GENOMIA
genetic laboratory

Result certificate #012345

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.

SAMPLE

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. Xantinuria 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: SOP188-MPS-canine, MPS, accredited method

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
GENOMIA genetic laboratory

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