

Identification of Mutation for Cerebellar Degeneration in the Gordon Setter

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 has been documented in the Gordon Setter as an autosomal recessive inherited disorder since at least the 1960s. 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.

The mutation was identified following extensive mapping of cerebellar degeneration in Old English Sheepdogs. Testing of the mutation discovered in Old English Sheepdogs revealed that both breeds of dog have the same mutation. This implies that it 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 Sheep dog and the Gordon Setter.

Cerebellar degeneration has been seen in Gordon Setters worldwide, including the US, Canada, Europe, and Australia. It has been documented in both conformation and field lines. While the mutation is old and dispersed in the breed, we do not expect it to be present at a high frequency in the breed. Genetic testing for the mutation will allow an assessment of the gene's frequency in the breed.

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 defective gene), or affected (two copies of the defective gene). Now that a genetic test is available, no dog has to be eliminated from breeding due to cerebellar degeneration carrier status. Quality carriers should 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 defective gene in the breed. In this way, selection against cerebellar degeneration will not impact the genetic health or diversity of the breed.

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