

## Result certificate #012345

Detection of c.73C>T mutation in VMD2 gene causing CMR1 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.73C>T in VMD2 gene causing CMR1 disease (Canine Multifocal Retinopathy type 1) in Great Pyrenees, English Mastiffs, Bullmastiffs, Australian Shepherd and related breeds was tested. The mutation forms premature stopkodon (R25X) in canine VMD2 gene; the gene is responsible for right forming of pigment epithelium in retina. 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 c.73C>T in VMD2 gene 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



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