

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.1451_1453delinsTACTACTA mutation in PNPLA1 gene causing ichthyosis in Golden Retriever breed was tested. In puppies of Golden Retrievers affected by ichthyosis, skin scaling is evident 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.

Mutation that causes ichthyosis in Golden Retriever 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: SOP171-ICTA, fragment analysis

Date of issue: 22.11.2022

Date of testing: 15.11.2022 - 22.11.2022

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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