

# Result certificate #012345

Detection of c.1728C>A mutation in ACADVL gene causing EIMM in German Hunting Terrier

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 mutation c.1728C>A in ACADVL gene causing exercise induced metabolic myopathy (EIMM) in German Hunting Terrier was tested. EIMM is a hereditary metabolic disease that results in weakness of limb and profound muscle pain during physical exercise (e.g during a hunt).

Further, the disease is characterized by increased level of creatine kinase in blood plasma that is a sign of muscle cell damage and by a rhabdomyolyse i.e. rapid breakdown of skeletal muscles resulting in release of the degradation products (mainly myoglobin) into bloodstream and subsequently into urine. The affected dogs show dark urine during and shortly after physical exercise as a consequence of the presence of hemoglobin or erythrocytes in urine. The symptoms of this disease are usually induced by stress, where the stress situation is not only physical exertion, but also exposure to cold.

Mutation that causes EIMM in German Hunting Terrier 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



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