



## Progressive Retinal Atrophy in Tibetan Terriers

Geneticists at the Kennel Club Genetics Centre at the Animal Health Trust have discovered a mutation that causes a form of progressive retinal atrophy (PRA) in Tibetan Terriers. We are calling this form of the disease **PRA3** to distinguish it from other, genetically distinct, forms of PRA that are caused by different mutations, including the previously reported RCD4 mutation that is also known to cause PRA in some Tibetan Terriers. Together the PRA3 and RCD4 mutations account for approximately half of Tibetan Terrier PRA cases that we investigated during our research, although the number of samples from dogs with PRA was small and these proportions might not be representative of the wider population. During the latter half of 2013 the AHT will collaborate with the Kennel Club (KC) to screen a random subset of KC registered Tibetan Terriers to determine the frequency of both mutations more accurately. The mutation(s) that cause PRA in the other affected Tibetan Terriers remain(s) unknown and cannot therefore be detected by any DNA test at present.

As for RCD4, the PRA3 mutation is recessive, meaning a dog needs to inherit two copies of the mutation to be clinically affected with PRA. PRA3 is a late-onset condition and clinical signs can usually be detected by an ophthalmologist from 4-7 years of age. The onset of RCD4 is variable, but is usually around 10 years of age. Any Tibetan terrier that has 2 copies of either the PRA-3 or RCD4 mutation will develop PRA, assuming it lives long enough to do so.

A DNA test for PRA3 will become available from the Animal Health Trust July 8<sup>th</sup>, 2013

Full details will be made available on our website shortly:

[http://www.aht.org.uk/cms-display/genetics\\_tests.html](http://www.aht.org.uk/cms-display/genetics_tests.html)

**May 8<sup>th</sup>, 2013**