GENOMIA
genetic laboratory

# Result certificate #218834

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