

Laboratory Report

Laboratory #: 296930
Order #: 134721
Ordered By: Denise Misaras
Ordered: March 15, 2022
Received: April 15, 2022
Reported: April 22, 2022

Call Name: Fringe
Registered Name: Regen's On The Edge
Breed: Labrador Retriever
Sex: Female
DOB: Dec. 2021
Registration #: SS31479101
Microchip #: 956000012754803

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/M	Carrier 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/WT	Normal (clear)
Hereditary Nasal Parakeratosis	<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>HCRT2</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)

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 for the 21 deleterious mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 21 mutations. In addition, we identified 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).

Recommendations:

No deleterious mutations were identified. Thus, this dog is not at an increased risk for the diseases caused by or associated with the mutations tested. Because this dog is "clear" of these mutations, this dog will only pass the normal genes on to its offspring. 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.

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 and dogs that inherit two copies of the *ATP7A* mutation will have an even lesser risk of copper toxicosis than those inheriting just a single copy. 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



Robert D. Westra, MS, DVM
Associate 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.