

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

- Dr Gary Johnson from the University of Missouri in the USA (UMO) and Dr Cathryn Mellersh from the Animal Health Trust in the UK (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 has been developed and is now available in both the USA and the UK, along with information about what the test can and cannot tell them. In the UK, the test is available from the AHT – further details concerning the procedure and costs are given below under the section headed “Testing Your English Springer Spaniels”.
- 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.

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 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, it is felt that 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.

Considerations for English Springer Spaniel Breeders:

The mutation is a risk factor for the development of PRA in English Springer Spaniels: Most of the dogs that were tested in the USA as "affected" are considered to have normal eyesight by their owners. Some of these dogs may develop PRA as they get older; however, there are many examples of old English Springer Spaniels that DNA test "affected" but, have subtle, if any, visual impairment.

On the other hand, 95% of the English Springer Spaniels with clinically recognized PRA test "affected." Erroneous diagnoses or a second rare form of

PRA may account for the 5% of English Springer Spaniels with PRA that do not test “affected.”

The USA study implies that the likelihood of developing PRA is approximately 20 times higher for English Springer Spaniels testing “affected” than it is for other English Springer Spaniels. This is strong evidence that testing “affected” is a major risk factor for PRA in English Springer Spaniels and indicates that the prevalence of English Springer Spaniel PRA can be reduced by breeding programmes that select away from the mutant gene. This can be accomplished by giving highest preference to breeding stock that test “normal,” intermediate preference to dogs that test “carrier” and lowest preference to dogs that test “affected.”

Recommendations to English Springer Spaniel Breeders:

Although we believe that English Springer Spaniel breeders should make efforts to reduce PRA in future generations of their line, we also believe that if the mutation is so common in the Breed, overly aggressive elimination of dogs testing affected or carrier from breeding consideration could have an overall detrimental effect on the Breed and could devastate successful breeding programmes.

A realistic approach when considering which English Springer Spaniels to select for breeding would be to consider dogs with the mutation to have a fault just as lack of working ability, poor top line, or imperfect gait would be considered faults.

Dogs that test “affected” with two mutant copies of the PRA gene should be considered to have a worse fault than “carriers” with only one mutant copy. English Springer Spaniel breeders could then continue to do what conscientious breeders have always done: make their selections for breeding stock in light of all of the dogs’ good points and all of the dogs’ faults. Using this approach over several generations should substantially reduce the prevalence of PRA while continuing to maintain or improve those qualities that have made English Springer Spaniels so popular.

Ongoing Research:

One problem with the above approach is that the clinical consequences of testing “affected” are, as yet, poorly defined. Thus, it is hard for breeders of English Springer Spaniels to determine how much priority should be given to selecting away from the mutation. To better understand the clinical consequences, we will continue to assess clinical eye examinations (BVA/ECVO & AHT) and other relevant medical records of the dogs that are tested. When the results of these studies become available, they will be posted at this Website.

Testing your English Springer Spaniels:

- A DNA test for PRA in the English Springer Spaniel is now available at the Animal Health Trust.
- The AHT will require **cheek swabs** for the DNA test.
- The cost of each test is £50 (including VAT). This includes the cost of the cheek swab sampling kit. A 20% discount is offered if 20 or more samples are sent in together in a single batch for testing.
- Animal Health Trust **DNA Testing Form** can be downloaded from the AHT website at www.aht.org.uk. Kits for taking cheek swabs are available by phoning 08700 509144 or via email to swab.request@aht.org.uk.
- Further information can be obtained by emailing dna.testing@aht.org.uk.
- Samples should be sent together with a completed DNA Testing Form and payment for each sample to:

Genetic Services, Animal Health Trust,
Lanwades Park,
Kentford,
Newmarket,
Suffolk CB8 7UU.

- For most tests offered by the AHT, 100 – 200 samples are routinely handled per month and turnaround time is usually a few weeks. However, this may take longer during very busy periods.
- Owners of all samples that the AHT has actively used in the research that has led to the development of this DNA test will be entitled to the genetic results of those samples free of charge in an email. The AHT will make a charge of £5.00 if the owner would like a certificate.
For samples to have been used in the research, they must be from affected dogs, or closely related to affected dogs.
All the samples that have been sent previously to the AHT will not have all been used, if they do not fall into the above categories.

Dr Cathryn Mellersh
Dr Gary Johnson

The Animal Health Trust (UK)
University of Missouri (USA)

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 minimise the risks to their future breeding programmes, whilst still breeding for quality, type and temperament.

UK ESS Breed Clubs – Joint Health Co-Ordinators:

Lesley Bloomfield
Louise Scott

lesley@fernlin.free-online.co.uk
louise@goldcliffe1.freeserve.co.uk