

Result certificate #012345

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

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic

Sample:

Sample: 08-12346 Date received: 01.01.2008 Sample type: buccal swab

Information provided by the customer

Name: Madame Théophile DEMO

Breed: Persian catDate of birth: 31.12.1909

Reg. number: (CZ)ABCD EF 123/45/XYZ

Microchip: 123456789012345

Sex: female

Date of sampling: 01.01.2008

The identity of the animal has been checked.

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: SOPAgriseq_feline, ngs

Date of issue: 06.01.2008

Date of testing: 12.06.2008 - 06.01.2008

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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