

## 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: 21-12345 Date received: 01.02.2021 Sample type: blood

## Information provided by the customer Name: Lassie DEMO Breed: Plemeno

Tattoo number: 1392013 Microchip: 123 456 789 012 345 Reg. number: REGQ12345 Date of birth: 1.1.2020 Sex: female Date of sampling: 01.02.2021 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: SOPAgriseq\_canine, ngs, accredited method

Date of issue: 06.02.2021 Date of testing: 01.02.2021 - 06.02.2021 Approved by: Mgr. Martina Šafrová, Laboratory Manager



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