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Result certificate #012345

Detection of mutations in CLCN1 gene causing MC in cats

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic
Sample:
Sample: 08-12346
Date received: 01.01.2008
Sample type: buccal swab

Information provided by the customer
Name: Madame Théophile DEMO
Breed: Persian cat
Date of birth: 31.12.1909
Reg. number: (CZ)ABCD EF 123/45/XYZ
Microchip: 123 456 789 012 345
Sex: female
Date of sampling: 01.01.2008
The identity of the animal has been checked.

Result: PREVIEW RESULT LINE

Explanation

Presence or absence of c.1930+1G>T, c.428_433+1del, c.991G>C mutations in CLCN1 gene causing Congenital Myotonia (MC) in cats was tested. Affected cats usually show muscle hypertrophy, are stiff and walk with a jerky gait with an abnormally short stride. They have a limited ability to open their jaws, swallow and make sounds.

Mutation that causes MC is inherited probably autosomally recessively which means that the disease develops only in those cats who inherit mutated allele from both parents; disease affects cats with P/P genotype only. The cats 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_feline, MPS

Date of issue: 06.01.2008
Date of testing: 12.06.2008 - 06.01.2008
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
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SAMPLE