

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: SOP182-CMR2, HRMA

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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