GENOMIA
genetic laboratory

Result certificate #218646

Detection of c.7437G>A mutation in exon 43 of VWF gene causing vWD type I

Customer: Dušan Kollárik, Belá 33, 03811 Belá-Dulice, Slovak Republic
Sample:
Sample: 23-01202
Date received: 23.01.2023
Sample type: buccal swab

Information provided by the customer
Name: GIANNA Bella Aurea
Breed: Golden Retriever
Microchip: 941 000 023 870 583
Reg. number: SPKP 3603/21
Date of birth: 15.5.2019
Sex: female
Date of sampling: 19.01.2023
The identity of the animal has been checked by MVDr. Lenka Blahušiaková, KVL 0814

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.7437G>A mutation in exon 43 of VWF gene causing vWD type I was tested. This mutation causes deficiency or failure of VWF (von Willebrand factor) which is called von Willebrand disease type I (vWD I). VWD manifests as bleeding which is most apparent in tissues having high blood flow shear in narrow vessels. VWD manifests oneself as a tendency to bleeding from skin and tissues.

VWD type I is the most often and simultaneously the least serious form of mammalian vWD. The disease is characterised by low plasma vWF concentration and normal vWF protein structure. VWD type I occurs, for example, in dog breeds Bernese Mountain Dog, Doberman Pinscher, Manchester terrier, Welsh Corgi Pembroke, all Poodles, Labradoodle, Goldendoodle.

Mutation c.7437G>A that causes VWDI is inherited as an autosomal recessive trait. That means the 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_TD, ngs

Date of issue: 03.02.2023
Date of testing: 23.01.2023 - 03.02.2023
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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