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# Result certificate #012345

## Detection of c.1749_1752del mutation in MAN2B1 gene causing Alpha-mannosidosis in the Persian 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.1749_1752del mutation in MAN2B1 gene causing Alpha-mannosidosis in the Persian cats was examined. Alpha-mannosidosis is a lysosomal storage disease. Clinical symptoms include neurological signs (ataxia, head tremor, nystagmus, aggressiveness, paralysis), skeletal abnormalities, growth retardation, gingivitis, and corneal and lenticular opacities. The disease is incurable, death occurs after about 6 months.

Mutation that causes Alpha-mannosidosis 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

verified and certified document
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