

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

Sample

Sample: 16-36307
Name: Courtmaster Wizard Of Oz
Breed: English Cocker Spaniel
Microchip: 981 000 008 167 618
Reg. number: 3015440
Date of birth: 13-10-2013
Sex: male
Date received: 27.02.2017
Sample type: buccal swab

Customer

Gerda van Empel
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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

Report date: 27.02.2017

Responsible person: Mgr. Martina Šafrová, Laboratory Manager



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