GENOMIA
genetic laboratory

Result certificate #060955:

Detection of c.693+304G>A mutation in the PKLR gene causing pyruvate kinase deficiency in cats by DNA sequencing

Sample
Sample: 15-03514
Name: IC Urschula von Skarrabäus*CH
Breed: Egyptian Mau
Date of birth: 7.7.2011
Reg. number: (CZ)ČSCH LO 1/12 MAU
Microchip: 756097200184667
Sex: female
Date received: 20.02.2015
Sample type: buccal swab
Sample certified by Vet/Tech or witness.

Customer
Jana Součková
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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: SOP144

Report date: 26.02.2015
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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