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Identification of Mutation for Cerebellar Degeneration in the Old English Sheepdog

The mutation causing cerebellar degeneration (also known as cerebellar abiotrophy, cerebellar ataxia, cerebellar cortical degeneration, CA) has been identified in the laboratory of Dr. Natasha Olby at North Carolina State University.

Cerebellar degeneration was first reported in the Old English Sheepdog in the 1970s. It causes a progressive loss of coordination resulting in the hallmark ataxic gait characterized by dramatic overstepping, particularly obvious in the forelimbs. Onset of signs ranges from 6 months to 4 years of age and disease progression tends to be slow, occurring over several years. It is inherited as an autosomal recessive trait, meaning that affected dogs have to have 2 copies of the mutated gene to develop signs of the disease.

The mutation was identified following extensive mapping of the disease in families of Old English Sheepdogs. The same mutation was also found in Gordon Setters with the same disease, implying that this is an old mutation that has existed in these populations of dogs at a very low level until more recent times. The mutation has not been described previously, and to date has only been found in the Old English Sheepdog and the Gordon Setter.

The Veterinary Genetics Laboratory at North Carolina State University will be offering genetic testing for the mutation to owners and breeders in early October. The cost of testing will be \$51 per dog, and the test can be run on blood, cheek swabs or semen. Please see http://www.cvm.ncsu.edu/vhc/csds/vcgl/index.html for more details.

Genetic test results for the mutation will be normal, carrier (one copy of the mutated gene), or affected (two copies of the mutated gene). Now that a genetic test is available, quality carriers can be bred to normal-testing dogs, preventing any affected dogs from being produced. Quality normal-testing offspring should replace the carrier parent to diminish the frequency of the mutated gene in the breed. In this way, selection against cerebellar degeneration will not impact the genetic health or diversity of the breed.

We are grateful for the support given to our research by breeders and owners of Old English Sheepdogs who donated DNA from their dogs, and by our collaborators, Drs. Dahlia Nielsen and Alison Mottsinger Reif of the Bioinformatics Research Center, North Carolina State University, Dr. Jerold Bell of the Tufts Cummings School of Veterinary Medicine, Dr. Andrew Singleton of

the Neurogenetics Laboratory at NIH, Drs. Kerstin Lindblad-Toh and Noriko Tonomura of the Broad Institute and Drs Dennis O'Brien and Gary Johnson of the University of Missouri College of Veterinary Medicine. Funding was provided by the AKC Canine Health Foundation and by the Old English Sheepdog Club of America.