GENOMIA
genetic laboratory

Result certificate #290758

Detection of c.880G>A mutation in LOC608697 gene causing CMS disease in Golden 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: Mutation was not detected (N/N)

Legend: N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

Explanation

Presence or absence of c.880G>A mutation in LOC608697 gene causing congenital myasthenic syndrome (CMS) disease in Golden Retrievers was tested. The disease is manifested by skeletal muscle weakness, especially after exercise, and a decrease in spinal reflexes. The first symptoms appear already in the first weeks or months of life. The disease progresses rapidly and most cases lead to the death or euthanasia of the affected puppy.

Mutation that causes CMS is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; disease affects dogs with P/P genotype only. The dogs with N/P genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

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

verified and certified document GENOMIA genetic laboratory

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