GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.597G>A mutation in CTSD gene causing NCL10 in American Bulldogs

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.597G>A mutation in CTSD gene causing NCL10 in American Bulldogs breed was tested. Neuronal Ceroid Lipofuscinosis type 10 (NCL10) is an inherited neurodegenerative disease characterised by accumulation of autofluorescent lipopigments (ceroid and lipofucsin) within the cells of the nervous system. Due to accumulation of lipopigments, the transfer of important information from the brain to muscles stops progressively. In affected dogs, progressive ataxia and hypermetria of hind limbs and forelimbs develop. The clinical signs of NCL10 occur first between the ninth month and three years of age and get progressively worse.

Mutation that causes NCL10 in American Bulldogs is inherited as an autosomal recessive trait. That means the 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, accredited method

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

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