

Result certificate #218645

Detection of c.5716G>A mutation in COL7A1 gene causing Epidermolysis Bullosa in Golden Retrievers

Customer: Dušan Kollárik, Belá 33, 03811 Belá-Dulice, Slovak Republic **Sample:** Sample: 23-01202 Date received: 23.01.2023 Sample type: buccal swab

Information provided by the customer Name: GIANNA Bella Aurea

Breed: Golden Retriever Microchip: 941 000 023 870 583 Reg. number: SPKP 3603/21 Date of birth: 15.5.2019 Sex: female Date of sampling: 19.01.2023 The identity of the animal has been checked by MVDr. Lenka Blahušiaková, KVL 0814

Result: Mutation was not detected (N/N)

Legend: N/N = wild-type genotype. N/P = carrier of the mutation. <math>P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

Explanation

Presence or absence of c.5716G>A mutation in COL7A1 gene causing Epidermolysis Bullosa in Golden Retrievers was tested. This is a skin disease characterized by the formation of blisters, abrasions, and scars all over the body. The skin is fragile, cornified and hard in places, and very susceptible to damage. Symptoms appear in new-born or very young puppies.

Mutation that causes Epidermolysis Bullosa in Golden Retrievers is inherited autosomally recessively which means that the disease develops only in those dogs who inherit mutated allele from both parents; 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: SOPAgriseq_canine_TD, ngs

Date of issue: 06.02.2023 Date of testing: 23.01.2023 - 06.02.2023 Approved by: Mgr. Martina Šafrová, Laboratory Manager



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