

Detection of c.1991\_1995delTTTCC  
mutation in ENAM gene causing  
Amelogenesis Imperfecta in Italian  
Greyhounds

**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)

**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 c.1991\_1995delTTTCC mutation in ENAM gene causing Amelogenesis imperfecta (AI) in Italian Greyhounds was tested. AI (congenital enamel hypoplasia) is a hereditary teeth disorder that is characterized by abnormalities in the enamel structure, composition and amount. Proteins that take part in the process of enamel mineralisation are enamelin (ENAM-gene), amelogenin (AMELX-gene) and acid phosphatase (ACPT). In Italian Greyhounds the amelogenesis imperfecta is caused by mutation in the enamelin gene. The tooth enamel of deciduous and permanent teeth is of poor quality and may be missing in some areas and is prone to attrition. The teeth have rough surface and brownish mottling occurs in areas where the enamel is thinning or missing. The teeth appear to be small and pointed with increased gaps between the teeth.

Mutation that causes AI 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, 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



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