

Laboratory Report

Laboratory #:	291849	Call Name:	Mercy
Order #:	132433	Registered Name:	Empress I Can only Imagine
Ordered By:	Dawn Nacey	Breed:	Labrador Retriever
Ordered:	Feb. 17, 2022	Sex:	Female
Received:	March 1, 2022	DOB:	Jan. 2020
Reported:	March 15, 2022	Registration #:	SS168221/01
		Microchip #:	992000000184244

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/M	Carrier Female
Copper Toxicosis (Labrador Retriever Type) ATP7B	<i>ATP7B</i>	WT/WT	Normal (clear)
Exercise-Induced Collapse	<i>DNM1</i>	WT/WT	Normal (clear)
Hereditary Nasal Parakeratosis	<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/M	Carrier

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. However, we identified one normal copy and one mutant copy of the DNA sequences for *ABCA4*. Thus, this dog is a carrier of Stargardt Disease. In addition, we identified one normal copy and one mutant copy of the DNA sequences for *ATP7A*. Thus, this dog carries one copy of the protective mutation for Copper Toxicosis (Labrador Retriever Type) ATP7A.

Recommendations:

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.

This dog was also 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. 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. 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.



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.

Coat Color and Trait Certificate

Call Name:	Mercy	Laboratory #:	291849
Registered Name:	Empress I Can only Imagine	Registration #:	SS168221/01
Breed:	Labrador Retriever	Microchip #:	992000000184244
Sex:	Female	Certificate Date:	March 15, 2022
DOB:	Jan. 2020		

This canine's DNA showed the following genotype(s):

Coat Color/Trait Test	Gene	Genotype	Interpretation
D Locus (Dilute)	<i>MLPH</i>	D/D	Non-dilute (does not carry dilute)
E Locus - e (Apricot/Cream/Red/Yellow, Common Variant Found in Many Breeds)	<i>MC1R</i>	E/e	Black (carries yellow/red)
L Locus (Long Hair/Fluffy) - Lh ¹ (Common Variant Found in Many Breeds)	<i>FGF5</i>	Sh/Sh	Shorthaired (does not carry long hair)

Interpretation:

This dog does not carry any copies of the d¹ or d² mutations and has a D locus genotype of **D/D** which does not result in the "dilution" or lightening of the pigments that produce the dog's coat color. This dog will pass one copy of **D** to 100% of its offspring and cannot produce d/d dogs.

This dog carries one copy of **E** and one copy of **e** which allows for the production of black pigment. However, this dog's coat color is also dependent on the K, A, and B genes. This dog will pass **E** on to 50% of its offspring and **e** to 50% of its offspring, which can produce a yellow/red coat (including shades of white, cream, yellow, apricot or red) if inherited with another copy of **e**.

This dog carries two copies of **Sh** which results in short hair. This dog will pass on **Sh** to 100% of its offspring.

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