GENOMIA
genetic laboratory

Result certificate #012345

Detection of IVS50+9T>G mutation in CEP290 gene causing PRA-rdAc disease 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: 123456789012345
Sex: female
Date of sampling: 01.01.2008
The identity of the animal has been checked.

Result: Mutation was not detected (N/N)

Legend: N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

Explanation

Presence or absence of (IVS50 + 9T>G) mutation in intron 50 of CEP290 gene causing PRA-rdAc (progressive retinal degeneration, rdAc = retinal degeneration in the Abyssinian cats) in cats was tested. PRA-rdAc is a late onset form of rod-cone degeneration. Vision loss progresses slowly and is variable. In most affected animals disease occurs between 3-5 years of age. The disease affects especially cats of following breeds: Abyssinian cats, Somali cats, Occicat, American Curl, Bengal, Balinese, Colorpoint Shorthair, Cornish Rex, Munchkin, Oriental Shorthair, Peterbald, Siamese, Singapura, Tonkinese.

Mutation that causes PRA-rdAc in cats is inherited as an autosomal recessive trait. That means the disease affects cats with P/P genotype only. The cats with N/P genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

Method: SOP172-PRA-rdAc, direct DNA sequencing

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

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