

Result certificate #012345

Detection of c.398C>T mutation in PIGN gene causing Paroxysmal dyskinesia Soft Coated Wheaten 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.

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.398C>T mutation in PIGN gene causing Paroxysmal dyskinesia in the Soft Coated Wheaten Terriers was tested. Paroxysmal dyskinesia is a genetic disorder characterized by episodes of involuntary movements. The involuntary movements are manifested as dystonia, while consciousness is retained. Episodes occur without previous exertion or fasting and can sometimes be triggered by excitement. They can last from a few minutes to several hours and can occur more than 10 times a day.

Mutation that causes Paroxysmal dyskinesia 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.

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



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