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Result certificate #290763

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: 25-05401
Date received: 04.03.2025
Sample type: buccal swab

Information provided by the customer
Name: Heidi Bella Aurea
Breed: Golden Retriever
Microchip: 941 000 026 557 633
Reg. number: SPKP 4826/24
Date of birth: 17.6.2023
Sex: female
Date of sampling: 28.02.2025
The identity of the animal has been checked by MVDr. Juraj Chorváth, 0011

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: 14.03.2025
Date of testing: 04.03.2025 - 14.03.2025
Approved by: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

verified and certified document
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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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