GENOMIA
genetic laboratory

Result certificate #050883:

Detection of c.344G>A mutation in the CAPN1 gene causing LOA by PCR-RFLP

Sample
Sample: 14-24302
Name: Nikita Kelsey Libami
Breed: Parson Russell Terrier
Tattoo number: 2011
Reg. number: CLP/PRT/2011
Date of birth: 14.7.2011
Sex: female
Date received: 08.09.2014
Sample type: buccal swab

Customer
Dagmar Kynclová
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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 c.344G>A mutation in the CAPN1 gene causing LOA (Late Onset Ataxia) was tested. The Late Onset Ataxia is characterized by lack of balance and incoordination of gait. The clinical symptoms are usually noticed between 6 months and 12 months of age. The main symptoms are stiffness of hind limbs, difficulty going up the stairs and incoordination when dumping. The disease is progressive and after the onset of the first signs the problems with balance and gait incoordination are increasing rapidly. The neurological examination of the affected dogs shows symmetric spino-cerebellar ataxia - cerebellar malfunction characterized by inability to carry out precise and quick movements of skeletal muscles.

Mutation that causes LOA is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. The dogs 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.

Test does not exclude present of mutation causing another type of spinocereberal ataxia.

Method: SOP146

Report date: 10.09.2014
Responsible person: Mgr. Barbora Bláhová, Analyst

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