GENOMIA
genetic laboratory

Result certificate #290761

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: 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)

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, ngs, accredited method

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 GENOMIA genetic laboratory

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