GENOMIA
genetic laboratory

# 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.

SAMPLE

## 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: SOP188-MPS-canine, MPS, 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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