

## **Genetic research in Alaskan Malamutes**

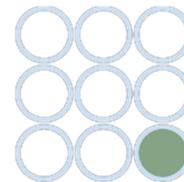
The Canine Genetics Research Group led by Professor Hannes Lohi at the University of Helsinki, Finland has been collecting DNA-samples from Alaskan Malamutes for chondrodysplasia and hereditary cataract research. The cataract research is done in collaborations with Dr. Cathryn Mellersh at the Animal Health Trust, UK. The aim of the research is to study the genetics of both diseases and to find the causative gene mutations. Identification of the causative mutations would enable the development of genetic tests that could be used for breeding purposes.

### **CHONDRODYSPLASIA (CHD)**

Alaskan Malamutes suffer from a genetic skeletal condition referred to as chondrodysplasia or dwarfism. The appearance of affected Malamutes is characterized by shorter stature and bowed front legs. The condition is thought to have an autosomal recessive inheritance pattern, which means that an affected dog has to inherit the causative mutation from both parents. At the moment, altogether ~250 Malamute samples are stored at our DNA bank, and 37 of these have been collected specifically for the chondrodysplasia study, including 10 affected dogs. In our genetic study, we will use two complementary research methods in order to discover the causative mutation: exome sequencing and genome-wide association mapping. We have already sequenced the exomes (i.e. protein coding regions of the genome) of two affected dogs and one known carrier. At the moment, we are analyzing a subset of the identified sequence variants in larger sample cohorts. The screening of 28 variants did not give positive results and we are currently screening an additional set of 27 variants. The next step in the study is to perform marker-based association mapping and to combine the sequence data with the results of the genome-wide mapping. Any new affected dogs would be highly welcome to participate in the study to expedite our gene discovery.

### **HEREDITARY CATARACT**

The first genetic study included affected and healthy Malamutes as well as Siberian Huskies and Samoyeds. Affected dogs had been diagnosed with bilateral posterior polar subcapsular cataract that is the more common form of cataract in these breeds. The average age of onset is about 3 years. The hypothesis was that the three breeds may share the same genetic risk for cataract. Based on our preliminary results a chromosomal region in one of the canine chromosomes is associated with cataract. The region includes several different genes which are currently being studied for mutations. To find the causative gene(s) the researchers need more samples from both cataract affected dogs and dogs that have been eye examined healthy.



## HOW TO PARTICIPATE

The samples are collected as blood samples (1-3 ml in EDTA tube). If blood sample is impossible to obtain, swab samples can be sent as well. A sample form should be filled of every dog and sent with the samples. Include a copy of the dog's pedigree and a copy of the eye examination report if the dog's eyes have been examined. Please contact the researchers for further, detailed instructions how to participate and how to send the samples.

**The research group would like to thank all dog owners who already have submitted samples and information from their dogs to the research.**

Further information [www.koirangeenit.fi/in-english/](http://www.koirangeenit.fi/in-english/) or contact:

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