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Result certificate #012345

Detection of g.38013703_38014019del mutation in exone 14 and 3'UTR region of FAN1 gene causing Fanconi syndrome in Basenji

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 g.38013703_38014019del mutation in exone 14 and 3'UTR region of FAN1 gene causing Fanconi syndrome in Basenji was tested. The Fanconi syndrome (FS) is a disease characterized by defects in epithelial transport of solutes in kidney proximal tubule. The impaired reabsorption of amino acids, glucose, sodium, calcium and phosphorus leads to acid-base balance disturbance and causes metabolic acidosis, aminoaciduria and increased glucosuria and phosphaturia.

Mutation that causes Fanconi syndrome in Basenji is inherited as an autosomal recessive trait. That means the 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOPAgriseq_canine, ngs

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
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