

Sample

Sample: 20-09208
Name: Hanny Fortuna Moravia
Breed: Schnauzer
Microchip: 203 098 100 387 798
Reg. number: CMKU/KS/8802/15/18
Date of birth: 8.7.2015
Sex: female
Date received: 16.04.2020
Sample type: buccal swab

Customer

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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: SOP171-DCM, fragment analysis

Report date: 24.04.2020

Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager



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