

Sample

Sample: 18-24523
Name: CARLSBERG
Breed: Schnauzer Miniature
Microchip: 941 000 019 533 876
Reg. number: PKR.II-128831
Date of birth: 24.VI,2016
Sex: male
Date received: 12.09.2018
Sample type: buccal swab

Customer

Monika Opoka
Gersona 20a, 3
30-818 Kraków
Poland

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 Schnautzer 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 Schnautzer 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: SOP173-MC, PCR-RFLP, 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%.

Report date: 20.09.2018

Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

