

Detection of c.3149_3150insC mutation in
the C17H2orf71 gene causing RCD4 in
several dog breeds

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 c.3149_3150insC mutation in the C17H2orf71 gene causing rod-cone dysplasia (RCD4) was tested. The mutation is related with RCD4 in breeds: Australian Cattle Dog, English Setter, Gordon Setter, Irish Red & White Setter, Irish Setter, Polish Lowland Sheepdog, Small Munsterlander, Standard Poodle and Tibetan Terrier. RCD4 is a form of progressive retinal atrophy (PRA) characterized by the degeneration of the photoreceptors in the retina. It results in vision loss and eventually complete blindness. The average age of RCD4 diagnosis is 10 years.

Mutation that causes RCD4 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), they are healthy but they can transmit the mutation on their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 50 % N/P and 25 % P/P.

In individuals with N/N and N/P result might develop any different form of PRA, due to mutations that were not detected by this test.

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



Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz

Without a written consent by the lab, the report must not be reproduced unless as a whole.

The result refers only to the tested sample, as received. Genomia is not responsible for the accuracy of the information provided by the customer.