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Result certificate #012345

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, syncopes, 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: SOP175-HCM-RAG, 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

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