

Detection of c.1190G>A mutation in COLQ
gene causing Dystroglycanopathy
in Devon Rex and Sphynx

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: 123456789012345

Sex: female

Date of sampling: 01.01.2008

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.1190G>A mutation in COLQ gene causing Dystroglycanopathy in Devon Rex and Sphynx was examined. Dystroglycanopathy manifests itself in muscle weakness affecting mainly the limb plexuses, in nervous system disorders and eye malformations.

Mutation that causes Dystroglycanopathy is inherited 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



Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz

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