

**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.618\_620delCCT mutation in HEXB gene causing Gangliosidosis type 2 (GM2) in Shiba-Inu was tested. GM2 is a lysosomal storage disorder that causes accumulation of GM2 ganglioside-glycolipid in various body tissues due to deficiency of the hexosaminidase enzyme. The symptoms of GM2 include small growth of affected puppies, ataxia, and head tremor. The first clinical signs occur at early age of the puppies; however, they become worse as the disease progresses. Later, there can be observed poor coordination of movements making walking almost impossible, general tremor, the dog is not able to eat, and its health condition leads to death or an indicated euthanasia.

Very similar clinical manifestations has a type 1 gangliosidosis (GM1), which has a different genetic cause and is not excluded by this test.

Mutation that causes GM2 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: SOP171-GM1\_GM2, fragment analysis

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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