

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.934_935delAG mutation in CLN5 gene causing Neuronal Ceroid Lipofuscinosis (NCL) in Golden Retrievers was tested. NCL is manifested by accumulation of lipopigments (coroid and lipofuscin) in the lysosomes. The clinical symptoms are progressive neurological signs including disorientation, worsening of motor functions, anxiety, aggression, seizures and problems with food intake. Usually visual impairment and loss of vision occur as well. The onset of the disease and its clinical course vary substantially between breeds. The first signs occur most often after 15 month of age. The degree of neurodegeneration increases with the age and all affected dogs develop psychological abnormalities and spasms. Changes in gait and posture – stumbling, leg stiffness, tremor - can be observed as well.

Mutation that causes NCL in Golden Retrievers 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: SOP188-MPS-canine, MPS

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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