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genetic laboratory

Result certificate #012345

Detection of c.2899C>T mutation in WNK4 gene causing hypokalemia 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.

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.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, 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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