

DNA testing for inherited eye diseases in dogs – FAQ's

What is DNA testing?

DNA is found within most cells of the body and contains all the genetic information of an animal. We can sample this DNA and test it to find out whether an animal is at risk of developing certain inherited diseases, or is at risk of passing them on to their offspring. This is called DNA testing.

The process usually involves taking a small blood sample from your dog, from which DNA is extracted. DNA can also be found in other cells in the body, so in some cases a mouth swab to collect cells from the inside of the cheek is all that is required.

Is DNA testing the same as DNA profiling?

No, DNA profiling describes a different technique that uses differences in DNA inheritance patterns to uniquely identify an animal. DNA profiling is primarily used for paternity testing, not to screen for genetic diseases.

Which eye conditions can be screened for with DNA testing?

Currently, DNA tests are available for the following eye conditions that are listed on Schedule A or B of the BVA/KC/ISDS Eye Scheme (see Appendix for details of the specific tests):

- **Collie Eye Anomaly (CEA)** in the Border Collie; Collie (Rough); Collie (Smooth); Lancashire Heeler; Shetland Sheepdog
- **Hereditary cataract (HC)** in the Australian Shepherd; Boston Terrier (early onset); French Bulldog; Staffordshire Bull Terrier (early onset)
- **Generalised Progressive Retinal Atrophy (GPRA)** in the Australian Cattle Dog; Collie (Rough); Dachshund (Miniature Long-Haired); Dachshund (Miniature Smooth-Haired); Finnish Lapphund; Glen of Imaal Terrier; Gordon Setter; Irish Setter (early and late-onset PRA); Miniature Schnauzer; Norwegian Elkhound; Poodle (Miniature); Poodle (Toy); Retriever (Chesapeake Bay); Retriever (Golden); Retriever (Labrador); Retriever (Nova Scotia Duck Tolling); Spaniel (American Cocker); Spaniel (Cocker); Spaniel (English Springer); Tibetan Spaniel; Tibetan Terrier; Welsh Corgi (Cardigan); Yorkshire Terrier
- **Primary Lens Luxation (PLL)** in the Bull Terrier (Miniature); Fox Terrier (Wire); Lancashire Heeler; Parson Russell Terrier; Sealyham Terrier; Tibetan Terrier.
- **Primary Open Angle Glaucoma (POAG)** in the Petit Basset Griffon Vendeen

Please note that research is ongoing and so new tests are constantly being developed. Contact info@thekennelclub.org.uk, aht.org.uk, or optigen.com for the latest information.

What are the benefits of DNA testing?

There are three main benefits of DNA testing:

1. It identifies 'at risk' animals before they develop clinical signs of the disease itself. Since many inherited eye diseases develop relatively late in life, this allows us to identify affected dogs before they reach breeding age
2. It allows us to identify carrier animals (which do not develop the disease themselves but may pass it onto their offspring)
3. It identifies genetically clear dogs that will be guaranteed not to pass the condition to any of their offspring and so can be used safely in a breeding programme

How does DNA testing compare with eye testing under the BVA Eye Scheme?

A DNA test only checks for a single inherited condition, whereas an eye examination screens your dog for a wide range of eye diseases. For example, the Labrador Retriever is known to be affected by 5 inherited eye diseases, but there is at present a DNA test available for only one of these (GPRA). Because of this we would strongly advise you to continue to bring your dog for regular eye examinations.

As well as detecting breed specific certified eye diseases, eye testing also plays an important surveillance role in identifying new inherited eye diseases within breeds, before they become an established problem. It also provides an expert health check for your dog's eyes, and can yield information about other, non-inherited, diseases of importance.

How do I get my dog DNA tested for inherited eye diseases?

DNA testing for some inherited eye conditions involves taking a blood sample, for others a cheek swab is sufficient – see next section for details.

Where a blood sample is needed, contact your own veterinary surgeon to arrange this. 2mls blood is required, taken into an EDTA blood tube that should be labelled with the owner's name, dog's name, the breed and the date.

Where a cheek swab is needed, a sterile cytobrush/applicator should be used. Some testing laboratories may be able to supply these to your veterinary surgeon, or advise them on where they can be purchased. It is important that your pet is not fed or given water for an hour prior to the swab, to reduce the risk of contamination.

Who can take the samples?

