

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 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, accredited method

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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