

## Geneticists identify a mutation for Primary Lens Luxation (PLL) in several breeds. (September 2009)

A mutation responsible for the development of primary lens luxation in many breeds of dogs has been identified by geneticists working in the Kennel Club Genetics Centre at the Animal Health Trust, led by Dr Cathryn Mellersh, in collaboration with Dr David Sargan (Cambridge University) and Dr David Gould (Davies Veterinary Specialists).

A DNA test for this mutation is expected to be available by late October, 2009.

**Primary Lens Luxation (PLL)** is a well-recognised, painful and blinding inherited eye condition that affects many breeds of dog. In affected dogs the zonular fibres that 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.

The team of scientists have identified a mutation that is responsible for the development of PLL in several breeds, including the Miniature Bull Terrier, the Lancashire Heeler, **Tibetan Terrier**, the Jack Russell Terrier, the Parson Russell Terrier, the Patterdale Terrier, the Sealyham Terrier and the Chinese Crested dog.

A DNA test soon to be made available at the Animal Health Trust can be used to determine a dog's genotype with respect to this mutation. Dogs will be identified as **CLEAR** (has two normal copies of the gene), **CARRIER** (has one normal copy and one mutated copy) and **AFFECTED** (has two copies of the mutation). Breeders will be given an estimate of each dog's risk of developing PLL depending on their genotype and will be able to make sensible breeding decisions that minimise the risk of producing dogs that will become affected by this serious and debilitating condition.

Breeders and owners will be able to test their dogs using a simple test kit that will be made available shortly. It will be possible to collect DNA from dogs to be tested via a simple cheek swab which will be included in the test kit, along with all information, necessary forms and full instructions.

**PLEASE NOTE: THIS TEST IS NOT YET AVAILABLE. FULL DETAILS WILL BE POSTED ON THIS WEBSITE AS SOON AS THEY HAVE BEEN FINALISED:**

<http://www.aht.org.uk/news.html#pll>

Owners who submitted samples for the PLL research prior to September 1, 2009 may request test results using a form that will also shortly become available on this website:

<http://www.aht.org.uk/news.html#pll>

We would like to sincerely thank all the many owners and breeders, from all over the world, who have contributed DNA and information from their dogs to this project. This discovery would not have been possible without them. We would also like to thank everybody who has made a financial donation to support the research.