GENOMIA
genetic laboratory

Result certificate #012345

Detection of mutations in SLC3A1 a SLC7A9 genes causing cystinuria in English and French Bulldogs

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 interpretation D
Genotype found: c.574A/A; c.2092A/A; c.649G/G

Explanation

Presence or absence of mutations c.574A>G in exone 2 and c.2092A>G in exone 10 of SLC3A1 gene and mutation c.649G>A in SLC7A9 gene causing cystinuria in English and French Bulldogs were tested. Possible interpretations of genetic test results:

A) Dogs carrying homozygous mutation in exone 2 and/or in exone 10 of SLC3A1 gene are at risk of cystinuria development, it means dogs with c.574G/G and/or c.2092G/G result. Cystinuria can also develop in dogs that carry the homozygous mutation c.649A/A or the heterozygous c.649G/A mutation in the SLC7A9 gene (incomplete recessive disorder). We recommend mating only with partners who do not carry any of these mutations.

B) Carriers at risk of cystinuria development are heterozyous in one or more mentioned mutations and always carry c.649G/A, it means dogs with c.574A/G and/or c.2092A/G and always c.649G/A result. We recommend mating only with partners who do not carry any of these mutations.

C) Carriers of cystinuria are heterozyous in one or two tested mutations of SLC3A1 gene, it means dogs with c.574A/G and/or c.2092A/G and c.649G/G result. We recommend mating only with partners who do not carry any of these mutations.

D) Dogs with c.574A/A, c.2092A/A, c.649G/G result are out of risk of cystinuria development (i.e. these mutations dependent cystinuria).

The test does not exclude existence of another, nowadays unknown, mutation which can cause cystinuria.

Method: SOP188-MPS-canine, MPS

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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SAMPLE