

Result certificate #012345

Detection of c.531-2A>G mutation in DMD gene causing GRMD in Golder 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: Xn/Xn

Explanation

Presence or absence of c.531-2A>G mutation in DMD gene causing Muscular Dystrophy in Golder Retrievers was tested. GRMD is a degenerative muscular disease causing loss of active muscular tissue and movement disorders. Disease is usually expressed in puppies around 8 weeks of age. Dogs with severe form die soon after diagnosis, while dogs with milder form of the disease can survive several years. GRMD is X-linked recessive disease.

Females have XX chromosomes. So females have three possibilities as regards GRMD:

XnXn - females with two normal X chromosomes = normal phenotype, a healthy female XnXm - females with one normal X (Xn) and one mutant X (Xm) = a female carrier.

XmXm - females with two mutated X chromosomes = an affected female

Males have XY chromosomes. So they have two possibilities as regards GRMD:

XnY - normal phenotype, a healthy male

XmY - an affected male; he inherited mutated X chromosome from his mother

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