

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: 123 456 789 012 345

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, MPS

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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Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at [www.genomia.cz](http://www.genomia.cz)

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