
Customer: Dr. Josette Debrincat, 20, Animal Doctors, Guze Bajada, HMR 2131 Hamrun, Malta**Sample:**

Sample: 24-26623

Date received: 01.10.2024

Sample type: blood

Information provided by the customer

Name: Scott Dell'Antica Contrada**Breed:** Border Collie

Microchip: 380 206 044 889 554

Reg. number: MKC BOR/14/2024

Date of birth: 18/12/2022

Sex: male

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: 13.10.2024

Date of testing: 01.10.2024 - 13.10.2024

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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