GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.1847del mutation in PDE6A gene causing RCD3 disease 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.

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 mutation c.1847del (previously called c.1940delA) in PDE6A gene causing PRA-rcd3 (Progressive Retinal Atrophy form Rod Cone Dysplasia 3) in Cardigan Welsh Corgi and Chinese Crested dog was tested. PRA-rcd3 is a cureless disease; affected individuals become usually blind in very early age.

Mutation that causes PRA-rcd3 in Cardigan Welsh Corgi and Chinese Crested dog is inherited as an autosomal recessive trait. That means the 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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