

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.996G>A mutation in AGXT gene causing PH I (Primary hyperoxaluria type I) in Coton de Tulear breed was tested. The PH disease is caused by insufficient function of a liver-specific enzyme alanine-glyoxalate aminotransferase (AGT, AGXT). PH is characterized by the accumulation of oxalate and subsequent precipitation of calcium oxalate crystals, primarily in the kidneys, leading to progressive kidney failure. If the storage capacity of the kidneys is exhausted, the crystals are accumulated in other tissues, for example in bones, joints, cartilages, retina and muscles.

Mutation that causes PH I in Coton de Tulear 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: SOP173-PHI, PCR-RFLP

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

Report verification code is: 12AB-CD34-GENO-MIA0-EFGH. You can verify report online at www.genomia.cz

Without a written consent by the lab, the report must not be reproduced unless as a whole.

The result refers only to the sample as received. Genomia is not responsible for the accuracy of the information provided by the customer.