GENOMIA
genetic laboratory

Result certificate #050777:

Detection of c.8392delC mutation in the CUBN gene causing IGS 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
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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 c.8392delC mutation in the CUBN gene causing IGS (Imerslund-Gräsbeck syndrome) or intestinal cobalamin malabsorption in border collies was tested. IGS is metabolic disorder in border collies. Signs appear early in 6 to 12 week of dog´s age and include failure to thrive and chronic loss of appetite. The affected dogs can suffer from neutropia, non-regenerative anaemia, anisocytosis and poikilocytosis, megaloblastic changes in bone marrow, reduction of Cbl level, methylmalonic aciduria and homocysteinemia.

Mutation that causes IGS 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), they are healthy but they can transmit the mutation on their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 50 % N/P and 25 % P/P.

Method: SOP137

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

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