

Detection of g.38013703_38014019del
mutation in exon 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 exon 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: SOP176-FS, ASA-PCR

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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