

Detection of c.669delA mutation in TTC8 gene causing GR-PRA2 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 Štekllová, 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 c.669delA mutation in TTC8 gene causing GR-PRA2 (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-PRA2 cases are clinically indistinguishable from other forms of PRA. The age of diagnosis is most commonly at a relatively late age of approximately 5 years.

Mutation that causes GR-PRA2 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-PRA2 together with GR-PRA1 and PRA-prcd. It is highly probable that other mutation responsible for this disease will be discovered in future.

Method: SOPAgriseq_canine_TD, ngs

Date of issue: 25.11.2022

Date of testing: 15.11.2022 - 25.11.2022

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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