

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

Sample

Sample: 17-08634
Name: HEREK Alfa - Pol - Cat, CZ
Breed: British Shorthair
Date of birth: 01.04.2016
Reg. number: (CZ)ČSCH LO 468/16/BSH
Microchip: 941 000 019 815 626
Sex: male
Date received: 31.03.2017
Sample type: buccal swab
The identity of the animal has been checked by
MVDr. Petra Domesová

Customer

Šárka Velčovská
Janovice 579
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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: 06.04.2017

Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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

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