GENOMIA
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Result certificate #112757

Detection of c.1451_1453delinsTACTACTA mutation in PNPLA1 gene causing ichthyosis in Golden Retriever

Sample
Sample: 18-17518
Name: Dasty od Rybníka Kamenný
Breed: Golden Retriever
Microchip: 941 000 019 285 674
Reg. number: ČLP/GR/18430
Date of birth: 8.7.2016
Sex: male
Date received: 29.06.2018
Sample type: blood
The identity of the animal has been checked by MVDr. Rostislav Klapka

Customer
Ing. Václav Frgal
Staré Město 259
79201 Staré Město
Czech Republic

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.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, accredited method

Report date: 06.07.2018
Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

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Genomia is accredited according to ISO/IEC 17025:2005 under #1549.

Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

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