

## Result certificate #012345

Detection of c.1449\_1452dup mutation in LTBP2 gene causing Glaucoma 3 in Siamese

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.1449\_1452dup mutation in LTBP2 gene causing Glaucoma 3 in Siamese cats was examined. Glaucoma is characterized by increased intraocular pressure, enlargement of the eyeball and elongation of the ciliary processes. Gradual damage to the optic nerve and loss of vision occurs as early as around 6 months of age.

Mutation that causes Glaucoma 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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