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Result certificate #300638

Detection of c.934_935delAG mutation in CLN5 gene causing NCL in Golden Retrievers

Customer: Míšek Pavel, Radošov 192, 36272 Kyselka, Czech Republic
Sample:
Sample: 22-31704
Date received: 15.11.2022
Sample type: buccal swab

Information provided by the customer
Name: Nelson Golden Victory
Breed: Golden Retriever
Tattoo number: BNY 4513
Microchip: 643 099 011 505 604
Reg. number: ČLP/GR/23461
Date of birth: 11.06.2021
Sex: male
Date of sampling: 14.11.2022
The identity of the animal has been checked by MVDr. Sabina Marie Šteklová, KVL 4726

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 mutation c.934_935delAG mutation in CLN5 gene causing Neuronal Ceroid Lipofuscinosis (NCL) in Golden Retrievers was tested. NCL is manifested by accumulation of lipopigments (coroid and lipofuscin) in the lysosomes. The clinical symptoms are progressive neurological signs including disorientation, worsening of motor functions, anxiety, aggression, seizures and problems with food intake. Usually visual impairment and loss of vision occur as well. The onset of the disease and its clinical course vary substantially between breeds. The first signs occur most often after 15 month of age. The degree of neurodegeneration increases with the age and all affected dogs develop psychological abnormalities and spasms. Changes in gait and posture – stumbling, leg stiffness, tremor - can be observed as well.

Mutation that causes NCL in Golden Retrievers 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: SOP171-NCL5-GR, fragment analysis

Date of issue: 05.06.2025
Date of testing: 27.05.2025 - 05.06.2025
Approved by: Ing. Irena Rusková, Analyst

verified and certified document
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