

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

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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