

Detection of mutation  
g.4411956\_441190delGTTT in VPS13B gene  
causing TNS in Border collies

**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 g.4411956\_4411960delGTTT in exon 19 of VPS13B gene causing Trapped Neutrophil Syndrome (TNS) in Border collie breed was tested. Due to this mutation the correct function of white corpuscles - neutrophils - is impaired. They take part in fighting bacterial infections and are important participants in acute inflammation. The failing of immune system can be seen in pups from as early as 2 weeks old and the pups die or are euthanized by approx. 4 months of age. The first symptoms may include apathy, loss of appetite, diarrhoea or poor mobility. Other symptoms depend on the type of infection the pup happens to contract.

Mutation that causes TNS in border collies 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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