

Result report

Detection of mutation in PKD1 gene

Owner address

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Details of animal

Sample: 08-31201
Animal: Vanilka z Lužinek, CZ
Breed: britská krátkosrstá
Year of bird: 2007
ID number: 978000001293515
Sex: female
Date received: 26.11.2008
Sample type: buccal swab

Result: N/N

Result codes:

N/N (negative/negative) normal homozygote

N/P (negative/positive) heterozygote

P/P (positive/positive) mutated homozygote

Result interpretation

Mutation 3284C>A in 29 exon of **PKD1** gene has been examined. This mutation brings on polycystic kidney disease. 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 (P/N) 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, Persian- and Exotic-outcrossed breeds (Selkirk Rex and Scottish Fold).

Method: SOP01

Report date: 01.12.2008

Responsible person: Mgr. Martina Šafrová, head of the laboratory