GENOMIA
genetic laboratory

Result certificate #087369

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

Sample
Sample: 16-34005
Name: Charlotte Champion Mewtwo, Cz
Breed: British Shorthair
Date of birth: 7.2.2014
Microchip: 953 000 010 187 350
Sex: female
Date received: 06.12.2016
Sample type: blood
The identity of the animal has been checked by MVDr. Petra Orthová

Customer
Lucie Břízová
Za Prachárnou 11
58605 Jihlava
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: 07.12.2016
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

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

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