GENOMIA
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Result certificate #157535

Detection of c.1623delG mutation in ATP13A2 gene causing NCL in Tibetan Terriers

Customer: Ellen Lind, Lundamovegen 307, 7232 Lundamo, Norway
Sample:
Sample: 20-24732
Date received: 06.10.2020
Sample type: buccal swab

Information provided by the customer
Name: Karamain Amritaya
Breed: Tibetan Terrier
Reg. number: FI43096/18
Date of birth: 19.07.2018
Sex: female
Date of sampling: 24.09.2020

Result: Mutation was detected in heterozygous status (N/P)

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.1623delG mutation in ATP13A2 gene causing Neuronal Ceroid Lipofuscinosis (NCL) in Tibetan Terriers was tested.

Mutation causing NCL in Tibetan Terriers 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP173-NCL-TT, PCR-RFLP

Date of issue: 19.10.2020
Date of testing: 06.10.2020 - 19.10.2020
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

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