

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.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 myokymia 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 spinocerebellar 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



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