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Result certificate #290756

Detection of mutations in MTMR2 a SH3TC2 genes causing Hypomyelinating polyneuropathy 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: MTMR2 Mutation was not detected (N/N)
Result: SH3TC2 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.1924C>T mutation in SH3TC2 gene and mutation c.1479+1G>A in MTMR2 gene causing Hypomyelinating polyneuropathy in Golden Retrievers was tested. Hypomyelinating polyneuropathy is a genetic disease affecting the peripheral nervous system. It results in insufficient production of myelin sheath, causing muscle weakness, loss of reflexes and difficulty coordinating movements.

Mutations that causes Hypomyelinating polyneuropathy are inherited probably as an autosomal recessive trait. That means the 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

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
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Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999