

Detection of c.418dup mutation in FASLG gene causing ALPS in british shorthair 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.

**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.418dup mutation in FASLG gene causing Autoimmune lymphoproliferative syndrome (ALPS) in british shorthair cats was examined. ALPS is a condition characterized by proliferation of lymphocytes due to a disorder of physiological cell death. Excessive numbers of lymphocytes lead to the development of autoimmune disorders. The disease develops in kittens from birth until weaning at the latest, and the manifestations include marked generalised lymphadenopathy, splenomegaly and haemolytic anaemia.

Mutation that causes ALPS 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



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

Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at [www.genomia.cz](http://www.genomia.cz)

Without a written consent by the lab, the report must not be reproduced unless as a whole.

The result refers only to the tested sample, as received. Genomia is not responsible for the accuracy of the information provided by the customer.