

Detection of c.634\_635insN[76] mutation in  
G6PC gene causing GSD1a in German  
Pinschers

**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.634\_635insN[76] mutation in G6PC gene causing Glycogenosis (GSD1a) in the German Pinschers was tested. Glycogenosis is a severe disorder of glycogen metabolism characterized by accumulation of glycogen, especially in the liver. Symptoms include severe liver enlargement, general failure to thrive, lethargy, coma and death.

Mutation that causes GSD1a 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: SOP176-GSD1, ASA-PCR

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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