GENOMIA
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Result certificate #223066

Detection of c.2237G>A mutation in GAA gene causing Glycogenosis in Scandinavian dog breeds

Customer: Aneta Pániková, Pribinova 3, 95301 Zlaté Moravce, Slovak Republic
Sample:
Sample: 23-05107
Date received: 03.03.2023
Sample type: buccal swab

Information provided by the customer
Name: Bessi Bienca Rosse
Breed: Finnish Lapphund (Suomenlapinkoira)
Microchip: 941 000 026 790 584
Reg. number: SPKP50
Date of birth: 08.11.2021
Sex: female
Date of sampling: 28.02.2023

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.2237G>A mutation in GAA gene causing Glycogenosis (GSDII) in Finnish Lapphund and Swedish Lapphund and the Lapponian herder was tested. The disease is caused by deficiency of a specific enzyme alpha-glycosidase active in lysosomes which is needed to breakdown glycogen to glucose in lysosomes. The clinical signs include progressive muscular weakness, vomiting caused by oesophageal dilatation, heart disease, myocardial hypotrophy and condition loss.

Mutation that causes GSDII is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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: SOP172-GSDII, direct DNA sequencing

Date of issue: 28.03.2023
Date of testing: 03.03.2023 - 28.03.2023
Approved by: Ing. Nikola Eretová, Analyst

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