

Detection of g.1,432,293G>A mutation in
HIVEP3 gene causing Progressive Retinal
Atrophy in Miniature Schnauzer

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 g.1,432,293G>A mutation in HIVEP3 gene causing Progressive Retinal Atrophy type PRA1 in Miniature Schnauzer was tested. Clinical symptoms usually appear at the age of 4 years. The symptoms start with night blindness which is followed with a slow deterioration of vision and complete blindness under any light conditions.

Mutation that causes PRA1 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

In Miniature Schnauzers, the finding of other mutations causing progressive retinal atrophy is expected, so also dogs with N/N and N/P results should be regularly ophthalmologically examined.

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 sample as received. Genomia is not responsible for the accuracy of the information provided by the customer.