

Laboratory Report

Laboratory #: 264376
Order #: 120394
Ordered By: Denise Misaras
Ordered: Oct. 14, 2021
Received: Nov. 5, 2021
Reported: Nov. 16, 2021

Call Name: Dava
Registered Name: Regen's Black Magic
Breed: Labrador Retriever
Sex: Female
DOB: Oct. 2018
Registration #: SS08524701
Microchip #: 956000005099517

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/M	Carrier
Hereditary Nasal Parakeratosis	<i>SUV39H2</i>	WT/WT	Normal (clear)
Hyperuricosuria	<i>SLC2A9</i>	WT/WT	Normal (clear)
Ichthyosis (Golden Retriever Type)	<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 20 of the deleterious mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 20 mutations. 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 *ATP7A*. Thus, this dog carries one copy of the protective mutation for Copper Toxicosis (Labrador Retriever Type).

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.

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



Casey R Carl, DVM
Associate Medical Director

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. These tests were developed and their performance determined by Paw Print Genetics®. This laboratory has established and verified the tests' accuracy and precision. Because all tests performed are DNA-based, rare genomic variations may interfere with the performance of some tests producing false results. If you think these 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.