

## Laboratory Report

<b>Laboratory #:</b>	326988	<b>Call Name:</b>	Stevie
<b>Order #:</b>	147036	<b>Registered Name:</b>	Regen's Little Black Dress
<b>Ordered By:</b>	Denise Misaras	<b>Breed:</b>	Labrador Retriever
<b>Ordered:</b>	Sept. 16, 2022	<b>Sex:</b>	Female
<b>Received:</b>	Oct. 3, 2022	<b>DOB:</b>	March 2022
<b>Reported:</b>	Oct. 11, 2022	<b>Registration #:</b>	SS33010601
		<b>Microchip #:</b>	956000010662444

### Results:

Disease	Gene	Genotype	Interpretation
Centronuclear Myopathy	<i>PTPLA</i>	WT/WT	Normal (clear)
Cone Degeneration (Labrador Retriever Type)	<i>CNGA3</i>	WT/WT	Normal (clear)
Congenital Myasthenic Syndrome (Labrador Retriever Type)	<i>COLQ</i>	WT/WT	Normal (clear)
Copper Toxicosis (Labrador Retriever Type) ATP7A	<i>ATP7A</i>	WT/WT	Normal/Clear Female
Copper Toxicosis (Labrador Retriever Type) ATP7B	<i>ATP7B</i>	WT/WT	Normal (clear)
Cystinuria (Labrador Retriever Type)	<i>SLC3A1</i>	WT/WT	Normal (clear)
Degenerative Myelopathy	<i>SOD1</i>	WT/WT	Normal (clear)
Elliptocytosis	<i>SPTB</i>	WT/WT	Normal (clear)
Exercise-Induced Collapse	<i>DNM1</i>	WT/M	Carrier
Hereditary Nasal Parakeratosis (Labrador Retriever Type)	<i>SUV39H2</i>	WT/WT	Normal (clear)
Hyperuricosuria	<i>SLC2A9</i>	WT/WT	Normal (clear)
Ichthyosis (Golden Retriever Type 1)	<i>PNPLA1</i>	WT/WT	Normal (clear)
Macular Corneal Dystrophy (Labrador Retriever Type)	<i>CHST6</i>	WT/WT	Normal (clear)
Myotubular Myopathy 1	<i>MTM1</i>	WT/WT	Normal/Clear Female
Narcolepsy (Labrador Retriever Type)	<i>HCRTR2</i>	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Cone-Rod Dystrophy 4	<i>RPGRIP1</i>	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Golden Retriever 2	<i>TTC8</i>	WT/WT	Normal (clear)
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration	<i>PRCD</i>	WT/WT	Normal (clear)
Pyruvate Kinase Deficiency (Labrador Retriever Type)	<i>PKLR</i>	WT/WT	Normal (clear)
Retinal Dysplasia/Oculoskeletal Dysplasia 1	<i>COL9A3</i>	WT/WT	Normal (clear)
Skeletal Dysplasia 2	<i>COL11A2</i>	WT/WT	Normal (clear)
Stargardt Disease	<i>ABCA4</i>	WT/M	Carrier

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

### Interpretation:

Molecular genetic analysis was performed for 22 specific mutations reported to be associated with disease in dogs (21 deleterious mutations and one protective mutation). We identified two normal copies of the DNA sequences in 20 of the deleterious mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 20 mutations. In addition, we identified two normal copies of the DNA sequences in the *ATP7A* gene tested. Thus, this dog does not carry the protective mutation for Copper Toxicosis (Labrador Retriever Type) *ATP7A*. However, we identified one normal copy and one mutant copy of the DNA sequences for *DNM1*. Thus, this dog is a carrier of Exercise-Induced Collapse. In addition, we identified one normal copy and one mutant copy of the DNA sequences for *ABCA4*. Thus, this dog is a carrier of Stargardt Disease.

## Recommendations:

Exercise-Induced Collapse is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *DNM1* gene, this dog is a carrier of this disease. Although dogs that carry only one copy of this mutation will not be clinically affected, if bred with another carrier, the pairing could produce affected offspring. To avoid producing affected offspring, this dog should be bred with dogs that are normal (WT/WT) for this gene. 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.

Stargardt Disease is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *ABCA4* gene, this dog is a carrier of this disease. Although dogs that carry only one copy of this mutation will not be clinically affected, if bred with another carrier, the pairing could produce affected offspring. To avoid producing affected offspring, this dog should be bred with dogs that are normal (WT/WT) for this gene. 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



**Christina J Ramirez, PhD, DVM, DACVP**  
Medical Director

Paw Print Genetics® performed the tests listed on this dog. The genes/diseases reported here were selected by the client. 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. The results included in this report relate only to the items tested using the sample provided. These tests were developed and their performance determined by Paw Print Genetics. This laboratory has established and verified the test(s)' accuracy and precision with >99.9% sensitivity and specificity. The presence of mosaicism may not be detected by this test. Non-paternity may lead to unexpected results. This is not a breed identification test. Because all tests performed are DNA-based, rare genomic variations may interfere with the performance of some tests producing false results. If you think any 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.