

**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)

**Explanation**

Presence or absence of c.1579G>A mutation in PLA2G2 gene causing Neuroaxonal dystrophy (NAD) in Papillon and Phalene breed was tested. NAD belongs to the group of rare inherited neurodegenerative diseases. The first symptoms appear around 3 months of age. There can be observed problems with gait, even complete limb paralysis, loss of coordination, head tremor or intention tremor, inability to stand. Further neurological symptoms occur from the 6 month of age, such as blindness, strabismus, loss of reaction to threat, difficulty swallowing and progressive cerebellar atrophy.

Mutation that causes NAD 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 on their offspring. Dogs with N/N genotype are without risk of NAD. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 50 % N/P (healthy carriers) and 25 % P/P (affected).

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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