



## **Progressive Retinal Atrophy in Miniature Long-haired Dachshunds**

Progressive retinal atrophy (PRA) is a term for retinal degenerations occurring in many breeds of dog. Many forms of PRA exist, each form being confined to one or a few breeds only. The disease results in a degeneration of the light-sensitive membrane at the back of the eye - the retina - resulting in loss of vision, and often leading to blindness.

The form of the disease occurring in miniature long-haired dachshunds can be diagnosed by electroretinography at a few months of age, although obvious clinical signs take longer to develop. The exact course of the disease can vary between individual dogs, and some don't develop symptoms until relatively late in life.

The disease is caused by a change to a gene involved in sight. This change, or mutation, occurred spontaneously, but once in the population has been inherited from generation to generation like any other gene. The mutation upsets the delicate processes involved in vision and causes the long-term degeneration seen. This form of PRA shows an autosomal recessive mode of inheritance: two copies of the defective gene (one inherited from each parent) have to be present for a dog to be affected by the disease. Individuals with one copy of the defective gene and one copy of the normal gene - called carriers - show no symptoms but can pass the defective gene onto their offspring. When two apparently healthy carriers are crossed, 25% (on average) of the offspring will be affected by the disease, 25% will be clear and the remaining 50% will be carriers.

There is currently no treatment for the disease. Breeding stock are regularly checked by eye examination, although this can only pick up affected dogs after symptoms have developed and will never detect the symptomless carriers.

The gene responsible has been identified at the Animal Health Trust and we have recently identified the mutation causing the disease. Using the information from this research, we have developed a DNA test for the disease. This test not only diagnoses dogs affected with this disease but can also detect those dogs which are carriers, showing no symptoms of the disease but producing affected pups. Under most circumstances, there will be a much greater number of carriers than affected animals in a population. It is important to eliminate such carriers from a breeding population since they represent a hidden reservoir of the disease that can produce affected dogs at any time.

The test is available from February 14<sup>th</sup>, 2005 and information on submitting samples is given below.

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

- CLEAR:** the dog has 2 copies of the normal gene and will neither develop PRA, nor pass a copy of the PRA gene to any of its offspring.
- CARRIER:** the dog has one copy of the normal gene and one copy of the mutant gene that causes PRA. It will not develop PRA but will pass on the PRA gene to 50% (on average) of its offspring.
- AFFECTED:** the dog has two copies of the PRA mutation and is affected with PRA. It will develop PRA at some stage during its lifetime, assuming it lives to an appropriate age.

Carriers can still be bred to clear dogs. On average, 50% of such a litter will be clear and 50% carriers; there can be no affecteds produced from such a mating. Pups which will be used for breeding can themselves be DNA tested to determine whether they are clear or carrier.

**Samples for testing, either blood (3mls in an EDTA tube) or cheek swabs, should be sent together with a completed DNA Testing form and a cheque for £60-00 (inc VAT) for each sample to Genetic Services, Animal Health Trust, Lanwades Park, Kentford, Newmarket, Suffolk CB8 7UU. DNA testing forms can be downloaded from our web site (<http://www.aht.org.uk>). DNA testing forms and cheek swabs can also be obtained by contacting Vikki Lett 01638 750659 ext 1223 or via e-mail to [vikki.lett@aht.org.uk](mailto:vikki.lett@aht.org.uk).**