GENOMIA
genetic laboratory

Result certificate #012345

Diagnostic test of spinal muscular atrophy

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 ~140 kb deletion in LIX1 and LNPEP genes, part of feline chromosome 1 (FCA1q), causing spinal muscular atrophy (SMA) in Maine Coon cat breed was tested. The progressive loss of the neuron function in the first few months of life leads to muscle weakness that first becomes apparent at 3-4 months of age. Affected kittens develop an odd gait with a sway of the hindquarters and sometimes connected with the transfer of the weight of the body to the toes. By 5-6 months of age they are too weak in the hindquarters to readily jump on furniture and are clumsy when jumping down. Length of the life depends on the severity of the affection.

Mutation that causes SMA in Maine Coon 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: SOP176-SMA, ASA-PCR

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

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999