

Result certificate #012345

Detection of c.1393C>T mutation in DNM2 gene causing CNM 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)

Explanation

Presence or absence of c.1393C>T mutation in DNM2 gene causing Centronuclear Myopathy (CNM) in Border Collies was tested. CNM is a defect of muscle fiber development. Initially, it is manifested by intolerance of muscle load, weakness of skeletal muscles and slightly disturbed walking. The disease is progressive and causes muscle atrophy and structural anomalies of muscle fibers, including nuclear centralization and mitochondrial abnormalities.

The CNM mutation is inherited autosomal dominant. This means that one copy of the mutated gene inherited from one of the parents is sufficient to show the symptoms of the disease.

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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