GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.1118C>T mutation in ATP13A2 gene causing NCL12 disease in Australian Cattle Dogs

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.1118C>T mutation in ATP13A2 gene causing Neuronal ceroid lipofuscinosis type 12 (NCL12) in Australian Cattle Dogs was tested. NCL is characterized by accumulation of lipid-like storage material (ceroid and lipofuscin) in nerve cells. The onset and clinical course of the disease are variable and individual. The affected dogs exhibit at least four of the following progressive neurological clinical symptoms: loss of vision, behavioural changes, cerebellar ataxia, tremors, decline of cognitive and motor functions, sleep disturbance and seizures. NCL includes neurological symptoms such as disorientation, states of anxiety and aggressive behaviour and problems with food intake. The degree of neurodegeneration increases with the age and mental abnormalities and spasm occur in all affected individuals. Changes in gait and posture can be observed – stumbling gait, limb stiffness. As there is no cure for this disease, it ultimately leads to death or euthanasia of the affected dog.

Mutation that causes NCL12 in Australian Cattle Dogs 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

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