

Result certificate #012345

Detection of c.964G>A mutation in SLC7A9

gene causing cystinuria type II-B

in Miniature Pinscher

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)

Explanation

Presence or absence of c.964G>A mutation in SLC7A9 gene causing cystinuria type II-B in Miniature Pinscher was tested. In affected individuals, cystine accumulates in the urine and leads to cystine crystals and urinary stones formation. These irritate the urinary tract, causing urinary stagnation and increasing the susceptibility of the urinary tract to infection. Clinically, cystinuria is manifested by dark and distinctly foul-smelling urine with an admixture of blood. Urination is painful and often unsuccessful.

Inheritance of the causal mutation of cystinuria type II-B is autosomal dominant. This means that it only takes one copy of the mutated gene inherited from one parent to cause symptoms of the disease.

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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