GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.786delC mutation in CUBN gene causing IGS in beagles

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.786delC mutation in CUBN gene causing IGS (Imerslund-Gräsbeck syndrome), intestinal cobalamin malabsorption, in beagles was tested. IGS is a metabolic disorder in beagles. Signs begin around 6 to 12 weeks of age, and include failure to thrive and chronic inappetance. Affected animals also demonstrate neutropenia, nonregenerative anemia, anisocytosis and poikilocytosis, megaloblastic changes of the bone marrow, decreased serum Cbl concentrations, methylmalonic aciduria, and homocysteinemia. Beagles with cobalamin deficiency develop a degenerative liver disease.

Mutation that causes intestinal cobalamin malabsorption in beagles 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 to their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

Method: SOPAgriseq_canine, ngs, accredited method

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
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