

## Result certificate #218647

Detection of c.1145G>C mutation in COL1A1 gene causing osteogenesis imperfecta in Golden Retrievers

**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)

## Explanation

Presence or absence of c.1145G>C mutation in COL1A1 gene causing osteogenesis imperfecta type 3 (OI) in Golden Retrievers was tested. OI is an inherited connective tissue disease characterized by thinning of the bones, leading to multiple fractures of the long bones and ribs. Clinical manifestations appear in puppies as young as a few weeks old.

Inheritance of the causal mutation is autosomal dominant. This means that it only takes one copy of the mutated gene inherited from one parent to cause symptoms of the disease.

Method: SOPAgriseq\_canine\_TD, ngs

Date of issue: 06.02.2023 Date of testing: 23.01.2023 - 06.02.2023 Approved by: Mgr. Martina Šafrová, Laboratory Manager



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