

## Ichthyosis

|                 |  |              |   |
|-----------------|--|--------------|---|
| Client Name:    | Liane van der Hoven (LIA001)   | Report No:   | ZO2021/2813/20210814/#12894   |
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| Profile:        | DG2021/20150   | Species:     | Canis lupus familiaris / Canine / Dog   |
| Name:           | Bonyl Kilted Ochre   | Microchip #: | 941000026460135   |
| Breed:          | Golden Retriever   |              |   |
| Test:           | [PNPLA1] Ichthyosis  |              |   |
| Results:        | c.1451_1453delTACTACTA   | WT/WT        | CLEAR   |

|  |  |                           |
|--|--|---------------------------|
| Sample Type: Whole Blood (EDTA)  | Extraction Method: DNA Extraction: D4069 | Test Type: Genetic Health |
| [PNPLA1] Ichthyosis  |  |                           |
| <p>Ichthyosis (PNPLA1) is mild, moderate, or severe scaling of the skin. This is visible in puppies and lasts throughout the lifespan of the canine. The scales darken in colour with age.</p> <p>This test detects the presence or absence of the indelACC/TATACTA mutation in the PNPLA1 gene associated with ichthyosis of Golden Retrievers.</p> <p>Ichthyosis is inherited as an autosomal recessive trait. Two mutant alleles are required for an individual to be affected by ichthyosis.</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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