GENOMIA
genetic laboratory

Result certificate #301457

Detection of c.634_635insN[76] mutation in G6PC gene causing GSD1a in German Pinschers

Customer: Marek Kučera, Na Betonce 1361/3, 15300 Praha 16- Radotín, Czech Republic
Sample:
Sample: 21-42684
Date received: 06.10.2021
Sample type: buccal swab

Information provided by the customer
Name: Naleks Treasure Uilsiya
Breed: German Pinscher
Microchip: 643 093 333 008 521
Reg. number: CMKU/NP/2387/-21/20
Date of birth: 10.09.2020
Sex: female
Date of sampling: 02.10.2021
The identity of the animal has been checked by Ing. Jana Kůsová, 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.634_635insN[76] mutation in G6PC gene causing Glycogenosis (GSD1a) in the German Pinschers was tested. Glycogenosis is a severe disorder of glycogen metabolism characterized by accumulation of glycogen, especially in the liver. Symptoms include severe liver enlargement, general failure to thrive, lethargy, coma and death.

Mutation that causes GSD1a 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: SOP176-GSD1, ASA-PCR

Date of issue: 13.06.2025
Date of testing: 04.06.2025 - 13.06.2025
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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