FACULTY OF HEALTH SCIENCES
UNIVERSITY OF COPENHAGEN

Facts about DNA test for Polyneuropathy in Alaskan Malamut

DATO 28 MARCH 2012

The disease

The disease involves a defect in the transmission of impulses from nerves to muscles. Clinical symptoms are usually detected at the age of 8 to 14 month. Initially clinical signs are subtle and characterized by slowly progressive exercise intolerance. Many dogs also express laryngeal paresis. As the disease progresses gait abnormalities and muscle atrophy is seen.

Heredity

Polyneuropathy in Alaskan Malamut follows a simple autosomal recessive inheritance. Autosomal means that the disease is not sex-linked and recessive means that the disease will only appear if a puppy receives the defective gene form both parents. All genes come in two copies and a disease is called recessive when a healthy gene can mask the presence of a defective gene. A dog that has one normal and one defective copy of the gene is called a carrier and it will not show signs of the disease. Only when both copies of the gene are defective the disease will show up - in this case as muscle fatigue.

The mutation

Polyneuropathy in Alaskan Malamut is comparable to an illness in humans called CMT4 (Charcot-Marie-Tooth Type 4) and to polyneuropathy in Greyhounds. The cause of these polyneuropaties is the presence of mutations in the *NDRG1*. We have shown that it is also a mutation in the *NDRG1* gene that causes Polyneuropathy in Alaskan Malamut. Based on this information a diagnostic test has been established. Thus, it is now possible to get your dog tested for the mutation using a DNA test.

The test and results

The test is performed on DNA extracted from EDTA stabilized blood or from DNA extracted from cheek swaps.

A certificate will be sent to the dog owner reporting on the genetic diagnosis of the dog:

- Homozygous normal
- Heterozygous carrier
- Homozygote sick

Homozygous normal means that the dog has two normal copies of the gene - so no mutations in the *NDRG1* gene. A dog with this result can be used for breeding without reservation.

Heterozygous carrier means that the dog has one normal copy of the gene and one defective copy. The dog is clinically completely healthy, but it will pass the mutation to half its offspring. As long as the dog is only mated to homozygous normal dogs it will not produce sick offspring. Heterozygote carriers can therefore be used in breeding, but only to homozygous normal breeding partners.

Homozygous sick means that the dog has two defective copies of the gene. These dogs should not be used for breeding.