

## Result certificate #012345

Detection of c.91G>C (A31P) mutation in MYBPC3 gene of Maine Coon 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.91G>C (A31P) mutation in MYBPC3 gene was tested. This mutation is considered to be associated with hypertrophic cardiomyopathy (HCM) in Maine Coon 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, syncopes, heart arrhythmia and cardiac murmur of different intensity. The disease manifestation starts at any age.

Cat without risk of development of HCM due to mutation A31P has genetic test result N/N (negative in both alleles). Cat in risk of development of HCM due to mutation A31P has genetic test result N/P or P/P (positive mutation finding in one or both alleles). Mutation is inherited as an autosomal dominant trait. Incomplete penetrance of the disease was described in heterozygotes (Longer et al. 2013).

Method: SOP175-HCM-MM, real-time PCR-ASA

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