

Laboratory Report

Laboratory #:	80797	Call Name:	Willow
Order #:	35919	Registered Name:	Thevenet Rebelde Sin Causa
Ordered By:	Ashley Flowers-Fade	Breed:	Golden Retriever
Ordered:	Feb. 14, 2018	Sex:	Male
Received:	Feb. 26, 2018	DOB:	April 2010
Reported:	March 12, 2018	Registration #:	SR79480701
		Microchip #:	985121013102452

Results:

Disease	Gene	Genotype	Interpretation
Degenerative Myelopathy	<i>SOD1</i>	WT/WT	Normal (clear)
Ichthyosis (Golden Retriever Type)	<i>PNPLA1</i>	M/M	At-Risk/Affected
Progressive Retinal Atrophy, Golden Retriever 1	<i>SLC4A3</i>	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Golden Retriever 2	<i>TTC8</i>	WT/M	Carrier
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration	<i>PRCD</i>	WT/WT	Normal (clear)

WT, wild type (normal); M, mutant; Y, Y chromosome (male)

Interpretation:

Molecular genetic analysis was performed for five specific mutations reported to be associated with disease in dogs. We identified two normal copies of the DNA sequences in three of the mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these three 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). In addition, we identified one normal copy and one mutant copy of the DNA sequences for *TTC8*. Thus, this dog is a carrier of Progressive Retinal Atrophy, Golden Retriever 2.

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.

Progressive Retinal Atrophy, Golden Retriever 2 is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *TTC8* gene, this dog is a carrier of this disease. Dogs that carry only one copy of this mutation will not be clinically affected. 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.



Blake C Ballif, PhD
Laboratory & Scientific 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.