GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.627C>G mutation in KCNJ10 gene causing SCA in 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.

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.627C>G mutation in KCNJ10 gene causing SCA (Spinocerebellar Ataxia) in Parson Russell Terriers, Jack Russell Terriers and Smooth-Haired Fox Terriers was tested. The clinical symptoms are usually noticed between 2 to 6 months of age. The disease is caused by degeneration of spinal nerves that carry information to the cerebellum. The symptoms are very similar to signs shown by dogs affected with Late Onset Ataxia (LOA) that begin with coordination difficulties when walking, running, turning and jumping. The problems with movement coordination can often progress. The majority of cases also develop myokymia, an involuntary twitching of the muscles. The mykomia also becomes progressively worse with the age and can result in generalized muscle spasms and over-heating. Some dogs may even have true epileptic seizures.

Mutation that causes SCA 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: 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

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