Puli – MRD	Retriever (Golden) – MRD, PRA, RPED, HC, G
Bedlington Terrier – TRD	Hungarian Vizsla – G	Retriever (Labrador) – MRD, TRD, PRA, RPED, HC
Belgian Shepherd Dog (all varieties) – HC	Irish Red and White Setter – HC	Retriever (Nova Scotia Duck Tolling) – PRA
Bichon Frise – HC	Irish Setter – PRA	Rottweiler – MRD
Border Collie – CEA, RPED, PLL, G	Irish Wolfhound – PRA	Sealyham Terrier – TRD, PLL
Boston Terrier – HC (two forms)	Japanese Shiba Inu – G	Shar Pei – POAG
Briard – RPED	Lancashire Heeler – CEA, PLL	Shetland Sheepdog – CEA, RPED
Bull Terrier (Miniature) – PLL	Large Munsterlander – HC	Siberian Husky – G, HC
Cavalier King Charles Spaniel – MRD, HC	Leonberger – G, HC	Spaniel (American Cocker) – MRD, G, PRA, HC,
Collie (Rough) – CEA, PRA, RPED	Lhasa Apso – PRA	Spaniel (Cocker) – G, PRA, RPED
Collie (Smooth) – CEA, RPED	Miniature Schnauzer – CHC, PRA, HC	Spaniel (English Springer) – MRD, G, PRA, RPED,
Dachshund (Miniature Long-Haired) – PRA	Norwegian Buhund – HC	Spaniel (Welsh Springer) – G, HC
Dandie Dinmont Terrier – G	Norwegian Elkhound – PRA	Spanish water Dog – G
Dobermann – PHPV	Old English Sheepdog – HC	Staffordshire Bull Terrier – PHPV, HC
Finnish Lapphund – PRA	Papillon – PRA	Swedish Vallhund – PRA
Fox Terrier (Smooth) – PLL	Parson Russell Terrier – PLL	Tibetan Spaniel – PRA
Fox Terrier (Wire) – PLL	Petit Basset Griffon Vendéen – POAG	Tibetan Terrier – PRA, PLL
French Bulldog – HC	Poodle (Miniature) – PRA	Welsh Corgi (Cardigan) – PRA, RPED
German Shepherd Dog – HC	Poodle (Standard) – HC	

CHS Eye Examination Scheme Litter Screening Checklist 2022

Alphabetical list of breeds and congenital inherited ocular diseases (those ophthalmoscopically identifiable during the neonatal stage) in the current Procedure Notes:

Bedlington Terrier – TRD	Hungarian Puli – MRD	Sealyham Terrier – TRD
Border Collie – CEA	Lancashire Heeler – CEA	Shetland Sheepdog – CEA
Cavalier King Charles Spaniel – MRD	Miniature Schnauzer – CHC	Spaniel (American Cocker) – MRD
Collie (Rough) – CEA	Retriever (Golden) – MRD	Spaniel (English Springer) – MRD
Collie (Smooth) – CEA	Retriever (Labrador) – TRD, MRD	Staffordshire Bull Terrier – PHPV
Dobermann – PHPV	Rottweiler – MRD	

Inherited eye diseases (NB: For a number of breeds a DNA test is available for certain eye conditions – please refer to current list)

CEA = Collie eye anomaly	MRD = Multifocal retinal dysplasia	PRA = Progressive retinal atrophy
CHC = Congenital hereditary cataract	PLL = Primary lens luxation	RPED = Retinal pigment epithelial dystrophy (formerly central progressive retinal atrophy = CPRA)
G = Goniodysgenesis/primary glaucoma	PHPV = Persistent hyperplastic primary vitreous	TRD = Total retinal dysplasia
HC = Hereditary cataract	POAG = Primary open angle glaucoma	

Examples of conditions recorded in comments section

Written descriptive comments can be made on both the Eye Examination Certificate and Litter Screening Eye Examination Certificate providing further information about already known inherited eye diseases and as a means of recording other conditions in the course of the clinical examination. Examples of such comments include:

- Known inherited eye disease that has been on occasion seen in a non-listed breed;
- Other breed-related anomalies and abnormalities of the eye and adnexa which may be inherited;
- Acquired ocular abnormalities which are important from a differential diagnosis standpoint (for example, post-traumatic damage, neoplasia, active/inactive inflammation);
- Ocular changes indicative of systemic disease.

