GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.1098_1103del mutation in SLC3A1 gene causing cystinuria type II-A in Australian Cattle Dogs and Koolies

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.1098_1103del mutation in SLC3A1 gene causing cystinuria type II-A in Australian Cattle Dogs and Koolies 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-A 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, 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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