GENOMIA
genetic laboratory

Result certificate #012345

Detection of deletion in RBM20 gene causing DCM in Schnauzers

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.2472_2493delGAAGGTCAAAATCTGCCCAGAA in RBM20 gene causing dilated cardiomyopathy (DCM) in Schnauzer and Giant Schnauzer was tested. The DCM is a condition in which the heart´s ventricular walls are stretched and the walls become thinner. Thinning of the walls causes enlargement of the ventricles and in most cases the left heart ventricle is affected first. These changes result in worsened function of the heart, the heart´s ability to contract and to pump the required amount of blood. The perfusion of the bodily organs is poor; systemic oedemas and pulmonary oedema occur resulting in dyspnoea and coughing. The symptoms of the disease also include ventricular arrhythmia and myocardial scarring. The first clinical signs occur between the first and the third year of age. At the early stage of DCM, the affected dogs show reduced tolerance to physical stress and overall weakness, fainting and collapse. The prognosis is not good and DCM is the most common cause of invalidity and untimely death of dogs.

Mutation that causes DCM in 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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