

Result certificate #012345

Detection of c.2668C>T mutation in DMD gene causing DMD in Australian Labradoodles

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/Y

Explanation

Presence or absence of c.2668C>T mutation in DMD gene causing Duchenne muscular dystrophy (DMD) in Australian Labradoodles 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: SOP188-MPS-canine, MPS, 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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