GENOMIA
genetic laboratory

Result certificate #050884:

Detection of c.627C>G mutation in KCNJ10 gene causing SCA by PCR-RFLP with detection by fragment analysis

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. 627C>G mutation in KCNJ10 gene causing SCA (Spinocerebellar Ataxia) in Parson Russell Terriers and Jack Russell Terriers was tested. The clinical symptoms are usually noticed between 2 to 6 months of age. The disease is caused by degeneration of spinal nerves that carry information to the cerebellum. The symptoms are very similar to signs shown by dogs affected with Late Onset Ataxia (LOA) that begin with coordination difficulties when walking, running, turning and jumping. The problems with movement coordination can often progress. The majority of cases also develop myokymia, an involuntary twitching of the muscles. The mykomia also becomes progressively worse with the age and can result in generalized muscle spasms and over-heating. Some dogs may even have true epileptic seizures.

Mutation that causes SCA 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: SOP150

Report date: 12.09.2014
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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