

Result certificate #050780:

Sample

Sample: 14-23808
Name: Darleyfalls All Zet To Do It
Breed: Border Collie
Microchip: 98100004416756
Reg. number: DK00418/2014
Date of birth: 20.01.2014
Sex: female
Date received: 03.09.2014
Sample type: buccal swab

**Detection of mutation
g.4411956_441190delGTTT in VPS13B gene
causing TNS in Border collies by fragment
analysis**

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 g.4411956_4411960delGTTT in exon 19 of VPS13B gene causing Trapped Neutrophil Syndrome (TNS) in Border collie breed was tested. Due to this mutation the correct function of white corpuscles - neutrophils - is impaired. They take part in fighting bacterial infections and are important participants in acute inflammation. The failing of immune system can be seen in pups from as early as 2 weeks old and the pups die or are euthanized by approx. 4 months of age. The first symptoms may include apathy, loss of appetite, diarrhoea or poor mobility. Other symptoms depend on the type of infection the pup happens to contract.

Mutation that causes TNS in border collies is inherited as an autosomal recessive trait. That means the 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: SOP88, accredited 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: 11.09.2014

Responsible person: Mgr. Barbora Bláhová, Analyst



Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

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

