

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

Customer: Alice Trojanová, Svatá 222, 26751 Svatá, Czech Republic

Sample:

Sample: 19-21318

Date received: 06.08.2019

Sample type: blood

Information provided by the customer

Name: Domus Letitia Goody

Breed: Schnauzer Miniature

Microchip: 688 032 000 030 589

Reg. number: CDK 3127/18ZSCH

Date of birth: 15.4.2018

Sex: male

Date of sampling: 02.08.2019

The identity of the animal has been checked by MVDr. Petr
Gblec

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: SOP173-PRA1, PCR-RFLP

Date of issue: 17.05.2021

Date of testing: 13.05.2021 - 17.05.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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