GENOMIA
genetic laboratory

Result certificate #036677:

Detection of g.9459_9460ins236 mutation in PLPLA gene causing CNM in Labrador Retrievers by PCR

Sample
Sample: 13-20220
Name: Kowalski Mio at BellissimaNera
Breed: Labrador Retriever
Microchip: 941000015239225
Date of birth: 16.04.2013
Sex: male
Date received: 26.07.2013
Sample type: buccal swab

Customer
Alena Šiplová
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Result: Mutation was not detected (N/N)

Explanation

Presence or absence of g.9459_9460ins236 mutation in PLPLA gene causing CNM (Centronuclear Myopathy or also HMLR (Hereditary Myopathy in Labrador Retrievers)) in Labrador Retrievers was tested. CNM is a genetically conditioned defect of muscle fibres development. The disease manifests in two weeks of puppies age, muscle atrophy gradually affects movement and swallowing muscles. Affected individuals die during the first weeks or months of life.

Mutation that causes CNM is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P (positive / positive) genotype only. The dogs with N/P (negative / positive) genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP86

Report date: 31.07.2013
Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

Dajbychová

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