

Detection of c.383del mutation in MOCOS gene causing Xanthinuria II in Cavalier King Charles Spaniels and English Cocker Spaniels

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 c.383del mutation in MOCOS gene causing Xanthinuria II in Cavalier King Charles Spaniards and English Cocker Spaniels was tested. Xanthinuria is characterized by the excretion of large amounts of xantine into urine, which then cause xanthin urinary stones. These stones irritate the urinary tract, cause urine stagnation, increase the susceptibility of urinary tract to infection and may lead to secondary kidney damage. Urine is dark and strongly smelly, with blood admixture. Urination is painful and often unsuccessful. The first symptoms are evident in the age range of 7 weeks up to 4 years.

Mutation that causes Xanthinuria II is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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, 25 % P/P and 50 % N/P.

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



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