GENOMIA
genetic laboratory

Result certificate #012345

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.

SAMPLE

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: SOP188-MPS-canine, MPS, 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

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