GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.693+304G>A mutation in the PKLR gene causing pyruvate kinase 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.693+304G>A mutation in the PKLR gene causing pyruvate kinase deficiency in cats (PK Def) was tested. Pyruvate kinase deficiency causes an inherited hemolytic disease. Perturbation of the regulatory enzyme pyruvate kinase decreases erythrocyte longevity and results in anaemia. Additional signs include lethargy, weakness, weight loss, jaundice and abdominal enlargement.

Mutation that causes PK Def 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, 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

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