GENOMIA
genetic laboratory

Result certificate #012345

Detection of mutations in GUSB gene causing Mucopolysaccharidosis VII 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.

SAMPLE

Result: PREVIEW RESULT LINE

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.1051G>A and c.[1423T>G;1426C>T] mutations in GUSB gene causing Mucopolysaccharidosis VII in cats was examined. Affected cats suffer from a wide range of manifestations of this disease: slowed growth, difficulty walking (internal rotation of the front paws, weight transfer to the front paws, reduced proprioception of the hind legs, luxation of the kneecap), epileptic seizures, flat and broad head with small distorted ears, corneal opacity, enlarged abdomen or thickening and excessive skin peeling.

Mutation that causes Mucopolysaccharidosis VII 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. The mutations are inherited independently, compound heterozygotes, i.e. carriers of both mutations, can also be affected.

Method: SOPAgriseq_feline, ngs

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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