Progressive Retinal Atrophy in Tibetan Terriers

**Progressive Retinal Atrophy (PRA)** is a well-recognised inherited condition that many breeds of dog are predisposed to. The condition is characterised by bilateral degeneration of the retina which causes progressive vision loss that culminates in total blindness. There is no treatment for PRA, of which several genetically distinct forms are recognised, each caused by a different mutation in a specific gene. The various forms of PRA are typically breed-specific, with clinically affected dogs of the same breed usually sharing an identical mutation. Clinically affected dogs of different breeds, however, usually have different mutations, although PRA-mutations can be shared by several breeds.

**Mutation Identified**

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 8th, 2013. Breeders using the test will be sent results identifying their dog as belonging to one of three categories:
CLEAR: these dogs have two normal copies of DNA. Clear dogs will not develop PRA as a result of the PRA3 mutation, although we cannot exclude the possibility they might develop PRA due to other mutations they might carry that are not detected by this test.

CARRIER: these dogs have one copy of the mutation and one normal copy of DNA. These dogs will not develop PRA themselves as a result of the PRA3 but they will pass the mutation on to approximately 50% of their offspring. We cannot exclude the possibility that carriers might develop PRA due to other mutations they might carry that are not detected by this test.

GENETICALLY AFFECTED: these dogs have two copies of the PRA3 mutation and will almost certainly develop PRA during their lifetime.

Advice

The mutation is recessive which means that all dogs can be bred from safely but carriers and genetically affected dogs should only be bred to DNA tested, clear dogs. About half the puppies from any litter that has a carrier parent will themselves be carriers and any dogs from such litters that will be used for breeding should themselves be DNA tested prior to breeding so appropriate mates can be selected.

It is advisable for all breeding dogs to have their eyes clinically examined by a veterinary ophthalmologist prior to breeding and throughout their lives, with at least one examination occurring when the dog is at least 8 years of age, so that any cases of PRA caused by additional mutations can be detected and that newly emerging conditions can be identified.

Frequently Asked Questions. If you have any questions about the PRA test please see if you can find an answer in our list of FAQs.