GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.803C>T mutation in CLCN1 gene causing Congenital Myotonia in Miniature 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 c.803C>T (p.Thr268Met) mutation in CLCN1 gene causing Congenital Myotonia (MC) disease in Miniature Schnauzer was tested. MC is characterized by disorder of the relaxation of muscle contraction resulting in muscle stiffness. The disease belongs to canalopathies; this means that it is caused by mutation in a gene that encodes the ion channel in muscle fibre. MC affected dogs have a stiff, stilted gait, so-called "bunny-hopping" type movement. In some cases an uncontrolled turning and downfalls may occur. In comparison with healthy animals, the affected Schnauzers have a superior prognathism and a shortened lower jaw, caused probably by contraction of jaw muscles.

Mutation that causes MC in Miniature Schnauzer is inherited as an autosomal recessive trait. That means the 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
Sensitivity (probability of correct identification of the defective form of the gene in heterozygous or mutated homozygous) is higher than 99%. Specificity (probability of correct identification of the normal form of the gene in a normal homozygous or heterozygous) is higher than 99%.

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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