Progressive Retinal Atrophy DNA Testing Frequently Asked Questions

What is Progressive Retinal Atrophy (PRA)?

PRA is an ongoing deterioration of the cells of the retina responsible for vision. Because of the progressive nature of the disease, this condition ultimately leads to complete blindness. In the English Springer Spaniel (ESS), the cells responsible for low light vision (the rod cells aka, "rods") deteriorate first, leading to diminished low light vision. Eventually the cells responsible for bright light vision (cone cells, aka "cones") deteriorate and bright light vision becomes diminished. Over time as these photoreceptors die off the dog eventually goes completely blind.

What is the difference between Early Onset and Late Onset PRA?

Originally it was thought that ESS affected with PRA would be completely blind by 3-5 years of age, when dogs are still relatively young. When our DNA cord1 test was discovered, we found that majority of the dogs that were DNA-affected with this mutation did not go blind by the expected age. Instead they were not losing vision until much later in life, at around 8-10 years of age, if at all. We were not aware that DNA-affected dogs could retain vision that long, so essentially we learned of a second form of PRA which we now term "Late Onset PRA". The original form of PRA is now termed "Early Onset PRA".

If my dog tests affected for the Cord1 Mutation, but remains visual, does that mean the test is wrong?

Because some dogs that have tested as "DNA cord1 affected" (DNA-affected) retain vision until old age, many breeders have concluded that the DNA cord1 test is not valid in the ESS. This is not a true statement. We need to look at this from a different perspective.

The early form of the PRA has been greatly diminished in the ESS due to diligent screening for the disease through annual AVCO (ECR, CERF) eye examinations on our breeding stock over the years. Now we know that we have a form of PRA that becomes a problem much later in the dog's life, is more insidious in its onset and is not always visibly apparent on the ACVO eye exam. We now believe that the reason we don't see vision loss or visible changes in the eye may be due to the fact that the dogs just don't live long enough to show signs of affliction. Perhaps if they lived to the age of 25 yrs, we would see complete blindness, but since their life expectancy is not that long, many never completely lose their vision.

How Can I Use the DNA cord1 Test Results In My Breeding Program?

You should use the test results in the same manner you would use the results of any other health screen we perform on our breeding stock. It is best to look at the pros and cons that any dog has to contribute to a particular breeding program and do an analysis of those pros and cons. If the pros outweigh the cons, then use the dog. The DNA test for PRA

allows us to use dogs of various genetic statuses in various ways to avoid eliminating the good traits that an individual may possess along with the PRA disease. Keep in mind that PRA is NOT a life threatening disease, so if your dog is affected, but not afflicted, you don't necessarily need to remove the dog from your breeding program, based solely on this one genetic disease.

Ideally we would all like to breed a clear individual to another clear individual, but at the present time, we do not have an abundance of clear individuals in the ESS. If you breed a clear dog to another clear dog, you will produce all clear offspring. If you bred a carrier dog to a clear dog, the offspring would be either clear or carrier. If you were to breed a carrier individual to another carrier, you would have the chance of producing clear offspring, carrier offspring as well as some affected individuals.

In an attempt to be able to retain some of the traits possessed by a DNA-affected dog the following are some possible scenarios that would allow you to go forward with an affected individual. Please remember, if you breed a DNA-affected animal to a clear animal, the worst you will produce in the resulting litter are carrier offspring. On the one hand (con), this increases the pool of carriers in the breed as a whole, but this would allow you to retain some of the exceptional qualities of your DNA-affected individual (pro), and ultimately move forward in your breeding program. If you then kept one of those carrier offspring and bred that to a clear individual, you will further decrease the number of DNA-carriers; by hopefully producing some clear (DNA-normal) individuals, as well as more carrier individuals. Another, yet less desirable, scenario is to breed a DNA-affected individual to a DNA-carrier individual; the catch in this scenario is that you could produce some affected puppies (con) as well as carriers in this type of breeding. The ideal approach in this scenario would be to keep one of the carrier offspring and move forward with that individual as described above. The most important thing to keep in mind is that the DNA test allows us to avoid eliminating some of the great dogs that may test DNA cord1 affected, and still continue to strive to eliminate the problem from our breeding programs. With this approach as we get more clear individuals to choose from, we would be able to breed to only to clear individuals and in the process decrease the numbers of carrier and affected individuals all together. So don't "throw the baby out with the bath water", use the information that the test provides for your breeding program wisely to move forward to completely eliminate the disease from your breeding program.

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