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Result certificate #095477

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

Sample
Sample: 17-16930
Name: Zmatrixu Jacco
Breed: Maine Coons
Date of birth: 17. 4. 2017
Sex: male
Date received: 21.06.2017
Sample type: buccal swab

Customer
Zuzana Žáková
Podhráz 11
53401 Holice
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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: SOP175-PKdef-cat, real-time PCR-ASA

Report date: 22.06.2017
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
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