GENOMIA
genetic laboratory

Result certificate #254279

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

Customer: Lenka Orzelová, Bohumínská 437/42, 710 00 Slezská Ostrava, Czech Republic
Sample:
Sample: 24-40118
Date received: 30.01.2024
Sample type: buccal swab

Information provided by the customer
Name: Carramia Royal Glow
Breed: Poodle Standard
Microchip: 941000027192388
Reg. number: CMKU/P/23254/22
Date of birth: 17.7.2022
Sex: female
Date of sampling: 27.01.2024
The identity of the animal has been checked by Ing. Nikola Eretová, Genomia s.r.o.

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, ngs

Date of issue: 12.02.2024
Date of testing: 30.01.2024 - 12.02.2024
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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