

## ICHTHYOSIS (PNPLA1)

REPORT NO.: ZO2020/790/0404/04

Client Name: IRENE KELLOCK  
 Kennel Name:  
 Client Address: 21 ELIZABETH DRIVE  
 FOREST HILLS  
 3610  
 Client Tel No.: 083 436 9057



Canine Name: MOCHAVULIN MKUZE  
 Breed: GOLDEN RETRIEVER  
 Microchip No.: 900250000234015  
 Genetic Test: ICHTHYOSIS (PNPLA1)  
 Result: CLEAR  
 Registration No.: ZA010702B16

### ICHTHYOSIS (PNPLA1)

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.

This test detects the indelACC/TATACTA in the PNPLA1 gene associated with ichthyosis in Golden Retrievers. Ichthyosis is inherited as an autosomal recessive trait. Two mutant alleles are required for an individual to be affected by ichthyosis.

Grall et al 2012. PNPLA1 mutations cause autosomal recessive congenital ichthyosis in golden retriever dogs and humans. Nature Genetics 44, pp140.

SAMPLE TYPE: EDTA BLOOD AMPULE  
 EXTRACTION METHOD: DNA EXTRACTION  
 TEST TYPE: SANGER SEQUENCE DETECTION

### BREEDING IMPLICATIONS

		MATERNAL CANDIDATE						
		CLEAR		CARRIER		AFFECTED		
		N	N	N	Del-Ins	Del-Ins	Del-Ins	
PATERNAL CANDIDATE	CLEAR	N	N	NN	NN	NN	NDel-Ins	NDel-Ins
	CARRIER	N	Del-Ins	50% CLEAR 50% CARRIER	50% CLEAR 50% CARRIER	50% CLEAR 50% CARRIER	25% CLEAR 50% CARRIER 25% AFFECTED	50% CARRIER 50% AFFECTED
	AFFECTED	Del-Ins	Del-Ins	ALL CARRIER	50% CARRIER 50% AFFECTED	50% CARRIER 50% AFFECTED	ALL AFFECTED	ALL AFFECTED

Disclaimer: This report does not disregard the existence of any unknown or rare variant gene that may cause Ichthyosis in the Golden Retriever breed.



It is the sender's responsibility to ensure the correctness of the information accompanying the samples. In no event shall Inqaba Biotechnical Industries (Pty) Ltd or its divisions be held liable for indirect, substantial or secondary damages of any kind. Results are usually made available within 7-14 days of receipt of samples. Please note that results are only released subject to payment.