

Customer: Dušan Kollárik, Belá 33, 03811 Belá-Dulice, Slovak Republic

Sample:

Sample: 25-05401

Date received: 04.03.2025

Sample type: buccal swab

Information provided by the customer

Name: Heidi Bella Aurea

Breed: Golden Retriever

Microchip: 941 000 026 557 633

Reg. number: SPKP 4826/24

Date of birth: 17.6.2023

Sex: female

Date of sampling: 28.02.2025

The identity of the animal has been checked by MVDr. Juraj Chorváth, 0011

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, accredited method

Date of issue: 14.03.2025

Date of testing: 04.03.2025 - 14.03.2025

Approved by: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

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

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