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Result certificate #106816

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

Sample
Sample: 18-05220
Name: Spendles Time Slips Away
Breed: English Cocker Spaniel
Microchip: 953 010 001 672 915
Reg. number: 3087275
Date of birth: 02-01-2017
Sex: male
Date received: 15.03.2018
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: 20.03.2018
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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

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