GENOMIA
genetic laboratory

Result certificate #290768

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

Customer: Dušan Kollárik, Belá 33, 03811 Belá-Dulice, Slovak Republic
Sample:
Sample: 25-05401
Date received: 04.03.2025
Sample type: buccal swab

Information provided by the customer
Name: Heidi Bella Aurea
Breed: Golden Retriever
Microchip: 941 000 026 557 633
Reg. number: SPKP 4826/24
Date of birth: 17.6.2023
Sex: female
Date of sampling: 28.02.2025
The identity of the animal has been checked by MVDr. Juraj Chorváth, 0011

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: SOPAgriseq_canine, ngs

Date of issue: 16.03.2025
Date of testing: 04.03.2025 - 16.03.2025
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

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