

## Result certificate #012345

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: 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



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

