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Result certificate #012345

Detection of mutation insertion in the RPGRIP1 gene causing CORD1 in dogs

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 mutation g.8228_8229insA29GGAAGCAACAGGATG in RPGRIP1 gene causing CORD1 disease (cone-rod dystrophy type 1) in dogs was tested. This mutation probably causes a frame shift and formation of premature stop codon. CORD1 is an early onset form of progressive retinal atrophy disease (PRA). Individuals with N / N result (negative / negative) do not carry the mutation. Individuals with N / P (negative / positive) are carriers of the mutation. In individuals with two mutated alleles (P/P, positive / positive) the CORD1 disease does not necessarily have to develop. The exact mechanism of CORD1 disease is unknown. Influence of other mutations or modifications at the level of transcription cannot be excluded.

Method: SOPAgriseq_canine, ngs
Sensitivity (probability of correct identification of the defective form of the gene in heterozygous or mutated homozygous) is higher than 99%. Specificity (probability of correct identification of the normal form of the gene in a normal homozygous or heterozygous) is higher than 99%.

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
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