

## Ichthyosis (Golden Retriever Type 1)

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|-----------------|-------------------------------------------------------------------------------------------|--------------|-------------------------------------------------------------------------------------|
| Client Name:    | Lauriaan Morton (LAU002)                                                                  | Report No:   | ZO2025/9673/20250804/#101092                                                        |
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| Profile:        | DG2025/93667                                                                              | Species:     | Canis lupus familiaris / Canine / Dog                                               |
| Name:           | Golden Starseed Blue (Mufasa)                                                             | Microchip #: | 972274200143122                                                                     |
| Breed:          | Golden Retriever                                                                          |              |                                                                                     |
| Test:           | [PNPLA1] Ichthyosis (Golden Retriever Type 1)                                             |              |                                                                                     |
| Results:        | c.1451_1453delTACTACTA                                                                    | WT/WT        | Clear                                                                               |

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| Sample Type: Whole Blood (EDTA)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | Extraction Method: DNA Extraction: D4069 | Test Type: Genetic Health |
| [PNPLA1] Ichthyosis (Golden Retriever Type 1)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     |                                          |                           |
| <p>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.</p> <p>This test detects the presence or absence of the indelACC/TACTACTA mutation in the PNPLA1 gene associated with ichthyosis Type 1 of Golden Retrievers.</p> <p>Ichthyosis is inherited as an autosomal recessive trait and requires two mutant alleles for an individual to be affected.</p> <p>References: Grall et al 2012. PNPLA1 mutations cause autosomal recessive congenital ichthyosis in golden retriever dogs and humans. Nature Genetics 44, pp140.</p> |                                          |                           |

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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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