GENOMIA
genetic laboratory

**Result certificate #012345**

Detection of c.1303G>A mutation in DPYS gene causing Dihydropyrimidinase deficiency 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: 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.1303G>A mutation in DPYS gene causing Dihydropyrimidinase deficiency in cats was examined. The disease is characterized by increased amounts of dihydrouracil and dihydrothymine in the urine. The disease is manifested by malnutrition, depression, vomiting and increased ammonia concentration in the blood.

Mutation that causes Dihydropyrimidinase deficiency 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, ngs

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

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999