

## 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, accredited method

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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