GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.1572+5G>A mutation in INPP5E gene causing HRFCD in Norwich Terrier

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.1572+5G>A mutation in INPP5E gene causing diffuse cystic renal dysplasia and hepatic fibrosis (HRFCD) in Norwich Terrier was tested. The disease is also referred to as hepatorenal fibrocystic disorder. It is a disease caused by defects in the morphology or function of cilia, the cellular organelles that are used for intercellular communication and transport of signalling molecules. The disease is characterized by the formation of cysts in the straight portion of the proximal tubule, and in the thin descending and ascending limbs of Henle´s loop and is lethal to puppies.

Mutation that causes HRFCD is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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: 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

verified and certified document
GENOMIA genetic laboratory

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