GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.126delG in HES7 gene causing SCD 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 mutation c.126delG in HES7 gene causing Spondylocostal Dysostosis (SCD, Comma Defect) in Miniature Schnauzer was tested. SCD is a severe hereditary disease that causes changes in axial skeleton (spine, ribs). The disorder is characterized by changes in vertebra shape, so-called hemivertebrae (wedge-shaped vertebrae), and rib anomalies (fusion, and reduction in number, tumour growth). The malformation of the thorax and of the spine affects the respiratory function of lungs and the function of the spine and therefore the affected puppies, due to the severity of skeletal abnormalities, are stillborn or die briefly after birth. Beside these defects, the newly born puppies show an abnormal reduction in body length, distinctive forehead and limb defects.

Mutation that causes SCD in Miniature Schnauzer 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: SOPAgriseq_canine, ngs, 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

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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