GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.91G>C (A31P) mutation in MYBPC3 gene of Maine Coon cats

Sample
Sample: 08-12346
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 received: 01.01.2008
Sample type: buccal swab
The identity of the animal has been checked.

Customer
Jan Novák
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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, accredited method

Report date: 01.01.2008
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

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