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Result certificate #112758

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

Sample
Sample: 18-17518
Name: Dasty od Rybníka Kamenný
Breed: Golden Retriever
Microchip: 941 000 019 285 674
Reg. number: ČLP/GR/18430
Date of birth: 8.7.2016
Sex: male
Date received: 29.06.2018
Sample type: blood
The identity of the animal has been checked by MVDr. Rostislav Klapka

Customer
Ing. Václav Frgal
Staré Město 259
79201 Staré Město
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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: 06.07.2018
Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

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