GENOMIA
genetic laboratory

Result certificate #050779:

Detection of c.619C>T mutation in CLN5 gene causing NCL in border collies by DNA sequencing

Sample
Sample: 14-23808
Name: Darleyfalls All Zet To Do It
Breed: Border Collie
Microchip: 981000004416756
Reg. number: DK00418/2014
Date of birth: 20.01.2014
Sex: female
Date received: 03.09.2014
Sample type: buccal swab

Customer
Jane Elene Christensen
Ibsensvej 59
2630 Taastrup
Denmark

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 mutation c.619C>T in CLN5 gene causing NCL in border collies was tested. The 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 are very individual. The rate of neurodegeneration increases together with the age. Mental abnormalities and ataxy usually develop 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 than 28th month of age.

Mutation that causes NCL in border collies is inherited autosomally recessively which means that the disease develops only in dogs who inherit mutated allele from both parents; 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.

Method: SOP38

Report date: 11.09.2014
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

