Paw Print Genetics
A division of Genetic Veterinary Sciences, Inc.

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Laboratory Report

**** Amended Report ****

Laboratory #:	462985	**Call Name:**	Sadie
Order #:	208790	**Registered Name:**	Breezy's God Bless The Girls
Ordered By:	Bree Moderow	**Breed:**	Labrador Retriever
Ordered:	Aug. 15, 2024	**Sex:**	Female
Received:	Aug. 26, 2024	**DOB:**	July 2024
Reported:	Sept. 13, 2024	**Registration #:**	SS48712202
Amended:	Sept. 13, 2024	**Microchip #:**	900235000066411

Results:

Disease	Gene	Genotype	Interpretation
Centronuclear Myopathy	*PTPLA*	WT/WT	Normal (Clear)
Chondrodystrophy with Intervertebral Disc Disease Risk Factor (CDDY with IVDD)	*CFA12 FGF4*	WT/WT	Normal (Clear) - No CDDY or Increased IVDD Risk
Cone Degeneration (Labrador Retriever Type)	*CNGA3*	WT/WT	Normal (Clear)
Congenital Myasthenic Syndrome (Labrador Retriever Type)	*COLQ*	WT/WT	Normal (Clear)
Copper Toxicosis (Labrador Retriever Type) ATP7A	*ATP7A*	WT/WT	Normal/Clear Female
Copper Toxicosis (Labrador Retriever Type) ATP7B	*ATP7B*	M/M	At-Risk/Affected
Cystinuria (Labrador Retriever Type)	*SLC3A1*	WT/WT	Normal (Clear)
Degenerative Myelopathy (Common Variant)	*SOD1*	WT/WT	Normal (Clear)
Ehlers-Danlos Syndrome (Labrador Retriever Type), Variant 1	*COL5A1*	WT/WT	Normal (Clear)
Ehlers-Danlos Syndrome (Labrador Retriever Type), Variant 2	*COL5A1*	WT/WT	Normal (Clear)
Elliptocytosis	*SPTB*	WT/WT	Normal (Clear)
Exercise-Induced Collapse	*DNM1*	WT/M	Carrier
Hereditary Nasal Parakeratosis (Labrador Retriever Type)	*SUV39H2*	WT/WT	Normal (Clear)
Hyperuricosuria	*SLC2A9*	WT/WT	Normal (Clear)
Ichthyosis (Golden Retriever Type 1)	*PNPLA1*	WT/WT	Normal (Clear)
Laryngeal Paralysis and Polyneuropathy (Leonberger Type 3)	*CNTNAP1*	WT/WT	Normal (Clear)
Macular Corneal Dystrophy (Labrador Retriever Type)	*CHST6*	WT/WT	Normal (Clear)
Myotonia Congenita (Labrador Retriever Type)	*CLCN1*	WT/WT	Normal (Clear)
Myotubular Myopathy 1 (Labrador Retriever Type)	*MTM1*	WT/WT	Normal/Clear Female
Narcolepsy (Labrador Retriever Type)	*HCRTR2*	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Cone-Rod Dystrophy 4	*RPGRIP1*	WT/WT	Normal (Clear)

Progressive Retinal Atrophy, Golden Retriever 2	*TTC8*	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration	*PRCD*	WT/WT	Normal (Clear)
Pyruvate Kinase Deficiency (Labrador Retriever Type)	*PKLR*	WT/WT	Normal (Clear)
Retinal Dysplasia/Oculoskeletal Dysplasia 1	*COL9A3*	WT/WT	Normal (Clear)
Skeletal Dysplasia 2	*COL11A2*	WT/WT	Normal (Clear)
Stargardt Disease	*ABCA4*	WT/WT	Normal (Clear)
Ullrich Congenital Muscular Dystrophy (Labrador Retriever Type 1)	*COL6A3*	WT/WT	Normal (Clear)
Ullrich Congenital Muscular Dystrophy (Labrador Retriever Type 2)	*COL6A3*	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 27 of the mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 27 mutations. However, we identified two mutant copies of the DNA sequences for *ATP7B*. Thus, this dog is at risk for/affected with Copper Toxicosis (Labrador Retriever Type) ATP7B. In addition, 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.

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 both copies of the *ATP7B* gene, this dog is at risk for/affected with 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. Breeding this dog is not recommended if you wish to eliminate this mutation from your lines because 100% of the offspring from a breeding between a dog carrying two copies of the *ATP7B* gene mutation (M/M) and an *ATP7B* normal dog (WT/WT) will inherit one copy (WT/M) of the mutation for Copper Toxicosis (Labrador Retriever Type) and approximately half of the offspring from a breeding between a dog carrying two copies of the *ATP7B* gene mutation (M/M) and a dog carrying one copy of the *ATP7B* gene mutation (WT/M) will inherit two copies of the mutation associated with Copper Toxicosis (Labrador Retriever Type). In either case, all puppies from this dog will be at an increased risk for developing copper toxicosis. Dogs related to this dog have an increased risk of developing Copper Toxicosis (Labrador Retriever Type). Additional testing for this mutation is indicated for related dogs.

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. Dogs that carry only one copy of this mutation will not be clinically affected. 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.

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.

NOTE: The following fields were adjusted at the client's request on Sep 13, 2024: Registered Name, Microchip Id

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.