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Result certificate #148771

Detection of c.115A>T mutation in exon 3 of COL4A4 gene causing Familiar Nephropathy in English Cocker Spaniels

Customer: Gerda van Empel, Dollemansstraat 2, 7223 KG BAAK, Netherlands

Sample:
Sample: 20-11922
Date received: 18.05.2020
Sample type: buccal swab

Information provided by the customer
Name: Eos Euphoria of Anton
Breed: English Cocker Spaniel
Microchip: 807 050 000 006 099
Reg. number: 3186435
Date of birth: 02-12-2018
Sex: male

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.115A>T mutation in exon 3 of the COL4A4 gene causing familial nephropathy (FN) in English Cocker Spaniels was tested. FN disorder is a fatal renal disease. Kidney failure arises between 6 months and 2 years of age of the dog. The first observed symptoms include vomiting, loss of appetite, excessive water consumption, and weight gain or loss.

Mutation that causes FN 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: SOP175-FN, real-time PCR-ASA, accredited method

Date of issue: 26.05.2020
Date of testing: 18.05.2020 - 26.05.2020
Approved by: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

verified and certified document
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