

Detection of c.3656\_3859delinsTGTCATTGG mutation in COL1A2 gene causing osteogenesis imperfecta in Beagles

**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.3656\_3859delinsTGTCATTGG mutation in COL1A2 gene causing osteogenesis imperfecta (OI) in Beagles was tested. OI is an inherited connective tissue disease characterized by extreme bone fragility that leads to numerous fractures.

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

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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