GENOMIA
genetic laboratory

Result certificate #027678:

Detection of c.7437G>A mutation in exon 43 of VWF gene causing vWD type I in several dog breeds by PCR-RFLP

Sample
Sample: 12-33822
Name: Calypso Čertovo kopýtko
Breed: německý pinč
Microchip: 900088000245677
Date of birth: 14.11.2010
Sex: female
Date received: 10.12.2012
Sample type: buccal swab

Customer
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Result: Mutation was not detected (N/N)

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 characteristic 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.

VWD type I 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP51, accredited method

Report date: 17.12.2012
Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

Dajbychová