GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.1360_1373dup mutation in ITGA2B gene causing thrombasthenia in Great Pyrenees

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.1360_1373dup mutation in ITGA2B gene causing thrombasthenia in Great Pyrenees was tested. Glanzmann thrombasthenia is a blood clotting disorder in which platelet function is impaired. Typical signs are small dots of blood on the skin and mucous membranes (petechiae) and increased bleeding during injuries or surgery. Other signs may include frequent nose bleeds or the presence of blood in the stool.

Mutation that causes thrombasthenia 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

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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