STORAGE DISEASE IN LAGOTTO ROMAGNOLOS – THE RESULT OF GENETIC RESEARCH

DEAR LAGOTTO ROMAGNOLO OWNER,

You have donated a sample from your Lagotto to the Canine Genetics Research Group at the University of Helsinki, Finland. Your dog's sample has now been tested for the storage disease mutation, and here we report the result:

<u>DOG'S RESULT: DELMINA LIR08/78080 IS normal</u>

STORAGE DISEASE IN LAGOTTOS

The genetic and clinical studies concerning the Lagotto storage disease have been performed as a collaboration between the Universities of Helsinki and Bern. Altogether 1300 Lagottos were initially tested as a part of the research. Out of these dogs, 11 % were carriers of the recessive mutation and 1 % were affected. The Lagotto storage disorder is characterized by movement incoordination, which is seen for instance as a staggering gait. Some affected dogs have also suffered from involuntary eye movements (nystagmus) and behavioral changes, such as aggression. The onset of symptoms has varied from 4 months to 4 years. The condition is progressive and affected dogs deteriorate to the point where they need to be euthanized. On the pathological level, storage material accumulates to the affected dogs' cerebellar neurons and other cells, causing dysfunction and cell death.

INTERPRETATION OF RESULTS

The results of our genetic study indicate that the Lagotto storage disorder is inherited through a single gene, in an autosomal recessive manner. In recessive disorders, the affected dogs have inherited two copies of a disease-causing gene mutation. All dogs inherit one copy of each gene from their dam and one copy from their sire. Those Lagottos that receive a **NORMAL** result have inherited two normal copies of the disease gene. Their risk of being affected is minimal and they do not pass the mutation down to their offspring. **CARRIER** dogs have inherited one normal copy and one with the mutation. Their risk of being affected is also minimal but they pass the mutation down to approximately half of their offspring. If a carrier dog is used for breeding, it should be mated with dog that has been tested as normal. In carrier/carrier-matings, the resulting offspring can be either affected, carrier or normal, whereas in carrier/normal-matings, only carrier or normal dogs are born. **AFFECTED** dogs have inherited the mutation from both parents, and so they have two copies of the mutation. Their risk of developing the disease is very high. It is not recommended to use affected dogs for breeding.

OTHER RESEARCH IN LAGOTTOS

In addition to the storage disorder, progressive cerebellar abiotrophy (CA) has been reported to occur in the Lagotto Romagnolo breed. The clinical signs and age of onset can overlap between the different disorders, and therefore it is not possible to make a diagnosis based just on the dog's symptoms. The genetic cause of the cerebellar abiotrophy has not been identified yet but our ongoing research is aiming to shed light to this. In addition, Lagotto samples have been collected to an adult-onset epilepsy study, and different behavioral studies. You can update your dog's health information and your own contact information through this electronic form: https://elomake.helsinki.fi/lomakkeet/31849/lomake.html?rinnakkaislomake=english. Up-to-date

health information is highly important for the different research project. Please let us also know if your dog is still healthy. The results of the storage disease study will be published in a scientific article, after which commercial test laboratories can offer a gene test.

ABOUT DNA-TESTING

A genetic test is reliable and repeatable. However, in all DNA-testing there is a slight margin of error that could be introduced for instance by the quality of the sample, dysfunction in the test method or errors in the sample handling process. An unexpected result could also be caused by an incorrect pedigree. Possible error sources have been eliminated in the best possible way. If any ambiguity rises regarding the result, the research group commits to clarifying the matter as far as possible.

Best regards,

The Canine Genetics Research Group

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