GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.1579G>A mutation in PLA2G2 gene causing NAD in Pappilons and Phalene

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

Presence or absence of c.1579G>A mutation in PLA2G2 gene causing Neuroaxonal dystrophy (NAD) in Papillon and Phalene breed was tested. NAD belongs to the group of rare inherited neurodegenerative diseases. The first symptoms appear around 3 months of age. There can be observed problems with gait, even complete limb paralysis, loss of coordination, head tremor or intention tremor, inability to stand. Further neurological symptoms occur from the 6 month of age, such as blindness, strabismus, loss of reaction to threat, difficulty swallowing and progressive celebellar atrophy.

Mutation that causes NAD 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 NAD. 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: SOPAgriseq_canine, ngs, 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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