GENOMIA
genetic laboratory

Result certificate #012345

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

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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