

**Detection of c.191+1G>A mutation in  
LMBR1L gene causing  
Hyposegmentation of granulocytes  
in Australian Shepherds****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.191+1G>A mutation in LMBR1L gene causing Hyposegmentation of granulocytes in the Australian Shepherds was tested. Granulocytes are a type of white blood cell whose main function is to help the body fight against infection. Hyposegmentation is a condition in which the nuclei of granulocytes have fewer segments than normal, resulting in a change in their shape and function.

Mutation that causes Hyposegmentation of granulocytes 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:** 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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Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at [www.genomia.cz](http://www.genomia.cz)

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