GENOMIA
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Result certificate #080226

Detection of c.10063C>A mutation in exon 29 of PKD1 gene in cats causing PKD disease

Sample
Sample: 16-17901
Name: Antibes Dello Stretto Incantato
Breed: Birma (SBI)
Date of birth: 25.6.2015
Microchip: 985 170 001 132 979
Sex: male
Date received: 29.06.2016
Sample type: buccal swab

Customer
Miturová Kateřina
Podlesí 75
75701 Vlašské Meziříčí
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Result: Mutation was not detected (N/N)

Explanation

Presence or absence of c.10063C>A mutation in exon 29 PKD1 gene causing polycystic kidney disease (PKD) in cats was tested. The disorder presents itself as the formation of fluid-filled renal cysts. The cysts disrupt the function of kidneys and can lead to the ultimate renal failure and death of affected animal.

Feline PKD is inherited as an autosomal dominant trait. That means the disease affects all cats bearing mutated PKD1 gene (result N/P, negative / positive). There are no healthy carriers of the disease. One positive parent is enough to transmit the mutation. When mating the affected heterozygote (N/P) with the healthy individual (N/N), the mutation is transmitted in 50 % of cases. Mutated homozygous (P/P) genotype is embryonic lethal. Mutation c.10063C>A in PKD1 gene was found among Persians, Siamese, Exotic, Ragdoll, and Persian- and Exotic-outcrossed breeds (Selkirk Rex and Scottish Fold).

Method: SOP173-PKD, PCR-RFLP, accredited method
Sensitivity (probability of correct identification of the defective form of the gene in heterozygous or mutated homozygous) is higher than 99%. Specificity (probability of correct identification of the normal form of the gene in a normal homozygous or heterozygous) is higher than 99%.

Report date: 08.07.2016
Responsible person: Mgr. Barbora Bláhová, Analyst

Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

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