

Detection of c.1955_1958+6del mutation in ARHGEF10 gene causing LPN1 in Leonbergers and Saint Bernards

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 c.1955_1958+6del mutation in ARHGEF10 gene causing polyneuropathy LPN1 in Leonbergers and Saint Bernards was tested. Polyneuropathy is characterized by non-traumatic involvement of peripheral nerves. The hereditary variant of this disease is severe, chronic, and progressive. Clinical signs appear at a young age, they include gait abnormalities, muscle atrophy, particularly of the pelvic limbs, and general weakness. In affected dogs, problems with breathing such as whistling, or inspiratory stridor (due to laryngeal paralysis) and dyspnoea are also common.

Mutation that causes LPN1 is inherited probably autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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

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Approved by: Mgr. Martina Šafrová, Laboratory Manager



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