

## ENGLISH SPRINGER SPANIEL CLUBS\*

**\*On behalf of:**

The English Springer Spaniel Club  
The English Springer Spaniel Club of Scotland  
The English Springer Spaniel Club of Wales  
The Lancs & Cheshire English Springer Spaniel Club  
The Southern English Springer Spaniel Society  
The Midland English Springer Spaniel Society  
The Northern English Springer Spaniel Society  
The South Western English Springer Spaniel Club

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### **UK Health Co-ordinators Statement to Breed Clubs** **Re DNA Test for PRA**

As everyone is no doubt aware, a worldwide statement was issued on 20<sup>th</sup> April 2007 by Dr. Cathryn Mellersh from the Animal Health Trust (AHT) and Dr. Gary Johnson from the University of Missouri USA, stating that a mutation has been identified that is a major risk factor for the development of PRA in English Springer Spaniels. They also announced that a DNA test for this mutation has been developed and is now available in both the USA and UK.

Following that announcement, a number of breeders, who had donated samples from clinically affected dogs and their close relatives to the original PRA research project at the AHT, were invited to request from them the genetic results for these dogs.

These breeders had, naturally, expected that their clinically affected dogs would be confirmed as genetically affected, but when some results showed this not to be the case (i.e. their dogs tested as either genetically clear or carriers), considerable confusion and alarm ensued concerning the validity of the DNA test.

It is the opinion of both Dr. Mellersh and Dr. Johnson, that, barring any errors in the testing procedures (possible but unlikely), these unexpected genetic results can be explained by either of the following:

- 1) In some rare instances, a dog can develop PRA as a result of carrying a single copy of the mutation (i.e. a genetic carrier), rather than two mutant copies as would be expected in affected dogs. This would be unusual, based on what is known, but cannot formally be excluded at this stage.
- 2) There is another form of PRA in the English Springer Spaniel caused by a different mutation in a different gene that, as yet, has not been identified. Multiple forms of the same eye disease occurring in the same breed are not unheard of, and it is entirely possible that some dogs can carry either or both mutations.

The original announcement made by Dr. Mellersh and Dr. Johnson, may, unintentionally, have given the impression that the new test was the whole answer with regard to PRA in the Breed. Whilst it certainly mentioned that a second form of PRA was a possibility, the figures quoted (which were based on statistics from the USA studies) suggested that this was rare. Unfortunately, with regard to being able to advise us on how the statistics for a second form of PRA might apply within the UK population, Dr. Mellersh was and is unable to give any estimates or make any predictions, as she only has a very limited number of 'affected' samples to work with, and these are currently nowhere near enough for her to make any valid judgement.

**The DNA test for PRA that has been made available is for one specific mutation in one specific gene (known as 'PRA Cord 1 Mutation' or RPGRIP1), and the test cannot, therefore, detect any other mutation that may be responsible for any other form of PRA. It does not, however, diminish its validity for this particular mutation, which is undoubtedly associated with PRA in English Springer Spaniels.**

**Statistics from the AHT show that approximately 20% of dogs have been found to be genetic carriers for the Cord 1 (RPGRIP1) mutation, with approximately 3% being genetically affected. Irrespective of whether a second form of PRA exists in the Breed, the prevalence of this mutation alone in the UK population is very significant.**

**The DNA test allows breeders to identify those dogs either carrying or affected by this mutation, and, through careful and sensible breeding over several generations, to eliminate it from their breeding. The fact that it is not the whole of the answer to PRA is, of course,**

**disappointing, but the fact that a DNA test is available for any form of PRA has to be a step forward.**

**This new test, as with any other, is simply another tool that breeders now have at their disposal should they wish to make the most of the information available to them. When making decisions on breeding, the results of any test should be taken in the overall context of the merits or weaknesses of each dog and the severity or consequences of the disease to which the test relates.**

With regard to PRA, it is the intention to carry out further research into both of the following areas:

- a) The identification of the mutation that causes a second form of PRA in the Breed. This will require samples from dogs that are diagnosed as clinically affected but their DNA test shows them not to be genetically affected for the Cord 1 (RPGRIP1) mutation.
- b) The identification of dogs that have been tested as genetically affected for the Cord 1 (RPGRIP1) mutation, but are not clinically affected. From a breeding point of view, it is the genetic status that counts, but Dr. Mellersh and Dr. Johnson have advised that other genes may influence the age of onset and rate of progression in dogs genetically affected for the Cord 1 (RPGRIP1) mutation, and further studies are required to explain this.

**Therefore, ESS owners and breeders, who wish to do so, should be encouraged to use the new DNA test for PRA in addition to continuing with clinical eye testing for all breeding stock.**

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