GENOMIA
genetic laboratory

Result certificate #012345

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

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic
Sample:
Sample: 08-12345
Date received: 25.11.2008
Sample type: blood

Information provided by the customer
Name: Lassie DEMO
Breed: ---
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 31.12.1909
Sex: female
Date of sampling: 25.11.2008
The identity of the animal has been checked.

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.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: 30.11.2008
Date of testing: 25.11.2008 - 30.11.2008
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

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