

Detection of c.619C>T mutation in CLN5 gene causing NCL5 in border collies and australian cattle dogs

Customer: Dr. Josette Debrincat, 20, Animal Doctors, Guze Bajada, HMR 2131 Hamrun, Malta

Sample:

Sample: 24-22121

Date received: 29.08.2024

Sample type: blood

Information provided by the customer

Name: Meg

Breed: Border Collie

Tattoo number: N/A

Microchip: 380 260 102 381 321

Reg. number: MKC BOR/06/2024

Date of birth: 21/11/2022

Sex: female

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 Neuronal Ceroid Lipofuscinosis type 5 (NCL5) in border collies and australian cattle dogs was tested. 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 ataxia 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 NCL5 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: SOPagrisseq_canine, ngs, accredited method

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Approved by: Mgr. Martina Šafrová, Laboratory Manager



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