GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.124+1G>A mutation in PPT1 gene causing NCL1 in Cane Corso

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.124+1G>A mutation in PPT1 gene causing Neuronal ceroid lipofuscinosis type 1 (NCL1) in Cane Corso 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 of NCL1 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

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 s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999