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Result certificate #200446

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: 22-08802
Date received: 07.04.2022
Sample type: buccal swab

Information provided by the customer
Name: AKIRA JARVI Mały Wiedeń
Breed: Finnish Lapphund (Suomenlapinkoira)
Microchip: 616 093 901 138 210
Reg. number: SPKP39
Date of birth: 25.07.2020
Sex: male

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: 12.04.2022
Date of testing: 07.04.2022 - 12.04.2022
Approved by: Mgr. Lucie Magoči, Analyst

verified and certified document GENOMIA genetic laboratory

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999