

## Result certificate #012345

Detection of c.367delG mutation in RNF170 gene causing NAD in Miniature American 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.367delG mutation in RNF170 gene causing Neuroaxonal Dystrophy (NAD) in Miniature American Shepherds was tested. NAD is a slowly progressive neurodegenerative syndrome. Affected dogs are typically young adults and show characteristic gait abnormalities, pelvic limb weakness and ataxia. However, despite these disabilities, the dogs have a relatively long lifespan.

Mutation that causes Neuroaxonal Dystrophy 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: SOP171-NAD\_MAS, fragment analysis

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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