

**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.736\_737insC mutation in PPT1 gene causing Neuronal ceroid lipofuscinosis type 1 (NCL1) in Dachshunds was tested. The characteristic feature of the NCL disease is excessive accumulation of waste compounds of lipid character (ceroid and lipofuscin) in the cells of the nervous system. The presence of a high content of lipofuscin and its increasing pressure affects and destroys the nerve cells in the cortex and the cerebellum and the retina cells. Symptoms in this type of NCL begin around nine months of age and include behavioural changes, nervousness, disorientation, ataxia, weakness, kyphosis, stiffness of gait, uncontrollable rhythmic head movements and visual disturbances.

Mutation that causes NCL1 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, accredited method

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549

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Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at [www.genomia.cz](http://www.genomia.cz)

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