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

Sample
Sample: 08-12345
Name: Lassie DEMO
Breed: ---
Tattoo number: 1392013
Microchip: 123 456 789 012 345
Reg. number: REGQ12345
Date of birth: 31.12.1909
Sex: female
Date received: 25.11.2008
Sample type: blood
The identity of the animal has been checked.

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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, accredited method
Report date: 25.11.2008
Responsible person: Mgr. Martina Šafrová, Laboratory Manager

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