Such observations are important if the abnormalities identified have welfare implications, particularly so if they might be passed on to subsequent generations. Some examples are conformational abnormalities which may cause ocular problems associated with excessive amounts of loose skin, imperfect eyelid anatomy (for example, entropion, ectropion and combinations of entropion and ectropion) and ocular-related disease associated with brachycephaly (for example, lagophthalmos and corneal damage).



Breed-related ocular conditions that may be inherited



Fig. 44

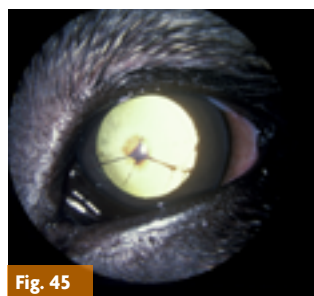


Fig. 45

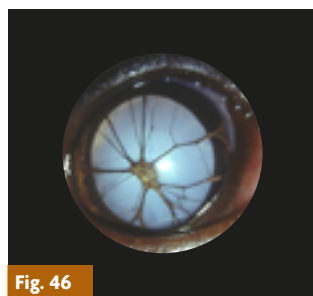


Fig. 46



Fig. 47

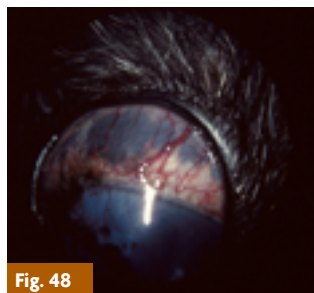


Fig. 48

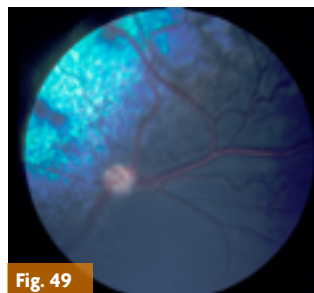


Fig. 49

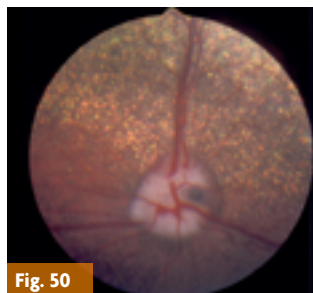


Fig. 50

Fig. 44: Multiocular defects in a Cocker Spaniel. The eye is microphthalmic and a congenital cataract is present. Retinal dysplasia was an additional finding. **Fig. 45:** Persistent pupillary membrane. Most of the remnants arise from the iris collarette and extend anteriorly to the cornea where a discrete opacity is present at the point of contact. **Fig. 46:** Persistent pupillary membrane. The remnants arise from the iris collarette and extend posteriorly to the lens. Extensive cataract formation is present. **Fig. 47:** Congenital hereditary nuclear cataract and uveitis in a Golden Retriever. Note the small darkly pigmented iris cyst in the pupillary aperture medially. **Fig. 48:** Ocular Melanosis in a Cairn Terrier – secondary glaucoma has resulted in an enlarged globe. **Fig. 49:** Optic nerve hypoplasia in a Miniature Poodle. **Fig. 50:** Papillary coloboma, seen as a dark round hole in the right side of the optic nerve.

