GENOMIA
genetic laboratory

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: 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

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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