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Result certificate #012345

Detection of c.1668delC mutation in GLB1 gene causing GM1 in Shiba-Inu

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.1668delC mutation in GLB1 gene causing Gangliosidosis type 1 (GM1) in Shiba-Inu was tested. GM1 is a lysosomal storage disorder that causes accumulation of the ganglioside-glycolipid in various body tissues, especially in the brain and the nervous system and in other internal organs. The disease is characterized by progressive neuromuscular dysfunction, disturbed growth, ataxia, motor disorders and head tremors. With the progress of the symptoms the affected dogs are not able to eat and to drink and later, the dog is unable to stand on its feet. The worsening of the health condition results in the death of the dog or in an indicated euthanasia.

Very similar clinical manifestations has a type 2 gangliosidosis (GM2), which has a different genetic cause and is not excluded by this test.

Mutation that causes GM1 in Shiba-Inu is inherited 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
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
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