

Detection of c.1445\_1447delinsTACTACTA  
mutation in PNPLA1 gene causing  
ichthyosis 1 in Golden Retrievers

**Customer:** Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic

**Sample:**

Sample: 21-12345

Date received: 01.02.2021

Sample type: blood

Information provided by the customer

**Name:** Lassie DEMO

**Breed:** Plemeno

Tattoo number: 1392013

Microchip: 123 456 789 012 345

Reg. number: REGQ12345

Date of birth: 1.1.2020

Sex: female

Date of sampling: 01.02.2021

The identity of the animal has been checked.

**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.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, ngs, accredited method

Date of issue: 06.02.2021

Date of testing: 01.02.2021 - 06.02.2021

Approved by: Mgr. Martina Šafrová, Laboratory Manager



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