GENOMIA
genetic laboratory

Result certificate #012345

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

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, accredited method

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

verified and certified document GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

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