

## Result certificate #012345

Detection of c.2504A>G mutation in VPS11
gene causing NAD in Rottweilers

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.2504A>G mutation in VPS11 gene causing Neuroaxonal dystrophy (NAD) in Rottweilers was tested. The first symptoms appear in puppies at approx. 3 months of age. There can be observed problems with gait (it may develop partial or complete limb paralysis), loss of coordination, head tremor or intention tremor, inability to stand (astasia) and further neurological symptoms occur from the 6th month of age, such as blindness, strabismus, loss of reaction to threat, difficulty swallowing and progressive celebellar atrophy.

Mutation that causes NAD is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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



Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999