

## **Result report**

### **Detection of A115T mutation in exon 3 of COL4A4 gene causing FN in English Cocker Spaniels by DNA sequencing**

#### **Customer**

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#### **Details of animal**

Sample: 09-12201  
Animal: Sonique Schönez  
Breed: English Cocker Spaniel  
Reg. number: 33047  
Microchip: 978000000658135  
Year of birth: 2006  
Sex: female  
Date received: 06.05.2009  
Sample type: blood

### **Result: Mutation was not detected (N/N)**

#### **Explanation**

Mutation A115T 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.

FN is inherited as an autosomal recessive trait. That means the 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 (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP19, unaccredited method

Report date: 15.05.2009

Responsible person: Mgr. Markéta Dajbychová, Veterinary Laboratory Manager