

PRA rcd4 (LOPRA) Some questions answered

(please read this alongside the AHT announcement)

I have been asked a number of questions on this subject, and the following answers try to throw light on the current situation.

1. What does it mean to be genetically affected but not yet clinically affected by PRA rcd4?

Unlike PRA rcd1 and CLAD, which can be seen in very young puppies, PRA rcd4 may not be visible to the owner or even to the vet or ophthalmologist until later in life. The dog is genetically affected from birth and a DNA test for PRA rcd4 will show this; however the clinical signs of deteriorating eyesight will not be present until sometime later in life and, in fact in a few cases, may never occur. The dog has the defective genes from birth although the clinical signs are not present and this must be understood when considering a breeding programme.

2. Explain the meaning of “homozygous for PRA rcd4”.

This is frequently referred to as having “two copies of the mutant gene” and thus being genetically affected.

In layman’s terms this means that the defective gene is **inherited from both parents**.

If the defective gene is inherited from only one parent the dog will be a “**carrier**” of the condition which means the defective gene can be passed to the offspring but this dog will never have this condition. This is typical of a recessive mutant gene and most of us are familiar with it in PRA rcd1 and CLAD.

3. Remind me what happens if an affected dog is mated to a clear.

AFFECTED to CLEAR >>>>>>>>>> 100% CARRIERS

AFFECTED to CARRIER >>>>>>>>>> 50% AFFECTED; 50% CARRIERS

CARRIER to CLEAR >>>>>>>>>>>> 50% CARRIERS; 50% CLEAR

CARRIER to CARRIER >>>>>>>>>>>> 25% AFFECTED; 50% CARRIERS; 25% CLEAR

4. How do we know there might be 30-40% of dogs in our breed that are carriers?

A random check was performed on a large number of DNA samples stored at the AHT and this provided the information. The large number of samples used by the AHT means that the proportion of carriers for that sample is likely to reflect the proportion throughout our breed.

5. Will we be told the individual results from this test run?

No. The AHT have permission to use the samples stored for research purposes i.e. in the development of a new test, and to provide a statistical analysis of the results but not to provide individual dog’s names or results.

The way forward is to test the dogs you own now, particularly your breeding stock, and to move forward from this.

The advice so far is to avoid producing genetically affected puppies – if you find you have an affected dog or a carrier with which you wish to breed only breed to a clear dog.

6. What do we know about another form of LOPRA that exists in the breed?

We know there is a third form of PRA in the breed as 3 dogs have been clinically identified as having PRA but their DNA shows that they do not have PRA rcd1 or PRA rcd4. It probably occurs at a younger age than PRA rcd4. It may be the cause of blindness in the younger dogs that also have the PRA rcd4 mutation. Further research will be needed to find the mutation if more cases are found.

7. How many dogs so far (July 2011) are homozygous for PRA rcd4?

We only know of 7, 6 of which have been named by their owners. I understand that there were very few in the research run but we have not been given further information on this. I do, however, have a personal story to tell as a result of this research run.

My experience has been with my old dog, Willow (Kirkavagh Karamita of Follidown), until now referred to in newsletters but not named because of the uncertainty involved in her condition at the time. During the research she was found to have two copies of (i.e. homozygous for) the PRA rcd-4 mutation **and** she was blind **and** she was 13 years old. This seemed to confirm the research programme but on examination by two highly respected ophthalmologists she was found **not** to have LOPRA. Her blindness was caused by typical problems of old age – some cataract and sclerosis of the lens. If she had lived longer she may have developed LOPRA but, very sadly, she died in April. (Incidentally, she coped very well in her familiar environment with her blindness but did need extra help and consideration because of her condition.)

Most of you will have read about her already but it provides an important case study and a good reason not to panic if the DNA test shows your dog to be homozygous for PRA rcd-4. Your dog may never go blind despite having the genetic mutation.

If you have any further questions, please email me and I will try to help.

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