

**ATROFIA PROGRESIVA DE RETINA**, también conocida como APR o PRA, en sus variantes y mutaciones prcd PRA (poco común en el Golden europeo, más en las líneas americanas), PRA 1 y PRA 2.

En qué consiste la enfermedad:

<http://visionveterinaria.es/?p=390>



Es posible desde 2012 de test genético de las mutaciones europeas por la Animal Health Trust

En resumen, cabe la posibilidad de tres resultados:

Clear o libre, en cuyo caso el perro no está genéticamente afectado por la enfermedad.

Carrier o portador: El animal no desarrolla la enfermedad pero puede transmitirla.

Affected o enfermo: El Golden es portador de la enfermedad y la transmite.

## PROGRESSIVE RETINAL ATROPHY IN THE GOLDEN RETRIEVER

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 AHT, in collaboration with scientists from at the Swedish University of Agricultural Sciences and Uppsala University, have identified a recessive mutation that is associated with the development of a form of PRA in the Golden Retriever, known as GR\_PRA2. The DNA test we are offering (from August 13<sup>th</sup> 2012) examines the DNA from each dog being tested for the presence or absence of this precise mutation and is thus a ‘mutation-based test’ and not a ‘linkage-based test’.

### **Other Forms of PRA**

The mutation is different from both i) the previously described mutation that cause the 'prcd' (progressive rod-cone degeneration) form of PRA that OptiGen ([www.optigen.com](http://www.optigen.com)) offers a DNA test for and ii) the mutation that causes the form of PRA known as GR\_PRA1, that the AHT offers a DNA test for.

The research we have carried out to identify the PRA mutation has revealed GR\_PRA2 accounts for approximately 15% of known PRA cases in the breed. In addition GR\_PRA2 has to date been identified in dogs from the USA, Canada, UK, Sweden and France.

### **DNA Test Results**

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 GR\_PRA2 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 GR\_PRA2 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 GR\_PRA2 mutation and will almost certainly develop PRA during their lifetime. The average age of diagnosis for dogs with GR\_PRA2 is 5 yo, although there is considerable variation within the breed.

#### **Advice**

Our research has demonstrated that the frequency of the GR\_PRA1 mutation in Golden Retrievers living in the UK is approximately 2%. 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 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](#).

