GENOMIA
genetic laboratory

Result certificate #012345

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

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic
Sample:
Sample: 21-12345
Date received: 01.02.2021
Sample type: blood

Information provided by the customer
Name: Lassie DEMO
Breed: Plemeno
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 1.1.2020
Sex: female
Date of sampling: 01.02.2021
The identity of the animal has been checked.

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: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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