GENOMIA
genetic laboratory

Customer: Míšek Pavel, Radošov 192, 36272 Kyselka, Czech Republic
Sample:
Sample: 22-31704
Date received: 15.11.2022
Sample type: buccal swab

Information provided by the customer
Name: Nelson Golden Victory
Breed: Golden Retriever
Tattoo number: BNY 4513
Microchip: 643 099 011 505 604
Reg. number: ČLP/GR/23461
Date of birth: 11.06.2021
Sex: male
Date of sampling: 14.11.2022
The identity of the animal has been checked by MVDr. Sabina Marie Šteklová, KVL 4726

Result: Xn/Y

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: SOP172-GRMD, direct DNA sequencing

Date of issue: 29.11.2022
Date of testing: 15.11.2022 - 29.11.2022
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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