

Detection of c.9772G>A mutation in the PKD1 gene causing BTPKD in English Bull Terriers

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.9772G>A mutation in PKD1 gene causing polycystic kidney disease (BTPKD) in English Bull Terriers was tested. The disease is characterized by the formation of fluid-filled cysts in the kidneys. These cysts endanger the function of kidneys and may result in kidney failure and death of the affected animal.

Mutation that is responsible for BTPKD is inherited as an autosomal dominant trait. That means the disease affects all dogs carrying mutated PKD1 gene (dogs with result N/P, negative / positive). When mating the affected heterozygote (N/P) with the healthy individual (N/N), the mutation is transmitted in 50 % of cases.

Method: SOPAgriseq_canine, ngs, accredited method

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549

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 tested sample, as received. Genomia is not responsible for the accuracy of the information provided by the customer.