

Ichthyosis (Golden Retriever Type 2)

Client Name:	Lauriaan Morton (LAU002)	Report No:	ZO2024/7488/20240611/#72403
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Profile:	DG2023/47485	Species:	Canis lupus familiaris / Canine / Dog
Name:	Pawshappyness Kiara of Mortonouro	Microchip #:	981 020 002 401 782
Breed:	Golden Retriever	Registration #:	ZA002971B23
Test:	[ABHD5] Ichthyosis (Golden Retriever Type 2)		
Results:	c.1006_1019del	WT/WT	Clear

Sample Type: Whole Blood (EDTA)	Extraction Method: DNA Extraction: D4069	Test Type: Genetic Health
[ABHD5] Ichthyosis (Golden Retriever Type 2) Ichthyoses are a group of inherited cornification skin disorders characterized by generalized dry skin, scaling and hyperkeratosis. Signs and symptoms may present in affected dogs from as early as 1-2 weeks of age, and may include flaky skin and a scruffy haircoat when compared to normal littermates. The skin of the abdomen may appear reddened, wrinkled, and covered by light brown scales. Scaled skin may become red-brown with brown scales adhered to the skin, and white to tan coloured scales throughout the coat. Affected dogs may also develop thickened footpads, and are at an increased risk for infections of the skin, ears, and footpads. This test detects the presence or absence of a 14 nucleotide deletion (GACTTCAACCAGAA; denoted as c.1006_1019del) in the ABHD5 gene. This deletion results in a frameshift and premature stop codon leading to a truncated protein. Ichthyosis is inherited as an autosomal recessive trait and requires two mutant alleles for an individual to be affected. References: Kiener, S., Wiener, D. J., Hopke, K., Diesel, A. B., Jagannathan, V., Mauldin, E. A., Casal, M. L., & Leeb, T. (2022). ABHD5 frameshift deletion in Golden Retrievers with ichthyosis. G3 (Bethesda, Md.), 12(2), jkab397.		

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The DNA profile is based on a preliminary marker panel that is subject to modification pending additional genetic information.

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