

Result certificate #012345

Detection of c.4223delA mutation in AGL gene causing GSDIlla in Curly Coated Retrievers

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 c.4223delA mutation in AGL gene causing GSDIlla in Curly Coated Retrievers was tested. Clinical signs of GSD are mild in the first year of life. The abnormally structured glycogen is progressively stored in liver and muscles. The signs usually develop after the first year of life, the dogs are lethargic, show intolerance to exercises, episodic hypoglycemia and collapses occur. The storage in livers may lead progressively to cirrhosis, storage in muscles results in progressive muscle and hearth atrophy.

Mutation that causes GSDIlla in Curly Coated Retrievers is inherited as an autosomal recessive trait. That means the 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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