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## **Laboratory Report**

**Laboratory #:** 29342 **Call Name:** Winnie

Order #: 11763 Registered Name: Whiskey Sanitacteam Day

Ordered By: Nancy Chadwick Breed: Golden Retriever

 Ordered:
 May 27, 2016
 Sex:
 Female

 Received:
 June 27, 2016
 DOB:
 July 2014

 Reported:
 July 8, 2016
 Registration #:
 SR88917501

**Microchip #:** 052106829

## **Results:**

Disease	Gene	Genotype	Interpretation
Degenerative Myelopathy	SOD1	WT/WT	Normal (clear)
Ichthyosis (Golden Retriever Type)	PNPLA1	M/M	At-Risk/Affected
Progressive Retinal Atrophy, Golden Retriever 1	SLC4A3	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Golden Retriever 2	TTC8	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration	PRCD	WT/WT	Normal (clear)
Sensory Ataxic Neuropathy	tRNA-Tyr	WT/WT	Normal (clear)

WT, wild type (normal); M, mutant

## Interpretation:

Molecular genetic analysis was performed for six specific mutations reported to be associated with disease in dogs. We identified two normal copies of the DNA sequences in five of the mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these five mutations. However, we identified two mutant copies of the DNA sequences for *PNPLA1*. Thus, this dog is at risk for/affected with Ichthyosis (Golden Retriever Type).

## **Recommendations:**

Ichthyosis (Golden Retriever Type) is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in both copies of the *PNPLA1* gene, this dog is at risk for/affected with this disease. Though there can be variation in the severity of the disease, affected dogs usually present between three weeks and one year of age with generalized scaling of the skin. Breeding of this dog is not recommended if you wish to eliminate this mutation from your lines because 100% of the offspring from a breeding between a *PNPLA1* at-risk/affected dog (M/M) and a *PNPLA1* normal dog (WT/WT) will be carriers (WT/M) of the mutation for Ichthyosis (Golden Retriever Type) and approximately half of the offspring from a breeding between a *PNPLA1* at-risk/affected dog (M/M) and a *PNPLA1* carrier dog (WT/M) will be at risk for/affected with Ichthyosis (Golden Retriever Type). Dogs related to this dog have an increased risk to be affected by or carry the mutated gene. Additional testing for this mutation is indicated for related dogs.

Paw Print Genetics™ has genetic counseling available to you at no additional charge to answer any questions about these test results, their implications and potential outcomes in breeding this dog.

Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration was performed under an exclusive sublicense from OptiGen®, LLC

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Christina J Ramirez, PhD, DVM, DACVP Medical Director Casey R Carl, DVM
Associate Medical Director

Normal results do not exclude inherited mutations not tested in these or other genes that may cause medical problems or may be passed on to offspring. These tests were developed and their performance determined by Paw Print Genetics™. This laboratory has established and verified the tests' accuracy and precision. Because all tests performed are DNA-based, rare genomic variations may interfere with the performance of some tests producing false results. If you think these results are in error, please contact the laboratory immediately for further evaluation. In the event of a valid dispute of results claim, Paw Print Genetics will do its best to resolve such a claim to the customer's satisfaction. If no resolution is possible after investigation by Paw Print Genetics with the cooperation of the customer, the extent of the customer's sole remedy is a refund of the fee paid. In no event shall Paw Print Genetics be liable for indirect, consequential or incidental damages of any kind. Any claim must be asserted within 60 days of the report of the test results.