Other breed-related ocular conditions of known and potential inheritance



Fig. 51



Fig. 52



Fig. 53

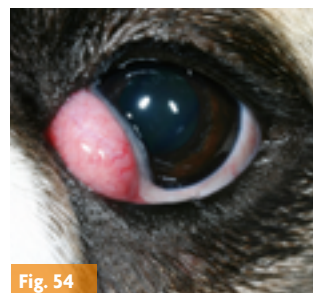


Fig. 54

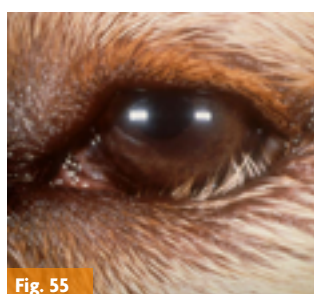


Fig. 55

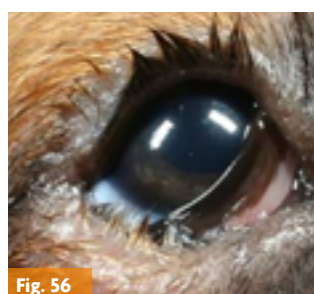


Fig. 56

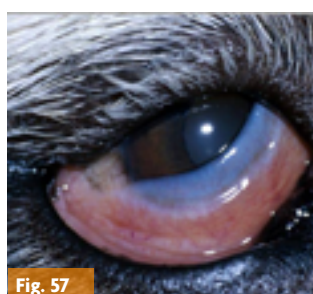


Fig. 57

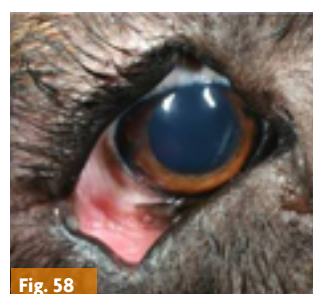


Fig. 58

Fig. 51: Pug – brachycephaly is associated with respiratory and ocular problems. The common anatomical-related problems that render the eye more susceptible to mechanical insult can be summarised as a prominent eye, macropalpebral fissure and lagophthalmos. Chronic pigmentary keratitis is also present in this dog. **Fig. 52:** Shar Pei – multiple skin problems (including poor eyelid conformation and entropion) associated with hereditary cutaneous hyaluronosis. **Fig. 53:** Neapolitan mastiff puppy – excessive skin folds and poor eyelid conformation. **Fig. 54:** Bulldog – prolapsed nictitans gland involving the third eyelid, subtle medial lower lid entropion. **Fig. 55:** Golden Retriever – lower eyelid entropion, left eye. **Fig. 56:** Boerbel puppy – severe bilateral entropion, right eye. **Fig. 57:** Great Dane – lower eyelid ectropion, right eye. **Fig. 58:** St Bernard – ‘diamond eye’ – combined entropion and ectropion, right eye.

Other breed-related ocular conditions of known and potential inheritance

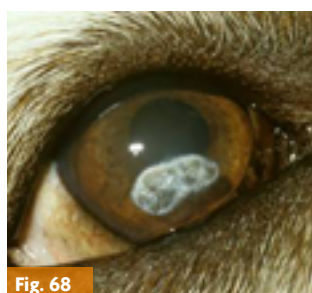
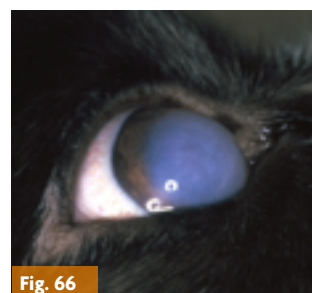
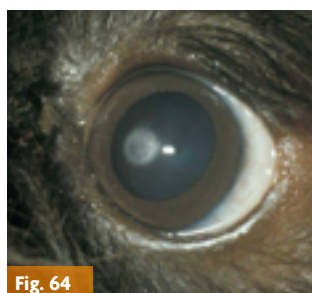
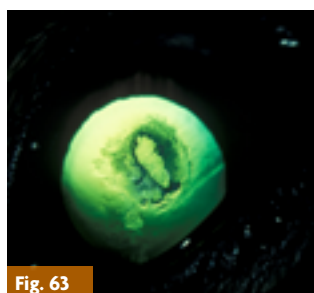
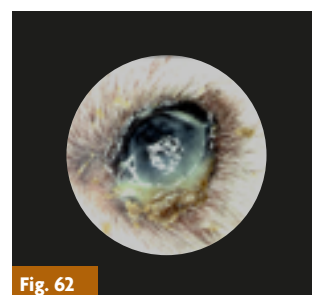
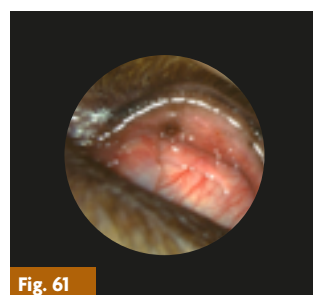
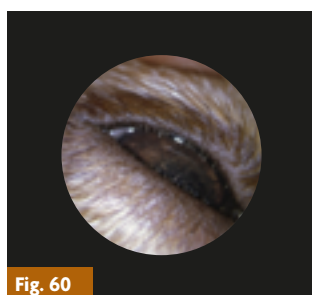
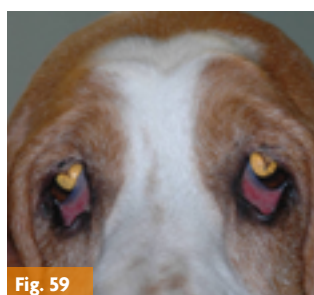


