

## Result certificate #012345

Detection of c.952\_53insC mutation in IQCB1 gene causing CRD2 in American Pit Bull Terriers

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 c.952\_53insC mutation in IQCB1 gene causing Cone-rod dystrophy 2 (CRD2) in American Pit Bull Terriers was tested. Affected dogs suffer from a severe and rapidly progressive loss of function of the cones, which is accompanied by a relatively slower loss of function of the rods. The disease leads to a significant deterioration of vision, both in light and in darkness, up to complete blindness.

Mutation that causes CRD2 is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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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