

Detection of c.8392delC mutation in the CUBN gene causing IGS in border collies

**Customer:** Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic

**Sample:**

Sample: 21-12345

Date received: 01.02.2021

Sample type: blood

Information provided by the customer

**Name:** Lassie DEMO

**Breed:** Plemeno

Tattoo number: 1392013

Microchip: 123 456 789 012 345

Reg. number: REGQ12345

Date of birth: 1.1.2020

Sex: female

Date of sampling: 01.02.2021

The identity of the animal has been checked.

**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: SOPAgriseq\_canine, ngs

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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