

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 catDate of birth: 31.12.1909

Reg. number: (CZ)ABCD EF 123/45/XYZ

Microchip: 123456789012345

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, ngs

Date of issue: 06.01.2008

Date of testing: 12.06.2008 - 06.01.2008

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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