

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.

Mutations found: c.574A/G; c.2092A/G; c.649G/A

Result interpretation B

Explanation

Presence or absence of mutations c.574A>G in exon 2 and c.2092A>G in exon 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 exon 2 and/or in exon 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 heterozygous 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 heterozygous 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: SOP172-SLC3A1,SOP173-SLC3A1-SLC7A, direct DNA sequencing

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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