

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: SOPAgriseq\_canine, ngs

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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