

# Result certificate #290764

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

**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:

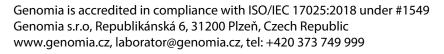
XnXn - females with two normal X chromosomes = normal phenotype, a healthy female XnXm - females with one normal X (Xn) and one mutant X (Xm) = a female carrier. XmXm - females with two mutated X chromosomes = an affected female

### 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





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