GENOMIA
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Result certificate #012345

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

Sample
Sample: 08-12345
Name: Lassie DEMO
Breed: ---
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 31.12.1909
Sex: female
Date received: 25.11.2008
Sample type: blood
The identity of the animal has been checked.

Customer
Jan Novák
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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 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: SOP173-EIMM, PCR-RFLP

Report date: 25.11.2008
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
GENOMIA genetic laboratory

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