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

- Dr Gary Johnson (UMC) and Dr Cathryn Mellersh (AHT) 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. We are awaiting confirmation from Dr Cathryn Mellersh at the AHT as to what the cost will be.

- The percentage of English Springer Spaniels testing as *affected* or *carrier* for this mutation is very high. (80% of the dogs tested in the USA during the research tested as affected or carrier for this mutation)

- It is likely to take several generations to reduce the frequency of this mutation in the ESS population.

- Additional research in the USA, 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.

- More additional important information about this discovery can be found at www.englishspringer.org and www.sesss.co.uk .

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 USA 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 ERG clinical 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 that 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 – In the USA only 20% of the 1100-plus ESS's genotyped during the research tested as clear or normal. 38% tested as carriers, and 42% tested as genetically affected. Should the same statistics follow in the UK/Europe, eliminating all dogs testing as affected from breeding programmes would have a major impact on the Breed, and would have the potential to devastate successful breeding programmes. 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.

The DNA test is accurate and valid in being able to determine the genetic status of each dog. However, it is not able to predict at what age a genetically affected dog may become clinically affected.

We are aware that the age at which dogs develop PRA can vary dramatically. Additional research is being carried out to help us understand why some genetically affected dogs develop PRA early and others later. This research is likely to take some time, and in the meantime, therefore, the AHT feels it is in the Breed's best interests to make the DNA test available now rather than wait 2 or 3 years until we understand the story completely. From experience with the Miniature Longhaired Dachshunds PRA, there is a wide range of variation, both in terms of clinical presentation and the degree of visual impairment that is associated with this mutation. Dogs that are DNA tested as being affected may themselves not develop the disease until relatively late in life, but it may well be possible for offspring of those dogs to display an earlier, more progressive form of PRA, depending on other genetic variants that they do or do not inherit.

PLEASE NOTE

For a detailed explanation of how this form of genetic inheritance is passed down, please refer to our document "Genetic Inheritance" at this Website.

All the UK ESS Breed Clubs will be consulted to agree a Code of Ethics and Guidance in order to help ESS breeders understand, assess and minimize the risks to their future breeding programmes.

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