

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: SOP175-PKdef-cat, real-time PCR-ASA

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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Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz

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