GENOMIA
genetic laboratory

Result certificate #012345

Detection of the c.1107del mutation in the CNP gene causing LSD in Dalmatians

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

The presence or absence of the c.1107del mutation in the CNP gene, which causes lysosomal storage disease (LSD) in Dalmatians, was examined. LSD is characterized by the accumulation of autofluorescent inclusions within lysosomes in the cerebral cortex, cerebellum, optic nerve, and cardiac muscle. In Dalmatians, the first neurological signs typically appear at approximately 18 months of age and progressively worsen over time. Clinical signs include anxiety, restlessness, circling behavior, hypersensitivity, cognitive decline, sleep disturbances, loss of coordination, urinary and fecal incontinence, and visual impairment.

The mode of inheritance is described as autosomal incompletely dominant with variable expressivity. Heterozygous individuals (N/P genotype) that have inherited the mutation from only one parent may exhibit milder, later-onset, or variable clinical signs. In contrast, homozygous individuals (P/P genotype) that have inherited the mutation from both parents typically develop a more severe or earlier-onset form of the disease. Individuals with the N/N genotype are not at risk of developing 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

verified and certified document
GENOMIA genetic laboratory

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