GENOMIA
genetic laboratory

# Result certificate #012345

## Detection of 15 bp deletion HEXB gene causing GM2 disease in Burmese 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: 123456789012345
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 15 bp deletion in HEXB gene causing GM2 gangliosidoses in Burmese cats was tested. GM2 gangliosidoses are progressive, fatal neuropathic lysosomal storage disease resulting from a deficiency of β-N-acetylhexosaminidase activity and causes accumulation of GM2 gangliosides in brain tissue. GM2 in Burmese cats has an early onset with severe signs by 3 months. Late signs include complete loss of hind limb use, blindness or epileptic like seizures. The kittens of Burmese cats affected by GM2 hardly survive for 6 months.

Delation that causes GM2 in Burmese cats is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. The dogs 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: SOP171-GM2, fragment analysis
Sensitivity (probability of correct identification of the defective form of the gene in heterozygous or mutated homozygous) is higher than 99%. Specificity (probability of correct identification of the normal form of the gene in a normal homozygous or heterozygous) is higher than 99%.

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
GENOMIA genetic laboratory

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