GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.107G>C mutation of ITGB2 gene causing CLAD disease in Irish Setters

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.

SAMPLE

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 mutation c.107G>C in exon 3 of ITGB2 gene (beta-2-integrin gene) causes CLAD (canine leukocyte adhesion deficiency) in Irish Setters was tested. The mutation causes Cys36Ser substitution in beta-2-integrin protein molecule; that leads to wrong protein conformation and function disruption. Integrins are located on the surface of white blood cells. Integrins intermediate cell-cell and cell-substratum adhesion reactions in the body. Integrins help leukocytes to incorporating into inflammatory tissues. If a part of integrin molecule is destroyed, there is a lack of immune response to the presence of infection in the body.

Mutation that causes CLAD in Irish Setters 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: SOP188-MPS-canine, MPS, 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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