GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.400+3A>C mutation in MKLN1 gene causing LAD in Bull Terriers and Miniature Bull Terriers

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.400+3A>C mutation in MKLN1 gene causing Lethal Acrodermatitis (LAD) in Bull Terriers and Miniature Bull Terriers was tested. LAD is a complex immune deficiency that is caused by defective zinc metabolic pathway or zinc absorption. Disease is characterized by severe retardation of growth and development, immune deficiency and skin lesions. Unpleasant lesions and swelling appear especially on the muzzle, around the eyes and ears. The skin on the feet is hard and cracked and crusted skin lesions and cracks develop on the footpads. The disease is often accompanied with diarrhoea and pneumonia. The clinical signs of LAD appear after the first week from the birth of the puppies. The affected dogs usually die before they reach two years of age and that due to infection or they are euthanized because of severe and painful lesions on the feet.

Mutation that causes LAD is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. The dogs with N/N genotype are healthy. 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

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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