Blood samples may only be taken by (or under the supervision of) a veterinary surgeon.

Cheek swabs may be taken by the owner/ handler. Cheek swabs and advice on sampling technique can be obtained from the Animal Health Trust (www.aht.org.uk).

Where should the samples be sent to?

See Appendix for a list of tests currently available, what samples are required and where these should be sent. Submission forms and instructions on packing/ shipping can be downloaded from the relevant web sites (Animal Health Trust: www.aht.org.uk; Optigen: www.optigen.com).

Optigen samples can be sent directly to the Optigen laboratory in the USA, or via van Haeringen Laboratorium in the Netherlands (email: info@vhlgenetics.com; website: www.vhlgenetics.com).

Can the DNA test result ever be at odds with the eye test result?

In the majority of cases the answer is no. However there are a few scenarios which may result in a discrepancy between DNA test and eye test results:

- An erroneous diagnosis during the eye test or a laboratory mistake during the DNA testing procedure could lead to conflicting results
- A dog affected with a 'late-onset' condition could have been DNA tested as a young dog, showing that it is genetically affected, but have clear eye examination results in the early part of its life before the onset of clinical signs
- On occasion, genetic eye diseases may show 'variable expression'. This means that some dogs, which according to the DNA test are affected, never go on to develop the disease. In such cases we presume that there must be something in the rest of these dogs' genetic makeup that is delaying or preventing the onset of clinical signs. However, from a breeding selection standpoint, the genetic result must take precedence; the dogs will pass the genetic mutation to their offspring, and these offspring will be at risk of developing the disease at the expected age
- It is also possible that future dual testing will reveal a scenario of a dog that, on its eye test, shows clinical signs typical of a particular inherited condition, but yet the DNA test is clear. Under such circumstances we would have to consider the possibility that there might be at least two different genetic causes of the disease in the breed.

Appendix

Details of disease subtypes or mutation tests available for inherited eye conditions listed on schedule A or B or the BVA/KC/ISDS Eye Scheme.

Disease	Breed	Disease subtype or mutation	Sample required	Laboratory
CEA	Border Collie Lancashire Heeler ; Rough Collie ; Smooth Collie; Shetland Sheepdog	Choroidal hypoplasia	Blood (EDTA)	Optigen
HC	Australian Shepherd ; Boston Terrier (early onset);French Bulldog; Staffordshire Bull Terrier (early onset)	HSF4	Cheek swab	AHT
GPRA	Australian Cattle Dog ; Finnish Lapphund; Norwegian Elkhound; Poodle (Miniature); Poodle (Toy); Retriever (Chesapeake Bay); Retriever (Golden); Retriever (Labrador; Retriever (Nova Scotia Duck Tolling); Spaniel (American Cocker); Spaniel (Cocker); Yorkshire Terrier	prcd	Blood (EDTA)	Optigen

GPRA	Collie (Rough)	rcd-2	Blood (EDTA)	Optigen
GPRA	Dachshund (Miniature Long-Haired); Dachshund (Miniature Smooth-Haired); Spaniel (English Springer)	cord-1/ CRD	Cheek swab	AHT
GPRA	Glen of Imaal Terrier	rcd-3	Blood (EDTA)	Optigen
GPRA	Golden Retriever	GR_PRA1 GR_PRA2 (+ prcd- see above)	Cheek swab	AHT
late onset GPRA (LOPRA)	Gordon Setter (late onset) Irish Setter (late onset)	rcd-4	Cheek swab	AHT
GPRA	Irish Setter (early onset)	rcd-1	Cheek swab Blood (EDTA)	AHT Optigen
GPRA	Miniature Schnauzer	Type-A PRA	Blood (EDTA)	Optigen
GPRA	Papillon	Pap_PRA1	Blood (EDTA)	Optigen
GPRA	Tibetan Spaniel	PRA3	Cheek swab	AHT
GPRA	Tibetan Terrier	rcd-4 PRA3	Cheek swab	AHT
GPRA	Welsh Corgi (Cardigan)	rcd-3	Blood (EDTA)	Optigen
PLL	Bull Terrier (Miniature); Fox Terrier (Wire); Lancashire Heeler; Parson Russell Terrier; Sealyham Terrier; Tibetan Terrier	ADAMTS17	Cheek swab	AHT
POAG	Petit Basset Griffon Vendeen		Cheek swab	AHT