

Detection of c.1358\_1359del mutation  
ADAMTS20 gene causing CLPS in Nova  
Scotia Duck Tolling retrievers

**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.1358\_1359del mutation ADAMTS20 gene causing cleft lip with or without cleft palate (CL/P, CLPS) in Nova Scotia Duck Tolling retrievers was tested. Cleft lip is a genetic disorder that occurs during the embryonic development of a puppy. Cleft lip may or may not extend to cleft palate. Clinical signs occur with cleft palate and include nasal discharge of fluid, sneezing, snorting, coughing, impaired ability to accept food and associated slowed growth and poor weight gain.

In Nova Scotia Duck Tolling Retrievers, there is another mutation in the DLX6 gene causing a cleft palate; this test does not detect the DLX6 gene mutation.

Mutation that causes CLPS in Nova Scotia Duck Tolling retrievers 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: 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



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