

Result certificate #024990:

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

Sample

Sample: 12-26006
Name: ALBERT Z VEJMINKU
Breed: English Cocker Spaniel
Reg. number: 35301
Microchip: -
Date of birth: 29.08.2010
Sex: male
Date received: 17.09.2012
Sample type: buccal swab

Customer

Gerda van Empel
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Result: Mutation was not detected (N/N)

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 (positive/positive) genotype only. The dogs with N/P (positive/negative) genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP19, accredited method

Report date: 24.09.2012

Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager


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