



Result report

Detection of C>A mutation in exon 29 of PKD1 gene in cats causing PKD by PCR-RFLP

Customer

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Details of animal

Sample: 09-15908
Animal: Nimoe von Imporio
Breed: Birma
Year of birth: 2008
ID number: ---
Sex: female
Date received: 15.06.2009
Sample type: buccal swab

Result: Mutation was not detected (N/N)

Explanation

Mutation 3284C>A in exon 29 of the 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 (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 – there is a 50 % risk of transmitting the disease. Mutated homozygous (P/P) genotype is embryonic lethal. Mutation 3284C>A in 29 exon of PKD1 gene was found among Persians, Siamese, Exotic, Ragdoll, and Persian- and Exotic-outcrossed breeds (Selkirk Rex and Scottish Fold).

Method: SOP01, 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: 18.06.2009

Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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