

## Result certificate #012345

Detection of c.866G>A mutation in ALDH5A1 gene causing SSADHD in Saluki

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.866G>A mutation in ALDH5A1 gene causing succinic semialdehyde dehydrogenase deficiency (SSADHD) in Saluki was tested. In the affected dogs, neurological abnormalities, including seizures and behavioural changes can be observed. They show atrophy of the cerebral cortex with vacuolization, increased levels of gamma-hydroxybutyric acid in blood serum, cerebrospinal fluid and brain, and increased levels of succinic semialdehyde in urine, cerebrospinal fluid, and brain.

Mutation that causes SSADHD 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, accredited method

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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