
EAOD Research Project - August 2016

The first priority of the ABCA Health & Education Foundation is to find the causal mutation(s) behind Early Adult Onset Deafness (EAOD) in the Border Collie, and to develop a mutation-based DNA test to identify dogs who are Affected by the disease, dogs who are Carriers, and dogs who are Normal.

Affected dogs begin to lose their hearing in early adulthood, most commonly between three and six years of age. Before that time, there is no way to distinguish a dog who will go deaf from this disease from one who won't, without a DNA test. Similarly, throughout the lifetime of a dog, there is no way to distinguish a dog who carries the disease and can pass it on to offspring from one who is genetically free of it, without a DNA test. This is a devastating disease for those who may have spent years training a pup, only to find that it will not be able to work during its most productive years because of deafness, and to realize that the dog may have been bred and passed on the disease genes to its progeny before there was any reason to suspect that the breeding could cause harm. Add to that the fact that EAOD has cropped up in some of the most talented and desirable working lines — in dogs with abilities that many want to perpetuate — and it's obvious that finding a definitive DNA test for this condition is a must.

Beginning back in 2007, the ABCA began supporting and encouraging research aimed at developing a DNA test for EAOD. Those efforts led to publication of a research paper in 2012 entitled "[Variation in Genes Related to Cochlear Biology Is Strongly Associated with Adult-Onset Deafness in Border Collies.](#)" However, they stalled out for long periods after that, leading to the formation of the HEF and a search for alternatives.

Our current initiative is a joint effort led by Dr. Hannes Lohi at the University of Helsinki and the Folkhalsan Research Center, working in conjunction with Dr. Jim Mickelson at the University of Minnesota. Dr. Lohi was one of the authors of the 2012 paper cited above, who pursued Border Collie deafness research independently thereafter, along with his other research interests, including epilepsy. Dr. Mickelson has performed research under grants from ABCA in the past, principally into Exercise-Induced Border Collie Collapse (BCC). Both Dr. Mickelson and Dr. Lohi are members of a large multi-national consortium of researchers who share data that includes hundreds of canine whole genome sequences. This will add power to the analysis of sequences and other samples we are able to obtain from the North American working Border Collie gene pool.

The study plan is comprehensive and well-designed, HEF has obtained some 400 DNA samples and more than 75 BAER tests for North American dogs as of the end of 2016, and we are hopeful that the project will lead to results within a year. To support it, we will need to acquire as many samples as possible, and BAER test a great number of dogs. We hope Border Collie owners will support us in our efforts. The long-term welfare of our breed is at stake.