

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 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, ngs, accredited method

Date of issue: 14.03.2025

Date of testing: 04.03.2025 - 14.03.2025

Approved by: Mgr. Markéta Dajbychová, Deputy Laboratory Manager



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