

Detection of c.1467_1491inv mutation in
HEXB gene causing GM2 domestic
shorthaired 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: 123 456 789 012 345

Sex: female

Date of sampling: 01.01.2008

The identity of the animal has been checked.

Result: Mutation was not detected (N/N)

Legend: N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

Explanation

Presence or absence of c.1467_1491inv mutation in HEXB gene causing Gangliosidosis (GM2) domestic shorthaired cats was examined. Gangliosidosis, also known as Sandhoff's disease, is a lysosomal storage disease which is characterized by progressive neuromuscular dysfunction and impaired growth from an early age.

Mutation that causes GM2 is inherited autosomally recessively which means that the disease develops only in those cats who inherit mutated allele from both parents; disease affects cats with P/P genotype only. The cats with N/P genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

Method: SOPAgriseq_feline, MPS

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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