GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.1752_1755delAACT mutation
in CNGA1 gene causing PRA
in Shetland Sheepdogs

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.

SAMPLE

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.1752_1755delAACT mutation in CNGA1 gene causing eprogressive retinal atrophy (CNGA1-PRA) in Shetland Sheepdogs was tested. The PRA starts by degeneration and loss of rod cells leading to so-called night blindness. It is followed by cone degeneration which is connected with visual impairment at bright light. The range of the age of the disease onset is relatively wide and in average the first signs of PRA become apparent at the age of 5 years.

Mutation that causes CNGA1-PRA in Shetland Sheepdogs is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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

verified and certified document
GENOMIA genetic laboratory

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