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Result certificate #012345

Detection of c.1793_1794ins36 mutation in RAPGEF6 gene causing LP in Miniature Bull Terriers and Bull 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 mutation c.1793_1794ins36 in RAPGEF6 gene causing Laryngeal Paralysis (LP) in Miniature Bull Terriers and Bull Terriers was tested. Laryngeal Paralysis (LP) is the inability to abduct the arytenoid cartilages during inspiration due to defective function of laryngeal nerves. This results in a partial to complete obstruction of airways and consequent respiratory distress. The clinical signs may vary and may include respiratory murmur, wheezing, cyanosis, voice impairment, extreme breathlessness when exercising, life-threatening episodes of breathing difficulties and suffocation. In some cases, the respiratory difficulties can be fatal.

Mutation that causes Laryngeal Paralysis (LP) in Miniature Bull Terriers and Bull Terriers 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: SOP188-MPS-canine, MPS

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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SAMPLE