GENOMIA
genetic laboratory

Result certificate #215107

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

Customer: Míšek Pavel, Radošov 192, 36272 Kyselka, Czech Republic
Sample:
Sample: 22-31704
Date received: 15.11.2022
Sample type: buccal swab

Information provided by the customer
Name: Nelson Golden Victory
Breed: Golden Retriever
Tattoo number: BNY 4513
Microchip: 643 099 011 505 604
Reg. number: ČLP/GR/23461
Date of birth: 11.06.2021
Sex: male
Date of sampling: 14.11.2022
The identity of the animal has been checked by MVDr. Sabina Marie Šteklová, KVL 4726

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.

If there is a reason to believe that the dog can suffer from retinal atrophy, it is recommended that the dogs are tested for GR-PRA1 together with GR-PRA2 and PRA-prcd. It is highly probable that other mutation responsible for this disease will be discovered in future.

Method: SOP171-GRPRA1,2, fragment analysis

Date of issue: 22.11.2022
Date of testing: 15.11.2022 - 22.11.2022
Approved by: Mgr. Martina Šafrová, Laboratory Manager

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999