GENOMIA
genetic laboratory

Result certificate #150051

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

Customer: Ольга Котишевская, улица Радищева, дом 3, квартира 86, 220007 Минск, Belarus

Sample:
Sample: 18-35322
Date received: 02.01.2019
Sample type: blood spot

Ordered on 2020-05-18.

Information provided by the customer
Name: Shining Wait S Taganiego Roga
Breed: Schnauzer Miniature
Tattoo number: GRM 97
Microchip: 643 094 100 443 024
Reg. number: BCU 183 - 003722
Date of birth: 26.07.2017
Sex: male

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 apear 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: SOP172-PRA1, direct DNA sequencing

Date of issue: 25.05.2020
Date of testing: 18.05.2020 - 25.05.2020
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

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