

Detection of c.1427T>C mutation in ARSB
gene causing Mucopolysaccharidosis VI 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.1427T>C mutation in ARSB gene causing Mucopolysaccharidosis VI in cats was examined. Clinical signs first appear at 6 to 8 weeks of age and include growth retardation, facial dysmorphism (broad cheeks with shortened nose and small ears), skeletal deformities, decreased flexibility of the cervical and lumbar spine, and degenerative joint disease.

Mutation that causes Mucopolysaccharidosis VI 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

Date of testing: 12.06.2008 - 06.01.2008

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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