

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

Sample

Sample: 16-30035
Name: Phoebe Jane Tonkin Artemis Gold
Breed: Golden Retriever
Microchip: 972 270 000 321 892
Reg. number: ČLP/GR/17452
Date of birth: 4.5.2015
Sex: female
Date received: 01.11.2016
Sample type: buccal swab
The identity of the animal has been checked by
MVDr. Marie Štekllová

Customer

Míšek Pavel
Radošov 192
36272 Kyselka
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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: 08.11.2016

Responsible person: Mgr. Martina Šafrová, Laboratory Manager



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