

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: Xn/Y

Explanation

Presence or absence of c.2841delT mutation in DMD gene causing Duchenne muscular dystrophy (DMD) in Border Collies was tested. The disease is characterized by progressive muscle weakness that is ultimately fatal. Clinical signs begin to appear in puppies between 8 and 10 weeks of age and include a stiff gait or shortened stride, inability to fully open the jaw, difficulty swallowing, excessive salivation and marked wasting of the muscles of the body and limbs.

The mutation is X-linked. This means that it is localized on the X chromosome. Males have an X and a Y chromosome, so they can only be healthy (Xn/Y) or affected (Xm/Y). Females have two X chromosomes, so they can either be healthy (Xn/Xn), carriers (Xn/Xm) or affected (Xm/Xm). Female carriers do not show clinical signs but are able to pass the mutant allele to their offspring.

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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