

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)

Explanation

Presence or absence of c.125C>T mutation in CNP gene causing Lysosomal storage disease (LSD) in Weimaraners was tested. The disease is characterized by the accumulation of autofluorescent inclusions in lysosomes in the cerebral cortex, cerebellum, optic nerve, and cardiac muscle.

Inheritance is described as autosomal, incompletely dominant with variable expressivity. Heterozygous individuals (N/P result) who inherit the mutation from only one parent may have milder, later-onset, or variable clinical signs. In contrast, homozygotes (P/P result) who inherit the mutation from both parents usually show a more severe or earlier-onset form of the disease. Individuals with an N/N result are not at risk of LSD.

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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