

Detection of c.2458C>T mutation of  
MYBPC3 gene causing HCM disease in  
Ragdoll 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: 123456789012345

Sex: female

Date of sampling: 01.01.2008

The identity of the animal has been checked.

**Result:** Mutation was not detected (N/N)

**Explanation**

Presence or absence of c.2458C>T MYBPC3 gene (R820W) in Ragdoll cats was tested. This mutation is considered to be associated with hypertrophic cardiomyopathy (HCM) in Ragdoll cats. In HCM, primary strengthening (hypertrophy) of the left cardiac ventricle wall and the cardiac septum occurs. HCM symptoms, that may accompany the disease, can be breath shortness, low physical activity from reduced mobility to legs paralysis, appetite decrease, cough, syncope, heart arrhythmia and cardiac murmur of different intensity. The disease manifestation starts at any age.

Mutation R820W in MYBPC3 gene is inherited as an autosomal dominant trait. That means the disease affects cats that carry one or two mutant alleles - cats with result N/P (negative / positive) or P/P (positive / positive). Cats with P/P genotype have more serious symptoms of HCM. Cats with N/N genotype are healthy. Mating N/P and N/N cats will be 50% healthy offspring (N/N) and 50% affected offspring (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](http://www.genomia.cz)

Without a written consent by the lab, the report must not be reproduced unless as a whole.

The result refers only to the tested sample, as received. Genomia is not responsible for the accuracy of the information provided by the customer.