GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.1107_1108delAG mutation in GJA9 gene causing LPN2 in Leonbergers

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)

Explanation

Presence or absence of c.1107_1108delAG mutation in GJA9 gene causing polyneuropathy LPN2 in Leonbergers 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 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 LPN2 is inherited incompletely dominant which means that one copy of the mutated gene inherited from one parent is enough to show symptoms of the disease. However, an affected homozygote (P/P) will differ in severity from a heterozygote (N/P).

Method: SOPAgriseq_canine, ngs, accredited method

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

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