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Result certificate #091764

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

Sample
Sample: 17-07916
Name: Silver Skydiver Advocate
Breed: Labrador Retriever
Microchip: 804 098 100 099 956
Reg. number: ČLP/LR/32361
Date of birth: 18.10.2016
Sex: male
Date received: 28.03.2017
Sample type: buccal swab

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

Legend: N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

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 genotype only. The dogs with N/P genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

Method: SOP176-CNM, ASA-PCR

Report date: 05.04.2017
Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

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