

## Laboratory Report

<b>Laboratory #:</b>	397734	<b>Call Name:</b>	GASTON
<b>Order #:</b>	182162	<b>Registered Name:</b>	DEVANLEY'S WHAT A GUY YOU ARE
<b>Ordered By:</b>	Judy Segedi	<b>Breed:</b>	Labrador Retriever
<b>Ordered:</b>	April 12, 2023	<b>Sex:</b>	Male
<b>Received:</b>	May 5, 2023	<b>DOB:</b>	March 2022
<b>Reported:</b>	May 16, 2023	<b>Registration #:</b>	SS32326202
		<b>Microchip #:</b>	956000013857432

### Results:

Disease	Gene	Genotype	Interpretation
Centronuclear Myopathy	<i>PTPLA</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/Y	Normal/Clear Male
Copper Toxicosis (Labrador Retriever Type) ATP7B	<i>ATP7B</i>	WT/M	At-Risk
Exercise-Induced Collapse	<i>DNM1</i>	WT/WT	Normal (Clear)
Hereditary Nasal Parakeratosis (Labrador Retriever Type)	<i>SUV39H2</i>	WT/WT	Normal (Clear)
Macular Corneal Dystrophy (Labrador Retriever Type)	<i>CHST6</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)
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/WT	Normal (Clear)

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

### Interpretation:

Molecular genetic analysis was performed for 12 specific mutations reported to be associated with disease in dogs (11 deleterious mutations and one protective mutation). We identified two normal copies of the DNA sequences in 10 of the deleterious mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 10 mutations. In addition, we identified one normal copy of the DNA sequences for *ATP7A* on the X chromosome. 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 *ATP7B*. Thus, this dog is at risk for Copper Toxicosis (Labrador Retriever Type) ATP7B.

### Recommendations:

Copper Toxicosis (Labrador Retriever Type) is inherited in an autosomal incomplete dominant fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *ATP7B* gene, this dog is at risk for this disease. Though Copper Toxicosis is more commonly seen in dogs having two copies of the mutated gene, dogs inheriting a single copy of the mutation also have an increased, though lesser, risk of developing Copper

Toxicosis. In addition, this disease appears to be sex-influenced in that female dogs inheriting one or two copies of the *ATP7B* mutation are at an increased risk of developing clinical disease compared to their male counterparts. Dogs with Copper Toxicosis have a decreased ability to excrete dietary copper from the body resulting in excessive copper storage in tissues and organs, including the liver, which can result in liver damage and subsequent cirrhosis. Though the age of onset and progression of the disease are variable, most affected dogs will present during middle age with non-specific signs of liver dysfunction including weight loss, lethargy, weakness, vomiting, diarrhea, and abdominal pain. In late stages of disease, affected dogs may develop signs of liver failure which include abdominal swelling, jaundice, and neurological dysfunction. Dogs found to have one or two copies of the mutation may benefit from certain preventative therapies. When a dog with a single copy of the *ATP7B* mutation (WT/M) is bred with another dog with a single copy of the same mutation (WT/M), there is risk of having affected pups. For each pup born to this pairing, there is a 25% chance the puppy will inherit two copies of the mutation (M/M) and a 50% chance the puppy will inherit one copy of the mutation (WT/M) and, in either case, may be susceptible to developing Copper Toxicosis. Dogs related to this dog have an increased risk to be affected by 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.