GENOMIA
genetic laboratory

Result certificate #024457:

Detection of c.619C>T mutation in CLN5 gene causing NCL in border collies by PCR-RFLP

Sample
Sample: 12-24630
Name: Bring me to life of Dashing Dawn
Breed: Border Collie
Reg. number: 2590/11
Microchip: 972274000073652
Date of birth: 24 August 2011
Sex: female
Date received: 07.09.2012
Sample type: buccal swab

Customer
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Result: Mutation was not detected (N/N)

Explanation

Mutation c.619C>T in CLN5 gene was tested. This mutation forms premature stopcodon; the mutated enzyme is 144 amino acids shorter and cannot full fill its normal function. Neuronal ceroid lipofuscinosis (NCL) is a neurodegenerative disorder that is characteristic by accumulation of lipopigments (coroid and lipofuscin) in the lysosomes. The beginning and clinical course of the disease vary greatly and are very individual. The rate of neurodegeneration increases together with the age, with psychical abnormalities and ataxy usually developing in all affected dogs. Increased restlessness, aggression, hallucinations, hyperactivity and epileptic attacks can be observed as well. Accompanying symptom is damaged retina due to lipopigment storage. Affected individuals rarely survive more 28th month of age.

Mutation that causes NCL in border collies is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; disease affects dogs with P/P (positive / positive) genotype only. The dogs with N/P (positive / negative) genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP39

Report date: 14.09.2012
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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