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Result certificate #148021

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

Sample
Sample: 20-10601
Name: Bienca Kabadula
Breed: Finnish Lapphund (Suomenlapinkoira)
Microchip: 941 000 022 618 478
Date of birth: 30.04.20218
Sex: female
Date received: 30.04.2020
Sample type: buccal swab

Customer
Aneta Pániková
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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

Report date: 06.05.2020
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

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