

**Result certificate #012230:****Sample**

Sample: 11-21413  
Name: Koral Rubínové srdce  
Breed: Border Collie  
Reg. number: 2461  
Microchip: -  
Date of birth: -  
Sex: unknown  
Date received: 03.08.2011  
Sample type: blood  
Sampling confirmed by veterinarian.

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

Kateřina Jančaříková  
Křižíkova 983/1  
289 22 Lysá nad Labem  
Czech Republic

**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: 09.08.2011

Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager



Genomia s.r.o, Teslova 3, 30100 Plzeň, Czech Republic, IČZ: 44929000  
www.genomia.cz, laborator@genomia.cz, tel: +420 378 051 410