Fig. 59: Basset Hound – combined entropion and ectropion and trichiasis. **Fig. 60:** American Cocker Spaniel – distichiasis. **Fig. 61:** Shetland Sheepdog: a group of ectopic cilia in the dorsal palpebral conjunctiva, left eye. **Fig. 62:** West Highland White Terrier – keratoconjunctivitis sicca, left eye. **Fig. 63:** Boxer – epithelial basement membrane dystrophy, right eye. **Fig. 64:** Cavalier King Charles Spaniel – crystalline corneal dystrophy (Schnyder-like corneal dystrophy), left eye. **Fig. 65:** Labrador Retriever – macular corneal dystrophy, right eye. Picture: Animal Health Trust. **Fig. 66:** English Springer Spaniel – corneal endothelial dystrophy, right eye. **Fig. 67:** German Shepherd Dog – corneal arcus (arcus lipoides corneae) secondary to hypothyroidism, right eye. **Fig. 68:** Golden Retriever – lipid keratopathy, right eye. **Fig. 69:** Labrador Retriever – iris melanoma, left eye. **Fig. 70:** Alaskan Malamute – pulverulent nuclear cataract, right eye. Opacities involving the lens or its capsule are frequently observed as incidental findings under the Eye Examination Scheme and there are many causes, of which possible inheritance is just one.

Some examples of other conditions observed as part of examination under the Eye Examination Scheme

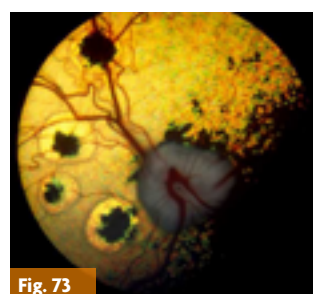
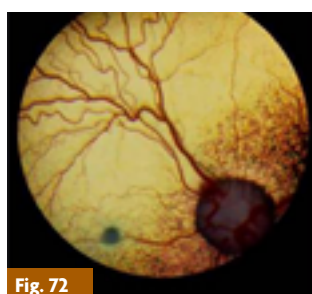


Fig. 71: Briard – a large nevus (black) seen lateral to the optic nerve (pink), right eye. **Fig. 72:** Focal granuloma probably due to ocular larva migrans, left eye. **Fig. 73:** Border Collie – inactive focal chorioretinopathy lesions (probably ocular larva migrans originally), right eye. **Fig. 74:** Several small intraretinal haemorrhages due to systemic hypertensive disease associated with hyperadrenocorticism.

Summary



■ The CHS Eye Examination Scheme offers a means of identifying the presence or absence of inherited eye disease in a variety of dog breeds. There is little doubt that conscientious breeders of all types of dog, both pure-bred and cross-bred, wish to use sound stock with known freedom from inherited eye disease and breed-related ocular disorders as part of their breeding programme. However, in the context of a comprehensive breeding programme, it is important to recognise that inherited problems without any impact on the dog's quality of life may well rank below maintaining genetic diversity and ensuring that breeding pairs are of good temperament and fit for function, an aspect of particular importance in working dogs. Understanding the welfare implications of inherited disease and breed-related ocular disorders is crucial and conditions that may be a cause of pain or blindness, require surgical correction, or lifelong medical therapy, should be regarded as priorities for elimination, as they have substantial effects on the individual's quality of life. All veterinary surgeons involved in clinical practice can help to achieve this ideal by:

- Checking puppies' eyes when they are seen for the first time.
- Informing all pet owners, not just breeders, about inherited eye diseases and the Eye Examination Scheme.
- Ensuring that owners recognise the need for eye examination in any dog which is to be used for breeding, and are aware of the importance of annual examination for dogs used regularly for breeding.
- In addition, older dogs (those over eight years of age) should be examined, in order to ascertain the dog's status in relation to possible later onset eye disease conditions and any changes that may have occurred with pre-existing inherited eye disease and, as a way of assessing ocular and general health.

