

## Laboratory Report

<b>Laboratory #:</b>	470357	<b>Call Name:</b>	Bones
<b>Order #:</b>	212622	<b>Registered Name:</b>	Elkens Digging Up Bones
<b>Ordered By:</b>	Leslie Black	<b>Breed:</b>	Labrador Retriever
<b>Ordered:</b>	Oct. 21, 2024	<b>Sex:</b>	Male
<b>Received:</b>	Nov. 7, 2024	<b>DOB:</b>	June 2021
<b>Reported:</b>	Dec. 6, 2024	<b>Registration #:</b>	SS27408201
		<b>Microchip #:</b>	956000013765500

### Results:

Disease	Gene	Genotype	Interpretation
Centronuclear Myopathy	<i>PTPLA</i>	WT/WT	Normal (Clear)
Chondrodystrophy with Intervertebral Disc Disease Risk Factor (CDDY with IVDD)	<i>CFA12 FGF4</i>	WT/WT	Normal (Clear) - No CDDY or Increased IVDD Risk
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>	M/Y	X-Linked Male Carrier
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 (Common Variant)	<i>SOD1</i>	WT/WT	Normal (Clear)
Ehlers-Danlos Syndrome (Labrador Retriever Type), Variant 1	<i>COL5A1</i>	WT/WT	Normal (Clear)
Ehlers-Danlos Syndrome (Labrador Retriever Type), Variant 2	<i>COL5A1</i>	WT/WT	Normal (Clear)
Elliptocytosis	<i>SPTB</i>	WT/WT	Normal (Clear)
Exercise-Induced Collapse	<i>DNM1</i>	WT/WT	Normal (Clear)
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)
Laryngeal Paralysis and Polyneuropathy (Leonberger Type 3)	<i>CNTNAP1</i>	WT/WT	Normal (Clear)
Macular Corneal Dystrophy (Labrador Retriever Type)	<i>CHST6</i>	WT/WT	Normal (Clear)
Myotonia Congenita (Labrador Retriever Type)	<i>CLCN1</i>	WT/WT	Normal (Clear)
Myotubular Myopathy 1 (Labrador Retriever Type)	<i>MTM1</i>	WT/Y	Normal/Clear Male
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/WT	Normal (Clear)
Ullrich Congenital Muscular Dystrophy (Labrador Retriever Type 1)	<i>COL6A3</i>	WT/WT	Normal (Clear)
Ullrich Congenital Muscular Dystrophy (Labrador Retriever Type 2)	<i>COL6A3</i>	WT/WT	Normal (Clear)

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

## Interpretation:

Molecular genetic analysis was performed for 29 specific mutations reported to be associated with disease in dogs. We identified two normal copies of the DNA sequences in 28 mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 28 mutations.

In addition, we identified one mutant copy of the DNA sequence for *ATP7A* on the X chromosome. Thus, this dog carries one copy of the protective mutation for Copper Toxicosis (Labrador Retriever Type) *ATP7A*.

## Recommendations:

This dog was tested for a genetic mutation of the canine *ATP7A* gene which partially protects against copper toxicosis in dogs that have inherited the *ATP7B* mutation described above. This dog carries one copy of the *ATP7A* gene mutation. Therefore, this dog may have a lesser risk of copper toxicosis than the risk associated with the inheritance of the *ATP7B* gene mutation alone. The *ATP7A* gene mutation is more effective at decreasing the risk of copper toxicosis in male dogs than females. However, since multiple factors (both genetic and environmental) play a role in causing copper toxicosis, the *ATP7A* mutation is not completely protective in either sex. Dogs that did not inherit the *ATP7B* gene mutation are not affected positively or negatively when they inherit one or two copies of the *ATP7A* gene mutation. Note: The *ATP7A* mutation is located on the X chromosome. Since males only have a single X chromosome they can only inherit a single copy of this mutation.

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.

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.