

**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 c.2899C>T mutation in WNK4 gene causing hypokalemia or hypokalemic polymyopathy (BHP) in cats was tested. The breeds that are at risk for BHP include: Burmese and outcrosses such as Burmilla, Bombay, Cornish Rex, Devon rex, Singapura, Sphynx, Australian Mist, Tiffanie and Tonkinese. The most characteristic clinical sign of BHP disease is a skeletal muscle weakness that is frequently episodic in nature, either generalized, or sometimes localized to the neck and thoracic muscles. The clinical signs of BHP become usually evident when kittens are two to six months of age, although some cases have not been detected until 2 years of age.

Mutation that causes BHP in cats is inherited as an autosomal recessive trait. That means the disease affects cats with P/P genotype only. The cats with N/P genotype are considered carriers of the disease (heterozygotes), they are healthy but they can transmit the mutation on their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 50 % N/P and 25 % P/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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