GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.618_620delCCT mutation in HEXB gene causing GM2 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.

SAMPLE

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: SOP188-MPS-canine, MPS, 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

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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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