

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic**Sample:**

Sample: 21-12345

Date received: 01.02.2021

Sample type: blood

Information provided by the customer

Name: Lassie DEMO**Breed:** Plemeno

Tattoo number: 1392013

Microchip: 123 456 789 012 345

Reg. number: REGQ12345

Date of birth: 1.1.2020

Sex: female

Date of sampling: 01.02.2021

The identity of the animal has been checked.

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: SOPAgriseq_canine, ngs

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager

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