

Detection of c.1445\_1447delinsTACTACTA mutation in PNPLA1 gene causing ichthyosis 1 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 c.1445\_1447delinsTACTACTA mutation in PNPLA1 gene causing ichthyosis 1 in Golden Retrievers breed was tested. Puppies suffering from ichthyosis 1 have scaly skin soon after birth. The skin scaling lasts through the whole life of the animal. The scales become dark and the skin dry and rough with the age of the animal. This disease does not usually cause itching. In severely affected animals, the disease can be complicated by secondary bacterial, fungal or parasitic infections. Golden retrievers also have recessively inherited ichthyosis 2, which is caused by a mutation in the ABHD5 gene.

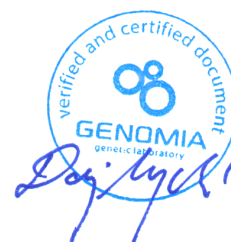
Mutation that causes ichthyosis 1 is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P (positive/positive) genotype only. The dogs with N/P (negative/positive) genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

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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