

**Customer:** Dušan Kollárik, Belá 33, 03811 Belá-Dulice, Slovak Republic**Sample:**

Sample: 23-01202

Date received: 23.01.2023

Sample type: buccal swab

Information provided by the customer

**Name:** GIANNA Bella Aurea**Breed:** Golden Retriever

Microchip: 941 000 023 870 583

Reg. number: SPKP 3603/21

Date of birth: 15.5.2019

Sex: female

Date of sampling: 19.01.2023

The identity of the animal has been checked by MVDr. Lenka  
Blahušiaková, KVL 0814**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: SOPAgriseq\_canine\_CP, ngs

Date of issue: 03.02.2023

Date of testing: 23.01.2023 - 03.02.2023

Approved by: Mgr. Martina Šafrová, Laboratory Manager

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