EAOD Research Project Update- September 2018

The ABCA Health & Education Foundation has received a progress report from Dr. Hannes Lohi's team studying EAOD, which says that they have essentially identified the causal gene and its likely causative variant. They need to complete some additional experiments to validate their conclusions prior to a peer-reviewed publication, but they hope to be able to submit their work for publication before the end of the year. That doesn't mean that the work will be published by then -- the peer review process can sometimes be lengthy -- but submission for publication is very important. It not only provides for evaluation of their work by other researchers, but also, once their results are published, it will be possible for any testing laboratory to develop and offer DNA tests for EAOD based on this research. Both the researchers and ABCA HEF agreed at the start that publication was essential, for the advancement of research and to permit competition to keep test prices lower.

In the meantime, based on the results they've achieved, the research team has begun the process of developing a gene test in collaboration with a very large, well-regarded testing company. The intent is to ensure that a test is available for veterinary diagnostic purposes and breeding decisions as soon as possible. The best current estimate for availability of the test is January 2019. If the test becomes commercially available from this company before other companies have been able to bring a test to market, there is a side benefit for us -- we will be able to get data that will best show the prevalence of EAOD in our dogs. Right now, a certain percentage of our dogs carries the EAOD mutation, but we have no way of knowing what that percentage is. It's the portion of the dogs who show up as Affected, Carrier or Normal when the test first becomes available that will give us this information. Later on, after the test has been on the market for awhile, these figures will gradually become less and less informative about the prevalence of the EAOD mutation, because more people will tend to test only suspect dogs, so the data will be skewed. Early on is when the sample of dogs being tested will be the most random, and will give us the best estimate of the true percentage of Carriers and Affecteds in our breed right now. That knowledge is very important in developing recommendations for breeding.

Dr. Lohi and his colleagues intend to continue and broaden their study to better understand this complex disease. Additional clinical studies will help in understanding its dimensions, including variations in age of onset and manifestations. For example, it is not yet certain that EAOD is 100% penetrant. There may be cases where dogs who carry two copies of the causative mutation do not become deaf. (This is similar to CEA. CEA has an autosomal recessive mode of inheritance, yet there are cases (often called "go normals") where a dog who carries two copies of the causative mutation, and therefore will pass that mutation on to its offspring, does not show symptoms of the disease. It's not yet known whether the same may be true of EAOD, or if so, how frequently this occurs.)

Dr. Lohi and his colleagues ended their report by thanking the ABCA Foundation for our "very helpful" and "much appreciated" support for this research. We in turn thank them for their hard work and the good results they have been able to achieve.