GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.344G>A mutation in the CAPN1 gene causing LOA in Jack and Parson Russell Terriers

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.

SAMPLE

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.344G>A mutation in the CAPN1 gene causing LOA (Late Onset Ataxia) in Jack Russell Terriers and Parson Russell Terriers was tested. The Late Onset Ataxia is characterized by lack of balance and incoordination of gait. The clinical symptoms are usually noticed between 6 months and 12 months of age. The main symptoms are stiffness of hind limbs, difficulty going up the stairs and incoordination when dumping. The disease is progressive and after the onset of the first signs the problems with balance and gait incoordination are increasing rapidly. The neurological examination of the affected dogs shows symmetric spino-cerebellar ataxia - cerebellar malfunction characterized by inability to carry out precise and quick movements of skeletal muscles.

Mutation that causes LOA 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.

Test does not exclude present of mutation causing another type of spinocereberal ataxia.

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

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