

## **IMPORTANT ANNOUNCEMENT TO THE ENGLISH SPRINGER SPANIEL WORLD WIDE COMMUNITY CONCERNING PROGRESSIVE RETINAL ATROPHY RESEARCH**

Dr Gary Johnson and Dr Cathryn Mellersh have identified a DNA mutation that is a major risk factor for development of Progressive Retinal Atrophy (PRA) in English Springer Spaniels.

- A DNA test is available for breeders, along with information about what the test can and cannot tell them.
- The percentage of English Springer Spaniels testing as *affected* or *carrier* for this mutation is very high.
- It is likely to take several generations to reduce the frequency of this mutation in the ESS population.
- Additional research, funded by the ESSFTA Foundation and the AKC Canine Health Foundation, has been initiated to help answer the questions that remain unexplained by the discovery of this mutation.
- Much more important information about this discovery can be found in the PRA section of the [www.CanineGeneticDiseases.net](http://www.CanineGeneticDiseases.net) website.

Dr Gary Johnson's Animal Molecular Genetics Laboratory at the University of Missouri-Columbia, in collaboration with Dr Cathryn Mellersh of the Animal Health Trust in England, has identified a mutation that is a major risk factor for development of Progressive Retinal Atrophy (PRA) in English Springer Spaniels (ESS). A DNA test for this mutation is now available for concerned ESS breeders to help them make breeding choices that can reduce the incidence of PRA in this breed. In the dogs tested during the research, 80% of the English Springer Spaniels tested as affected or carrier for this mutation, so it is important that breeders have a clear understanding of what this test can and cannot tell them. It is very likely to take several generations to reduce the frequency of this mutation in the ESS population. Additional research has been initiated to help answer the questions that remain unexplained by discovery of this mutation.

### **THE DISCOVERY**

Dr Mellersh recently published information on a mutation found to cause a recessive cone-rod form of PRA in Miniature Longhaired Dachshunds. In a limited survey, Dr. Mellersh also found the mutation to be present in ESS. Because of this, Masters student, Xuhua Chen, from Dr Johnson's laboratory tested over 1100 ESS DNA samples and found that dogs that inherited the mutation from both their sire and dam were approximately 20 times more likely to develop PRA compared to other ESS. Preliminary electroretinogram (ERG)

studies by Dr Kristina Narfstrom, Laboratory for Comparative Ophthalmology, University of Missouri-Columbia, suggest that ESS have a cone-rod form of PRA similar to that found in the Dachshunds.

It is important to note that there are a large number of dogs that have tested as genetically affected, but are reported as clinically normal by their owners. This is also similar to the situation in Miniature Longhaired Dachshunds. With the wide range of age of onset observed for PRA in ESS, it may be that many of these dogs will develop symptoms eventually. It is also possible that that these dogs have some loss of visual function that has not yet been detected by the owner.

### **GOOD NEWS – BAD NEWS**

The good news – a DNA test is now available. This test clearly identifies dogs that are clear (have 2 normal copies of the gene), those who are carriers (have one normal copy of the gene and one mutated copy of the gene), and those who are at much higher risk for developing PRA (have 2 mutated copies of the gene). Wise use of this test can reduce the incidence of dogs at risk for PRA in future generations.

The bad news – only 20% of the 1100-plus ESS's genotyped during the research tested as clear, or normal. Thirty-eight percent tested as carriers, and 42% tested as genetically affected. Obviously, eliminating all dogs testing as affected from breeding programs will have a major impact on the breed, and has the potential to devastate successful breeding programs. Reducing the incidence of dogs at risk for PRA while maintaining genetic diversity and positive qualities present in the breed is likely to be a slow process and will take several generations.

Information about the benefits of this test, testing forms and instructions, and suggestions for using the test to reduce the frequency of PRA in this breed is available online in the PRA section of [www.CanineGeneticDiseases.net](http://www.CanineGeneticDiseases.net) and at [www.aht.org.uk](http://www.aht.org.uk) .