GENOMIA
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Result certificate #077330:

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

Sample
Sample: 16-10405
Name: Marveil Zucchero
Breed: English Cocker Spaniel
Microchip: 380 260 080 295 807
Reg. number: 3015442
Date of birth: 16-11-2014
Sex: male
Date received: 22.04.2016
Sample type: buccal swab

Customer
Gerda van Empel
Dollemansstraat 2
7223kg 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 ASA-PCR, accredited method

Report date: 26.04.2016
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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

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