

Detection of c.2420G>A mutation in PDE6B causing PRA-RCD1 disease in Irish Setters

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.2420G>A in PDE6B gene (gene for beta-subunit cGMP phosphodiesterase enzyme (PDE)) causing PRA-RCD1 disease in Irish Setters was tested. The mutation forms premature stopcodon; the mutated enzyme is 49 amino acids shorter and cannot fulfil its normal function. PRA-RCD1 (Progressive Retinal Atrophy form Rod-Cone dysplasia 1) is hereditary eye defect. Photoreceptor degeneration starts in early age. Degeneration is completed in 2 years when affected dog is totally blind.

Mutation that caused PRA-RCD1 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: 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



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