GENOMIA
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Result certificate #012345

Detection of c.1333G>A mutation in TPO gene causing Congenital hypothyroidism in 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.

SAMPLE

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.1333G>A mutation in TPO gene causing Congenital hypothyroidism in domestic shorthaired cats was examined. Hypothyroidism is a lack of thyroid gland activity. It is characterized by a large skull with shortened jaw and ears, a square torso and short limbs, often combined with spinal deformity and delayed tooth growth. The typical feature is a goitre, an enlarged thyroid gland. Hypothyroidism is also characterized by mental deficiency and abnormalities of the nervous system. Other less specific features include hypothermia, anorexia, obesity, constipation or lethargy.

Mutation that causes Congenital hypothyroidism 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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