

Cerebellar ataxia in the Italian Spinone

Cerebellar ataxia (CA) in the Italian spinone is a serious neurological disease. In affected dogs the cerebellum, which is the part of the brain responsible for the coordination of motor movement in the body, becomes diseased. Affected dogs are born normal, but clinical signs, which include the development of an unsteady gait, loss of coordination and poor balance usually appear in the first weeks or months of life. The disease is progressive; symptoms worsen during the first year of the dog's life such that most dogs are euthanased before they are 1 year old. There is no treatment for the disease, which has an autosomal recessive mode of inheritance.

The genetic mutation that is responsible for the condition probably occurred spontaneously in a single dog but once in the population has been inherited from generation to generation like any other gene. The disorder shows an autosomal recessive mode of inheritance: a dog has to inherit two copies of the defective gene (one from each parent) for it to be affected by the disease. Individuals with one copy of the defective gene and one copy of the normal gene are called carriers and show no clinical signs but can pass the defective gene onto their offspring. When two apparently healthy carriers are crossed, 25% (on average) of the offspring will be affected by the disease, 25% will be clear and the remaining 50% will be carriers.

The region of the genome that contains the causal mutation has recently been identified at the Animal Health Trust. Using the information gained from this research, we have developed a linkage-based DNA test for the disease which we estimate will give an accurate result for between **95% and 98%** of dogs tested. This test will detect those dogs which are carriers of the CA mutation, and those that are clear of the mutation.

Breeders will be sent results identifying their dog as belonging to one of three categories:

CLEAR: This DNA test detects markers close to the gene responsible for CA - our research has indicated that the test is 95-98% accurate. CLEAR dogs have 2 copies of

the normal gene and will neither develop cerebellar ataxia, nor pass a cerebellar ataxia gene to their offspring.

CARRIER: This DNA test detects markers close to the gene responsible for CA - our research has indicated that the test is 95-98% accurate. CARRIER dogs have one copy of the normal gene and one copy of the mutant gene that causes cerebellar ataxia. They will not develop cerebellar ataxia but will, if bred from, pass on the cerebellar ataxia gene to, on average, 50% of its offspring..

AFFECTED: This DNA test detects markers close to the gene responsible for CA - our research has indicated that the test is 95-98% accurate. AFFECTED dogs have 2 copies of the mutant gene that causes cerebellar ataxia and will develop the disease..

Carriers can still be bred to tested clear dogs. On average, 50% of such a litter will be clear and 50% carriers; there can be no affecteds produced from such a mating. Pups which will be used for breeding can themselves be DNA tested to determine whether they are clear or carrier.

Tests can be ordered through our Webshop <http://www.ahtdnatesting.co.uk>.