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Result certificate #087342

Detection of 2601_2602insC mutation in SLC4A3 gene causing GR-PRA1 disease in Golden Retrievers

Sample
Sample: 16-33934
Name: FINNA Bella Aurea
Breed: Golden Retriever
Microchip: 941 000 016 041 679
Reg. number: SPKP 3007
Date of birth: 23.3.2015
Sex: female
Date received: 16.01.2017
Sample type: buccal swab

Customer
Dušan Kollárik
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03811 Belá-Dulice
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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 2601_2602insC in SLC4A3 gene causing GR-PRA1 (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 GR-PRA1 cases 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-PRA1 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, 25 % P/P and 50 % N/P.

Method: SOP171-GRPRA1, fragment analysis

Report date: 23.01.2017
Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

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