Up to date information on the CHS Eye Examination Scheme can be obtained from BVA's website, www.bva.co.uk/chs

DNA testing for inherited eye diseases

By Cathryn Mellersh

Most of the inherited eye diseases for which DNA tests are currently available are ‘simple’ or single gene diseases. This means that the disease is a result of a single mutation; no other genes or environmental factors are involved. For these diseases the results of DNA tests are easy to interpret and an individual dog’s risk of developing the disease can be estimated with virtual certainty from the DNA test results. Most simple inherited eye diseases have a recessive mode of inheritance. Every dog has two copies of each gene, one inherited from the dam and one from the sire, so any individual dog has one of three possible genotypes with regard to each single gene disorder:

- *normal or clear*, with two normal copies of the relevant gene
- *carrier*, with one normal copy of the relevant gene and one recessive, mutant copy
- *genetically affected*, with two copies of the recessive mutation.

Recessive mutations cause a loss of function of a gene. Carriers that have inherited a single copy of the normal gene from one parent and a single copy of a mutant gene from the other parent, usually have sufficient normal protein encoded by the normal gene to have healthy functioning eyes. It is only when a dog inherits a faulty gene from both parents that it becomes clinically affected. Consequently, if a mutation is recessive, then dogs with zero (normal/clear) or one copy of the mutation (carriers) will remain clinically free of the disease, although carriers will pass the mutation onto around half of their offspring. Dogs with two copies of the mutation (genetically affected) will almost certainly develop the disease during their lifetime. The age at which dogs typically develop clinical signs depends on the disease, so although a dog with two copies of a particular disease mutation is genetically affected from birth it may not become clinically affected until later in life. It follows, therefore, that a genetically affected dog may be clinically unaffected at the time it has an eye examination, especially if it is very young at the time. Some carriers can be identified by pedigree analysis, once an affected dog has been diagnosed; for example, the clinically normal parents of an affected puppy are both carriers, as are all the clinically normal offspring of clinically affected animals.

The past 10 years have seen remarkable progress in the field of canine molecular genetics. Since the publication of the canine genome sequence in 2004, the genetic tools available to researchers have become increasingly sophisticated and the ease with which mutations responsible for inherited eye diseases in dogs can be identified has increased accordingly. **Table 1** contains details of the genes that have been associated with inherited eye diseases in breeds currently certified under the Inherited Eye Disease Status section of the Certificate of Examination (previously known as Schedule A). Once a particular mutation has been identified, it is usually a relatively simple task to develop a DNA test that can be used to determine an individual dog’s genotype with respect to the disease. Worldwide there are now many facilities offering canine DNA tests. The process of DNA testing involves the submission of a sample of a dog’s DNA to an appropriate testing laboratory. The DNA can usually be submitted

as a simple cheek swab that an owner can take themselves, although some tests/laboratories may require a blood sample. The testing laboratory analyses the DNA for the presence or absence of the relevant mutation and will report back, usually within a few weeks, with the result (the dog’s ‘genotype’).

DNA testing and eye examinations should be regarded as complementary; one does not replace the need for the other. During an eye examination the ophthalmologist examines the eye and adnexa, so will detect any ocular abnormality a dog may have. DNA tests, on the other hand, usually only detect a single, specific mutation, and cannot be used to detect all abnormalities a dog may be suffering from, or detect newly emerging conditions within a breed. For example, Golden Retrievers can suffer from three different forms of progressive retinal atrophy (PRA), known as i) progressive rod cone dystrophy (prcd), ii) Golden Retriever PRA 1 (GR_PRA1) and iii) Golden Retriever PRA2 (GR_PRA2). These diseases are caused by mutations in three different genes so a DNA test for prcd, for example, does not provide any information about a dog’s risk of developing GR_PRA1 or GR_PRA2. Routine eye examination could detect the clinical signs of PRA, providing the dog was old enough to be showing clinical signs, but would not determine which form a dog was suffering from. DNA tests are able to detect carriers, which an eye examination cannot do, and a DNA test can be used from birth to determine whether a dog is genetically affected, before it may have developed clinical signs of disease.

