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Result certificate #012345

Detection of IVS10-1G>A BIN1 mutation in BIN1 gene causing IMGD in Great Danes

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic
Sample:
Sample: 08-12345
Date received: 25.11.2008
Sample type: blood

Information provided by the customer
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 of sampling: 25.11.2008
The identity of the animal has been checked.

Result: Mutation was not detected (N/N)

Explanation

Presence or absence of IVS10-1G>A mutation in BIN1 gene causing inherited myopathy in Great Danes was tested. Symptoms of degenerative muscle disorder begin between 3 and 6 months of age. The first signs include exercise intolerance, weakness and tremors. Frequent signs are overall stiff gait and short stride. In the progressive stage of the disease, the exercise results in exhaustion to the point of collapse. The clinical signs get worse under load, stress and low temperatures. The severity of symptoms and the progression rate are variable.

Mutation that causes IMGD is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P (positive/positive) genotype only. The dogs with N/P (negative/positive) genotype are considered carriers of the disease (heterozygotes), they are healthy but they can transmit the mutation on their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 50 % N/P (healthy carriers) and 25 % P/P (affected).

Method: SOP172-IMGD, direct DNA sequencing

Date of issue: 30.11.2008
Date of testing: 25.11.2008 - 30.11.2008
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document
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