

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 mutation c.491T>C in CLN8 gene causing Neuronal Ceroid Lipofuscinosis (NCL) in English Setters was tested. This mutation leads to p.L164P substitution in CLN8 protein. NCL is a neurodegenerative disorder that is characteristic by accumulation of lipopigments (coroid and lipofuscin) in the lysosomes. The beginning and clinical course of the disease are very individual. The rate of neurodegeneration increases together with the age. Mental abnormalities and ataxia usually develop in all affected dogs. Increased restlessness, aggression, hallucinations, hyperactivity and epileptic attacks can be observed as well. Accompanying symptom is damaged retina due to lipopigment storage. Affected individuals rarely survive more than 28th month of age.

Mutation that causes NCL in English Setters 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: SOPAgriseq_canine, ngs

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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