GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.735delT VWF mutation in VWF gene causing vWD type III in Shetland Sheepdogs

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 detected in heterozygous status (N/P)

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 mutation c.735delT in VWF gene causing vWD type III (von Willebrand disease type III) in Shetland sheepdogs was tested. VWD type III is the most severe form of vWDs, which leads to very strong life-threatening bleeding. VWD type III is characterized by total absence of von Willebrand factor (vWF). VWD III occurs in breeds Shetland Sheepdogs, Scottish Terrier and Kooikerhondje.

Mutation c.735delT in VWF gene is inherited as an autosomal recessive trait. That means the vWD typ III affects dogs with P/P genotype only. The dogs with N/P genotype are considered carriers of the disease (heterozygotes). Carriers have lower level of vWF; their homeostasis is normal. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 50 % N/P and 25 % P/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

verified and certified document
GENOMIA genetic laboratory

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