Genomia
genetická laboratoř

L 1549

Result report #002746

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

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Veterinarian Dr. Fáy József
confirmed sample origin - an optimal form.

Details of animal
Sample: 10-02102
Animal: COLLIES FROM SUSY BONITA
Breed: Border Collie
Reg. number: 926/05
Microchip: -
Year of birth: 2005
Sex: female
Date received: 25.01.2010
Sample type: blood

Result: Mutation was not detected (N/N)

Explanation

Mutation c.619G>C 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, unaccredited method
Sensitivity (probability of correct identification of the defective form of the gene in heterozygous or mutated homozygous) is higher than 99%. Specificity (probability of correct identification of the normal form of the gene in a normal homozygous or heterozygous) is higher than 99%.

Report date: 03.02.2010
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

Genomia is accredited according to ISO 17025 under #1549.
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