

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

Sample

Sample: 19-02823
Name: Nickel and Dimes False Alarm
Breed: English Cocker Spaniel
Microchip: 528 140 000 583 853
Reg. number: 2974837
Sex: female
Date received: 04.02.2019
Sample type: buccal swab

Customer

Gerda van Empel
Dollemanstraat 2
7223 KG BAAK
Netherlands

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: 06.02.2019

Responsible person: Mgr. Martina Šafrová, Laboratory Manager



Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