Dogs that are to be used for breeding should have all DNA tests that are relevant to their breed (unless they are hereditarily clear, see *Recording Results* overleaf). They should also be examined and certified under the Eye Examination Scheme, prior to breeding and subsequently, as outlined in the introduction to this pamphlet, including a final examination when the dog is over eight years of age. In those breeds in which inherited congenital/neonatal eye diseases are known or suspected it may also be sensible to carry out litter screening of puppies as outlined earlier. Dogs that are carriers of disease mutations can be bred from safely. Provided all carriers are paired with DNA-tested clear mates, only clear and carrier puppies will be born; no clinically affected dogs will be produced and breeders can select a clear dog to breed on from the resulting litters. **Table 2** details the outcomes of mating dogs with different genotypes (with respect to a recessive mutation) and whether they can result in clinically affected offspring.

Table 1
Genes associated with inherited eye disease in dogs

Disease	Locus or abbreviation	Gene	Breed
Autosomal dominant progressive retinal atrophy	ADPRA	<i>RHO</i>	Bull Mastiff, English Mastiff
Canine multifocal retinopathy	CMR1	<i>VMD2/BEST1</i>	Bull Mastiff, Great Pyrenees, English Mastiff
	CMR2	<i>VMD2/BEST1</i>	Coton de Tulear
	CMR3	<i>VMD2/BEST1</i>	Lapponian Herder
Collie eye anomaly	CEA	<i>NHEJ1</i>	Border Collie, Lancashire Heeler , Rough Collie, Shetland Sheepdog, Smooth Collie
Cone degeneration	CD	<i>CNGB3</i>	Alaskan Malamute, German Shorthaired Pointer
Cone-rod dystrophy		<i>NPHP4</i>	Standard Wire-Haired Dachshund
	CRD3	<i>ADAM9</i>	Glen of Imaal Terrier
	CORD1 (CRD4)	<i>RPGRIP</i>	Miniature longhaired Dachshund
Congenital stationary night blindness	CSNB	<i>RPE65</i>	Briard
Dwarfism with retinal dysplasia (oculoskeletal dysplasia)	DRD2 (OSD2)	<i>COL9A2</i>	Samoyed
	DRD1 (OSD1)	<i>COL9A3</i>	Labrador Retriever
Early retinal degeneration	ERD	<i>STK38L</i>	Norwegian Elkhound
Generalised progressive retinal atrophy	gPRA	<i>CCDC66</i>	Schappendoes
Hereditary cataract	HC, EHC	<i>HSF4</i>	Boston Terrier, French Bulldog, Staffordshire Bull Terrier
	HC	<i>HSF4</i>	Australian Shepherd
Macular corneal dystrophy	MCD	<i>CHST6</i>	Labrador Retriever
Photoreceptor dysplasia	PD	<i>PDC</i>	Miniature Schnauzer
Primary lens luxation	PLL	<i>ADAMTS17</i>	Australian Cattle Dog, Chinese Crested Dog, Fox Terrier, Jack Russell Terrier, Jagdterrier, Lancashire Heeler, Miniature Bull Terrier, Parson Russell Terrier, Patterdale Terrier, Rat Terrier, Sealyham Terrier, Tenterfield Terrier, Tibetan Terrier, Volpino Italiano, Welsh Terrier, Wire-haired Fox Terrier, Yorkshire Terrier
Primary open angle glaucoma	POAG	<i>ADAMTS17</i>	Basset Hound, Beagle, Petit Basset Griffon Vendeen, Shar Pei
Progressive retinal atrophy	PRA	<i>CNGB1</i>	Papillon
	GR_PRA1	<i>SLC4A3</i>	Golden Retriever
	GR_PRA2	<i>TTC8</i>	Golden Retriever
	PRA	<i>MERTK</i>	Swedish Vallhund
Progressive rod-cone degeneration	PRCD	<i>PRCD</i>	Multiple breeds, including American Cocker Spaniel, Australian Cattle Dog, Chesapeake Bay Retriever, Cocker Spaniel, English Springer Spaniel, Golden Retriever, Labrador Retriever, Miniature Poodle, Rough Collie, Toy Poodle
Rod cone degeneration	RCD4	<i>C2orf71</i>	Gordon Setter, Irish Setter, Tibetan Terrier
Rod cone dysplasia	RCD2	<i>RD3</i>	Rough Collie, Smooth Collie
	RCD3	<i>PDE6A</i>	Cardigan Welsh Corgi
	RCD1	<i>PDE6B</i>	Irish Setter, Sloughi
X-linked progressive retinal atrophy	XL PRA2	<i>RPGR</i>	Mixed breed dogs
	XL PRA1	<i>RPGR</i>	Samoyed, Siberian Husky

