GENOMIA
genetic laboratory

Result certificate #218833

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: 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 detected in heterozygous status (N/P)

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_TD, ngs

Date of issue: 03.02.2023
Date of testing: 23.01.2023 - 03.02.2023
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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