GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.829T>C mutation in CLN6 gene causing NCL6 in Australian Shepherds

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.829T>C mutation in CLN6 gene causing neuronal ceroid lipofuscinosis type 6 (NCL6) in Australian Shepherds was tested. The NCL disorder is characterized by excessive accumulation of waste lipopigment compounds primarily in the cells of the nervous system. The nerve cells in the cortex and the cerebellum and the retinae cells are affected and destroyed due to the high content of lipofuscin and its increasing pressure. The signs may include loss of vision, behavioural changes, worsening of motor and cognitive abilities, seizures which may look like epileptic seizures. The affected dogs start showing signs of NCL6 around a year and a half of age. It ends by premature death usually within one year after the first signs appear.

Mutation that causes NCL6 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), they are healthy but they can transmit the mutation on their offspring. Dogs with N/N genotype are without risk of NCL6. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 50 % N/P (healthy carriers) and 25 % P/P (affected).

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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