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Result certificate #012345

Detection of c.482G>A mutation in VMD2 gene causing CMR2 disease 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)

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.482G>A in VMD2 gene was tested. This mutation causes CMR2 disease (Canine Multifocal Retinopathy type 2) in Coton de Tulears. VMD2 gene is coding a protein bestrophin which is responsible for right forming of pigment epithelium in retina. Described gene mutation causes aminoacid change p.G161D in sequence of bestrophin. Clinically, rose-grey colored lesions are remarkable in retina. CMR disease usually arises before 4th month of age in an affected puppy. Total blindness usually comes in higher age.

Mutation that causes CMR2 disease 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: 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

verified and certified document
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