GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.160G>A mutation in HCRTR2 gene causing narcolepsy in Dachshunds

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.160G>A mutation in HCRTR2 gene causing narcolepsy in Dachshunds 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: 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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