GENOMIA
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Result certificate #290766

Detection of c.934_935delAG mutation in CLN5 gene causing NCL 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 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: 20.03.2025
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Approved by: Mgr. Martina Šafrová, Laboratory Manager

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