GENOMIA
genetic laboratory

Result certificate #012345

Detection of mutations in SLC7A9 gene related to cystinurie type B development 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.

Result: PREVIEW RESULT LINE

Explanation

Presence or absence of c.706G>A, c.1175C>T, c.881T>A mutations in SLC7A9 gene related to cystinurie type B development in cats were tested. Mutation c.881T>A has been identified in Maine Coon, Siamese and Siberian cats and in the Sphynx breed. Cystinuria is a disorder of amino acid transport in the kidneys. In affected cats the accumulation of cystine in the urine occurs, the formation of cystine crystals and the formation of urinary stones. These irritate the urinary tract, causing stagnation of urine and increasing the susceptibility of the urinary tract to infection. Clinically, cystinuria is manifested by dark and distinctly foul-smelling urine with an admixture of blood. Urination is painful and often unsuccessful.

The N/N result means that the cat is not at risk of cystinuria B development. The presence of mutation in homozygous (result P/P) or heterozygous (result N/P) state means that the cat is at risk of cystinuria B development. Not enough data are yet available to determine the mode of inheritance. Autosomal recessive or autosomal dominant inheritance is assumed.

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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SAMPLE