

## FAQs regarding the PLL DNA Test

**Q. *I have always been told PLL is a recessive condition but now the AHT is saying that carriers can develop the condition; why is this and what does that mean?***

**A.** Our extensive research has shown that for some breeds a very small number of carriers do develop bilateral, primary lens luxation whereas dogs that do not have the PLL mutation do not. 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. This means that carriers should be considered to have a low (but not zero) risk of developing PLL.

**Q. *Why do some carriers develop PLL whereas the majority don't?***

**A.** At the current time we simply don't know. A combination of genetic and environmental factors is likely to contribute to an individual dog's risk of developing the condition. Because these factors are not understood at this present time it is not possible to predict which carriers will develop PLL and all carriers should, therefore, be considered to be at the same low risk. 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)).

**Q. *What is the exact risk of a carrier developing PLL?***

**A.** The ideal way to calculate the exact number of carriers that develop PLL would be to clinically monitor a large number of carriers over their whole lives and record how many develop PLL and how many don't. This is obviously a long-term process; in the mean time we have estimated the risk using the information we have for the several thousand DNA samples that have been donated to our PLL research over the years.

**Q. *Why does the AHT advise that all carriers have their eyes examined by a veterinary ophthalmologist from the age of 2, whereas genetically affected dogs should have their eyes examined from the age of 18 months?***

**A.** We have some preliminary evidence that the carriers that develop PLL may do so at a slightly older age than genetically affected dogs, which have two copies of the mutation.

**Q. *Given that carriers have a low risk of developing PLL surely it makes sense to only breed with clear dogs?***

**A.** Unfortunately it doesn't. Our original research showed that for some breeds as many as 45-50% of the dogs were carriers; eliminating all these dogs from the breeding population would reduce the genetic diversity of the breed dramatically and could ultimately be very detrimental. We suggest breeders DNA test all their potential breeding stock and continue to breed with their best dogs, regardless of their PLL genotype. However, carriers and genetically affected dogs should only be mated to clear dogs and all resulting puppies should be tested to identify any carriers which should subsequently be monitored throughout their lives.

**Q.** ***I always thought that once a DNA test was available for PLL we could avoid producing any more affected dogs – now we are being told that carriers, which have a low risk of developing PLL, should be bred with. Surely this will result in more carriers being born, some of which will develop PLL?***

**A.** Yes, this is correct. However, by ensuring that carriers are only mated to clear dogs, no genetically affected dogs need now be produced. Approximately half of the puppies that result from a carrier x clear mating will be carriers but each of those will only have a low risk of developing PLL. We are not suggesting carriers should be bred with forever; merely that in the first instance quality dogs should be selected for breeding, regardless of their genotype, so that the gene pool isn't reduced dramatically. If carriers are only ever mated to clear dogs each resulting puppy has a 50% of being clear of the PLL mutation and sooner or later the 'pick of the litter' will happen to be a clear dog that can be bred on.

**Q.** ***Is the information & breeding advice being offered applicable to all PLL-affected breeds?***

**A.** The AHT's PLL research to date has focused on Miniature Bull terriers, Lancashire Heelers and, to a lesser extent, Tibetan terriers because these were the breeds for which we had the most DNA samples. As we proceed to DNA test dogs for the PLL mutation we will generate data regarding the frequency of the PLL mutation and the numbers of genetically affected, clear and carrier dogs within each breed; we will make this breed-specific data available in due course. The length of time it takes to generate robust data for each breed will depend on the numbers of dogs of each breed that we test.

**Q.** ***Do I need a vet to take the swabs or can I take them myself?***

**A.** The AHT will test the DNA of any dog, regardless of whether the owner or a vet takes the swabs, but a vet must take the EDTA blood sample. However, some breed clubs will only recognise DNA test results from dogs whose identification was verified by a vet at the time the swab/blood was taken. You are therefore advised to check with your relevant Breed Club.