

Result certificate #234064

Detection of c.1006_1019del mutation in ABHD5 gene causing ichthyosis 2 in Golden Retrievers

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 Šteklová, KVL 4726

Result: Mutation was not detected (N/N)

Explanation

Presence or absence of c.1006_1019del mutation in ABHD5 gene causing ichthyosis 2 in Golden Retrievers breed was tested. The first symptoms of skin keratinisation begin to develop soon after birth. The degree of symptom expression varies between individuals – from an initial slight formation of light-coloured scales to the gradual formation of larger areas of dark scales. In some cases, secondary infections occur in the affected areas of the skin due to bacteria or fungi. Golden retrievers also have recessively inherited ichthyosis 1, which is caused by a mutation in the PNPLA1 gene.

Mutation that causes ichthyosis 2 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

Date of issue: 11.07.2023 Date of testing: 23.06.2023 - 11.07.2023 Approved by: Mgr. Martina Šafrová, Laboratory Manager



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