PRA Update

DNA testing for PRA [rcd-4] began in March 2011 and since then steady progress has been made in reducing the amount of carriers and increasing the number of clears. It is the breeds goal to eventually eradicate this condition but not at the detriment of the breed.

I asked Dr Tom Lewis, Quantitative Geneticist at the Kennel Club for his thoughts on the PRA figures I have been collating. He says, *'..the bar chart you provide shows that there have been no affected individuals born since 2012. To my mind, this demonstrates the benefit of DNA tests-ensuring only puppies are born that will not suffer from the disease.*

However, as you correctly identify, we would also like to see a decline in the frequency of the disease causing mutation in the breed [i.e. eradicating the causal genetic variant]. I looked at figures for dogs born in the years with the pie charts in the document [2000,2009 and 2016]. From these it looks as though the frequency of the mutation is falling from 45% in 2000 to 32% in 2009 to 13% in 2016 born. There are some caveats and biases we should acknowledge [e.g. the 2000 sample may be biased towards dogs thought to be affected or carriers; and more recently we are biasing towards 'clear' animals in the sample by the presence of hereditary clear animals, however, this does

look as though the disease causing mutation is successfully being selected against.

From here on, I would recommend carrying on as you have been- testing and deriving hereditary clear status, and judiciously using clear and some carriers in breeding, all the while ensuring that no affected individuals are born. I would hope, in a few years' time, to see the frequency of the disease causing mutation has continued to fall and so see a proportional increase in the number of clear, and a decline in the number of carrier individuals'

Since the start of PRA, clinical eye tests have been recommended and this is still the case. It is a way to monitor the condition in DNA tested affected dogs and also is an important part of monitoring for other forms of PRA or any other emerging eye condition.

Dogs that have been identified as genetically affected with PRA rcd-4 may be coming to an age where they are starting to show clinical signs of the condition. This is to be expected and owners/breeders shouldn't be alarmed.

In 2016 two rcd-4 carriers showed clinical signs of PRA indicating there is another form of PRA in the breed. The AHT currently has five samples from Gordon setters exhibiting a different form to rcd-4. Two samples are from the UK, the other three are from the US. The first dog has had its DNA tested

against other breeds PRA DNA tests but didn't match. The other samples will be tested in June. The AHT's Give A Dog A Genome is looking at this other form of PRA in the Gordon Setter with a hope of accelerating research and getting a DNA test.

Currently the protocol is exactly the same as when rcd-4 was first discovered;

Clinical eye tests, particularly prior to breeding.

Any affecteds found should have a DNA sample sent to the AHT.

There are many forms of PRA and clinically they cannot be distinguished. They tend to be reported under the heading of GPRA [Generalised Progressive Retinal Atrophy]- it is only when the mutation is found that they are named. The age of onset is variable so this is not an effective method of deciding what type of PRA a dog has. PRA may be picked up clinically because an owner has noticed a problem or it may be picked up early through a routine eye test, in some cases before the dog has actually shown any signs of sight problems.

At the moment if a rcd-4 clear or carrier dog shows PRA, it is likely to be a different form and should be investigated. If a rcd-4 affected dog shows PRA it is mostly likely to be rcd-4 though another form cannot be ruled out yet.

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