

**Result certificate #022126:**

**Detection of 2601\_2602insC mutation in SLC4A3 gene causing GR-PRA disease in Golden Retrievers by fragment analysis**

**Sample**

Sample: 12-18301  
Name: Flying Heart Famous Gold  
Breed: Golden Retriever  
Reg. number: 13793  
Microchip: 972 270 000 034 870  
Date of birth: 17.09.2009  
Sex: female  
Date received: 02.07.2012  
Sample type: buccal swab  
When sampling the identity was verified.

**Customer**

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

**Explanation**

Presence or absence of 2601\_2602insC in SLC4A3 gene causing GR-PRA (Golden Retriever Progressive Retinal Atrophy) was tested. Disease is characterized by loss of vision due to degeneration of the photoreceptor cells of the retina. Most PRA cases in Golden Retrievers are clinically indistinguishable from other forms of PRA. The age of diagnosis is most commonly at a relatively late age of approximately 6 years.

Mutation that causes GR-PRA is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P (positive/positive) genotype only. The dogs with N/P (negative/positive) 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: SOP100

Report date: 10.07.2012

Responsible person: Mgr. Kateřina Štampachová, Analyst



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