



Cerebellar Ataxia

American Staffordshire Terrier

NCL-A Test

Cerebellar Ataxia

Cerebellar Ataxia is a neurological disorder. This disease is also called Cerebellar Cortical Degeneration or Neuronal Ceroid Lipofuscinosis (NCL).

The first signs of the disease usually appear between 3 and 5 years of age in affected dogs. More rarely, early or late-onset symptoms can appear between 1.5-3 years or after 5 years. Symptoms are loss of balance, difficulty cornering, and falling when shaking their head. When they became unable to walk without falling repeatedly, owners usually make the difficult choice to euthanize. The clinical diagnosis is realized by excluding others diseases and confirmed by a MRI (Magnetic Resonance Imaging).

Expression and mode of inheritance

Cerebellar Ataxia is inherited as an autosomal recessive trait. This means a dog must have 2 defective copies (one from each parent) of the mutated gene to be affected by the disease. The DNA test provides an answer among three possible situations:

DNA test result	The dog is	Genetic status	Will develop the disease ?	Will transmit the genetic anomaly?
Normal homozygous	Clear	2 normal copies of the implicated gene	NO	NO
Heterozygous	Carrier	1 normal copy and 1 defective copy of the implicated gene	NO	YES, statistically to 50% of its progeny
Mutated homozygous	Affected	2 defective copies of the implicated gene	YES	YES, to 100% of its progeny

The DNA test, a reliable and validated test

Identification of the implicated gene and validation of the test resulted from collaborative research efforts between the laboratory of Molecular and Cellular Genetics (Dr Marie Abitbol, Dr Laurent Tiret) and the laboratory of Neurobiology (Dr Stéphane Blot) from the Alfort School of Veterinary Medicine (ENVA), as well as the CNRS-University of Rennes and ANTAGENE.

The technology underlying the test is patented world-wide by INRA and ENVA. ANTAGENE has an exclusive international license to provide this test.

Pilot studies achieved during the development of the test revealed a high frequency of carrier dogs in the french population of AmStaff (30%). In addition, collaboration with Dr Natasha Olby (North Carolina State University, Raleigh) confirmed that american dogs carry the same disease-causing mutation as the one identified in french dogs.

The DNA test is reliable, easy to do from a cheek swab (cytobrush) that can be done at any age as soon as the animal is identified (by tattoo or chip). Once validated by the test, the genetic status of the dog will not change throughout his/her life.

Early DNA screening allows for:

- detection of affected dogs before the first symptoms,
- easy identification of this disease before considering other time and money-consuming exams,
- aide in selecting sires and dams to be bred,
- avoiding selling dogs who will eventually become affected,
- controlling the dissemination of the genetic defect in the breed.

Ordering

Kits for sampling have to be ordered by email. They will be mailed to your postal address. Directions for sampling are included and must be authenticated by a veterinarian. A certificate with genetic results will be mailed to the owner.

For more information on ordering kits, our diagnosis services, and current genetic research on dogs and cats, please consult our web site at www.antagene.com

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