

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_2493delGAAGGTCAAATCTGCCAGAA` 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

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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