Table 2
Recessive inheritance — expected outcomes of breeding combinations

Combination of dogs	Outcome	Possibility of clinically affected offspring?
Clear X Clear	All puppies will be clear	No
Clear X Carrier	Approx. 50% of puppies will be clear Approx. 50% of puppies will be carriers	No
Clear x Affected	All puppies will be carriers	No
Carrier x Carrier	Approx. 25% of puppies will be clear Approx. 25% of puppies will be affected Approx. 50% of puppies will be carriers	Yes
Carrier x Affected	Approx. 50% of puppies will be affected Approx. 50% of puppies will be carriers	Yes
Affected x Affected	All puppies will be affected	Yes

Recording results

Eye examination results for all certified conditions are recorded in The Kennel Club's Registration Database, with the information being added to the relevant field of the dog's record. A similar process is in place to deal with the results of DNA testing for inherited eye disease. Breed clubs that have one or more DNA test are encouraged to establish an Official DNA Testing Scheme for the condition(s), whereby the DNA test results for individual dogs are sent directly to The Kennel Club and are added to the dog's registration data. For those puppies resulting from two DNA "clear" tested dogs, an automatic status of "hereditarily clear" is assigned and published as described below.

For breeds examined that are not currently certified under the Eye Examination Scheme, but, where the dog is registered with The Kennel Club, a note will be added to the dog's record on The Kennel Club's database confirming that an examination has taken place and the date of the examination.

The addition of health screening results to The Kennel Club Registration Database triggers dissemination via a number of different routes. The result(s) will appear:

- On any new registration certificate issued for the dog
- On the registration certificates of any of the dog's future progeny
- In The Kennel Club Breed Records Supplement (BRS), a quarterly publication

The Kennel Club also maintains lists of DNA test results for all Official DNA Testing Schemes on the health pages of its website (www.thekennelclub.org.uk).

Appeals procedure

Eye Examination Certificate

Any appeal against the results of an eye examination must be lodged in writing with the BVA *within 30 days* of the examination. BVA's address is shown overleaf.

The owner may then take the dog, together with the certificate issued by the first panellist, for examination by the Chief Panellist, a panellist from the Eye Panel Working Party, or another panellist. The second panellist will charge the normal fee. If the second panellist agrees with the first panellist the appeal will be deemed to have failed and the second panellist will inform BVA accordingly. In such an event no further appeal is possible.

The decision of the Chief Panellist at second examination is final, as also is the decision of a panellist from the Eye Panel Working Party acting in consultation with the Chief Panellist and provided that the Chief Panellist agrees with the findings reported. In all other circumstances, if the second panellist disagrees with the first panellist the dog shall be referred to the Chief Panellist for further examination without additional fee to the owner. The decision of the Chief Panellist will be final and the Chief Panellist will advise BVA of the result accordingly. Owners may choose to see the Chief Panellist as the second panellist once an appeal has been lodged with BVA. If so, the normal fee will be charged.

The final result of any appeal must be received by BVA *within 90 days* of examination, otherwise the first result may be sent to The Kennel Club and/or the International Sheep Dog Society for publication.

Litter Screening Eye Examination Certificate

Any appeal against the results of litter screening must be notified to BVA and/or Chief Panellist as soon as possible so that suitable arrangements may be made for the whole litter to be re-examined before the puppies reach 12 weeks of age. No appeal will be granted unless all the puppies are re-examined on the same occasion and the puppies are less than 12 weeks of age.

CHS Eye Examination Scheme

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February 2023

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