



## Primary Lens Luxation

**Primary Lens Luxation** (PLL) is a well-recognised, painful and blinding inherited eye condition that affects many breeds of dog, particularly terrier and terrier-type breeds including (but not restricted to) Miniature bull terriers, Tibetan terriers, Jack and Parson Russell terriers, Lancashire Heelers and Chinese Crested dogs, also the Australian Cattle Dog, Jagd Terrier, Patterdale Terrier, Rat Terrier, Sealyham Terrier, Tenterfield Terrier, Toy Fox Terrier, Volpino Italiano, Welsh Terrier, Wire-haired Fox Terrier and Yorkshire Terrier.

In affected dogs the zonular fibres which support the lens breakdown or disintegrate, causing the lens to fall into the wrong position within the eye. If the lens falls into the anterior chamber of the eye glaucoma and loss of vision can quickly result.

Scientists at the AHT have identified a mutation that is associated with the development of PLL in several breeds of dog. The DNA test we are now offering examines the DNA from each dog being tested for the presence or absence of this precise mutation. It is thus a 'mutation-based test' and not a 'linkage-based test'.

Breeders will be sent results identifying their dog as belonging to one of three categories:

**CLEAR:** these dogs have two normal copies of DNA. Our research has demonstrated clear dogs will not develop PLL as a result of the mutation we are testing for, although we cannot exclude the possibility they might develop PLL due to other causes, such as trauma or the effects of other, unidentified mutations.

**CARRIER:** these dogs have one copy of the mutation and one normal copy of DNA. Our research has demonstrated that carriers from some breeds have a very low risk of developing PLL. The majority of carriers do not develop PLL during their lives but a small percentage do. This has been particularly noted for the Miniature Bull Terrier during our study and is also suggestive in the Lancashire Heeler. For Tibetan Terriers our study did not show any evidence to suggest that carriers will develop PLL. For the Miniature Bull Terrier we estimate that between 2% and 20% of carriers will develop the condition, although we believe the true percentage is nearer to 2% than 20%. We do not currently know why some carriers develop the condition whereas the majority do not, and we advise that all carriers have their eyes examined by a veterinary ophthalmologist every 6- 12 months, from the age of 2, throughout their entire lives. The Animal Health Trust welcomes samples from dogs with a robust diagnosis of PLL that DO NOT have two copies of the mutation. Owners can submit samples as cheek swabs to our research department by contacting Bryan McLaughlin (email: [bryan.mclaughlin@aht.org.uk](mailto:bryan.mclaughlin@aht.org.uk)).

**GENETICALLY AFFECTED:** these dogs have two copies of the mutation and will almost certainly develop PLL during their lifetime. We advise that all genetically affected dogs have their eyes examined by a veterinary ophthalmologist every 6 months, from the age of 18 months, so the clinical signs of PLL are detected as early as possible.

### **Breeding Advice**

Our original research demonstrated that the frequency of the PLL mutation is extremely high in the PLL-affected breeds that we have studied in depth. This means that allowing only CLEAR dogs to breed could have a devastating effect on breed diversity and substantially increase the likelihood of new inherited diseases emerging. Therefore, we strongly advise breeders to consider all their dogs for breeding, regardless of their PLL genotype. GENETICALLY AFFECTED and CARRIER dogs can be bred with, but should only be bred to DNA tested, CLEAR dogs. All puppies from any litter that has at least one CARRIER parent should be DNA tested, so that the CARRIERS can be identified and followed clinically throughout their lives. This practise should be followed for at least one or two generations, to allow the PLL mutation to be slowly eliminated from the population without severely reducing the genetic diversity of breeds at risk.

**Frequently Asked Questions.** If you have any questions about the PLL test please see if you can find an answer in our list of FAQ's on our website [http://www.aht.org.uk/genetics\\_pllfaq.html](http://www.aht.org.uk/genetics_pllfaq.html)

**Tests can be ordered through our Webshop <http://www.ahtdnatesting.co.uk>.**