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Result certificate #012345

Detection of mutation c.1024G>T in TRPV4 gene causing Osteochondrodysplasia in cats

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 cat
Date of birth: 31.12.1909
Reg. number: (CZ)ABCD EF 123/45/XYZ
Microchip: 123 456 789 012 345
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.1024G>T mutation in TRPV4 gene causing Osteochondrodysplasia in cats was examined. The mutation causes folding of the ears, musculoskeletal defects such as malformations of the limbs and tail tips or progressive destruction of the joints.

Inheritance of this mutation is incompletely autosomal dominant. This means that one copy of the mutated gene inherited from one of the parents is sufficient to cause the trait (result N/P). However, an affected homozygote (result P/P) who has inherited the mutation from both parents will differ from a heterozygote in the strength of the traits and the severity of the manifestations. A cat without osteochondrodysplasia has a result of N/N.

Method: SOPAgriseq_feline, MPS

Date of issue: 06.01.2008
Date of testing: 12.06.2008 - 06.01.2008
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
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