

## Result certificate #012345

Detection of c.1105+5G>A mutation in HCRTR2 gene causing narcolepsy in Labrador Retrievers

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.1105+5G>A mutation in HCRTR2 gene causing narcolepsy in Labrador Retrievers was tested. Narcolepsy is a sleeping disorder characterised by shortened sleep latency and cataplexy (sudden temporary loss of muscle tone without loss of consciousness). Affected dogs are sleepy, unable to stay awake for extended periods of time and fall asleep faster than normal dogs. Cataplectic episodes occur usually in response to a positive emotional stimulus (food, play). The first signs of cataplexy are a buckling of both hind legs and a drooping of neck. The muscle weakness lasts for a few seconds to minutes. During the episodes of collapse the dog remains conscious, has its eyes open and is able to track objects with its eyes. If the collapse lasts more than a few minutes, the dog may fall asleep during the episode.

Mutation that causes narcolepsy is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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.

Method: SOPAgriseg canine, ngs, accredited method

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Approved by: Mgr. Martina Šafrová, Laboratory Manager

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