

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: Xn/Xn**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:** 06.02.2021**Date of testing:** 01.02.2021 - 06.02.2021**Approved by:** Mgr. Martina Šafrová, Laboratory Manager

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Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

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