

Sample

Sample: 17-29146
Name: Kate Diamond in the heart
Breed: Golden Retriever
Microchip: 900 182 001 000 442
Reg. number: ČLP/GR/19246
Date of birth: 8.2.2016
Sex: female
Date received: 03.11.2017
Sample type: blood
The identity of the animal has been checked by
MVDr. Jana Kalendová

Customer

Martin Luňák
Urbánkova 1282
67602 Moravské Budějovice
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Result: Xn/Xn

Explanation

Presence or absence of c.531-2A>G mutation in DND 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: 07.11.2017

Responsible person: Mgr. Martina Šafrová, Laboratory Manager



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