GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.6210+1G>A and c.4726C>T mutations in COL6A3 gene causing Muscular dystrophy in Labrador Retrievers

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: PREVIEW RESULT LINE

Explanation

Presence or absence of c.6210+1G>A and c.4726C>T mutation in COL6A3 gene causing Muscular dystrophy, Ullrich type, in Labrador Retrievers was examined. Clinical manifestations usually appear in puppies and include mainly muscle weakness and excessive joint mobility.

The disease develops in homozygotes who inherit the same mutation from both parents (P/P), in compound heterozygotes who inherit a different mutation from each parent (P/P compound heterozygote), and in heterozygotes who carry only one mutation (N/P). Individuals with a result of N/N do not carry the causal mutation for muscular dystrophy.

Method: SOPAgriseq_canine, ngs

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

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