

Sample

Sample: 16-33934
Name: FINNA Bella Aurea
Breed: Golden Retriever
Microchip: 941 000 016 041 679
Reg. number: SPKP 3007/18
Date of birth: 23.3.2015
Sex: female
Date received: 16.01.2017
Sample type: buccal swab

Customer

Dušan Kollárik
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Ordered on 2019-05-02.

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

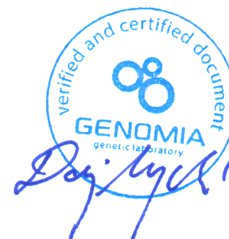
Males have XY chromosomes. So they have two possibilities as regards GRMD:

XnY - normal phenotype, a healthy male
XmY - an affected male; he inherited mutated X chromosome from his mother

Method: SOP172-GRMD, direct DNA sequencing

Report date: 07.05.2019

Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager



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