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Result certificate #104893

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

Sample
Sample: 18-01714
Name: Jasmín Měsíční kámen
Breed: British Shorthair
Date of birth: 2.1.2017
Microchip: 203 098 100 397 057
Sex: female
Date received: 23.01.2018
Sample type: blood

Customer
Monika Bubílková
Pšeník 5
63900 Brno
Czech Republic

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: 30.01.2018
Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

verified and certified document
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Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

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