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Result certificate #012345

Detection of c.1042_1044del mutation in IDUA gene causing Mucopolysaccharidosis I in 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.1042_1044del mutation in IDUA gene causing Mucopolysaccharidosis I in cats was examined. Affected cats are deficient in the lysosomal enzyme alpha-L-iduronidase, which is part of the glycosaminoglycan degradation pathway. The disease is manifested by flat and broad cheeks, a large head with small ears, thickened neck skin, broad cervical vertebrae and hip subluxation. Other symptoms include an abnormal gait, corneal opacity and a heart murmur due to mitral insufficiency may occur.

Mutation that causes Mucopolysaccharidosis I 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, ngs

Date of issue: 06.01.2008
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Approved by: Mgr. Martina Šafrová, Laboratory Manager

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