

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.7384G>C mutation in ALMS1 gene causing Hypertrophic Cardiomyopathy (HCM4) in cat breeds like American Shorthair, Exotic Shorthair, Munchkin, Scottish Fold and Sphynx was tested. In HCM4, there is thickening (hypertrophy) of the heart muscle, especially of the left ventricular wall. Clinical symptoms include breathing difficulties, coughing, lethargy and weakness.

Mutation that causes HCM4 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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Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz

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