GENOMIA
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Result certificate #012345

Detection of c.497_508del mutation in ALX1 gene causing Frontonasal dysplasia 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: 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)

Explanation

Presence or absence of c.497_508del mutation in ALX1 gene causing Frontonasal dysplasia in Burmese cats was examined. Frontonasal dysplasia is an inherited disorder of the development of the skull in the area between the forehead and nose, resulting in facial deformity. Complications include respiratory problems, eye abnormalities or eating problems.

The inheritance of the mutation is autosomal semi-dominant. This means that the phenotype of heterozygotes (N/P result) differs in the degree of expression from both affected homozygotes (P/P result) and healthy individuals (N/N result). Affected homozygous kittens are born alive; however, their condition is incompatible with life and requires euthanasia. Heterozygous cats are characterized by a brachycephalic phenotype typical of Burmese cats.

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

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