GENOMIA
genetic laboratory

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, accredited method

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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