

Result certificate #024458:

Sample

Sample: 12-24630
Name: Bring me to life of Dashing Dawn
Breed: Border Collie
Reg. number: 2590/11
Microchip: 972274000073652
Date of birth: 24 August 2011
Sex: female
Date received: 07.09.2012
Sample type: buccal swab

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

Customer

Diana Krassovari
Lonyai utca 1.
2330 Dunaharaszti
Hungary

Result: Mutation was not detected (N/N)

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 (positive/positive) genotype only. The dogs with N/P (negative/positive) 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: 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: 15.09.2012

Responsible person: Mgr. Martina Šafrová, Laboratory Manager


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Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic, VAT#: CZ25212991
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

