GENOMIA
genetic laboratory

Result certificate #012345

Detection of mutation in GBE1 gene causing GSD IV disease in Norwegian forest 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 mutation in GBE1 gene causing GSD IV disease in Norwegian forest cats was tested. GCD IV (glycogen storage disease IV) is a disorder of glycogen metabolism, especially deficiency of amylo-(1,4 ---> 1,6)-transglykosilase (GBE = glycogen branching enzyme). Reason for the disorder is complex rearrangement of genomic DNA in GBE1 gene, constituted by a 334 bp insertion at the site of a 6.2 kb deletion that extends from intron 11 to intron 12. Abnormal glycogen is accumulated in myocytes, hepatocytes or neurons; that causes lethal tissue demage. Affected kittens die from hypoglycemia very early after birth. Surviving individuals die before 5 month of age for progressive muscular degeneration.

Mutation in GBE1 gene is inherited as an autosomal recessive trait. That means the disease affects cats with P/P genotype only (positive / positive). The cats with N/P genotype (negative / positive) are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP176-GSDIV, ASA-PCR

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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