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

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Laboratory Report

Laboratory #:	475869	**Call Name:**	Sal
Order #:	215523	**Registered Name:**	SGR's Salvatore Mundi
Ordered By:	Colleen Goularte	**Breed:**	Labrador Retriever
Ordered:	Dec. 5, 2024	**Sex:**	Male
Received:	Dec. 20, 2024	**DOB:**	May 2024
Reported:	Jan. 22, 2025	**Registration #:**	
		Microchip #:	

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/Y	Normal/Clear Male
Copper Toxicosis (Labrador Retriever Type) ATP7B	*ATP7B*	WT/M	At-Risk
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/WT	Normal (Clear)
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/Y	Normal/Clear Male
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 28 of the mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 28 mutations. However, we identified one normal copy and one mutant copy of the DNA sequences for *ATP7B*. Thus, this dog is at risk of Copper Toxicosis (Labrador Retriever Type) ATP7B.

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 one copy of the *ATP7B* gene, this dog is at risk for 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. When a dog with a single copy of the *ATP7B* mutation (WT/M) is bred with another dog with a single copy of the same mutation (WT/M), there is risk of having affected pups. For each pup born to this pairing, there is a 25% chance the puppy will inherit two copies of the mutation (M/M) and a 50% chance the puppy will inherit one copy of the mutation (WT/M) and, in either case, may be susceptible to developing Copper Toxicosis. Dogs related to this dog have an increased risk to be affected by 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.

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Coat Color and Trait Certificate

Call Name: Sal
Registered Name: SGR's Salvatore Mundi
Breed: Labrador Retriever
Sex: Male
DOB: May 2024

Laboratory #: 475869
Registration #:
Microchip #:
Certificate Date: Jan. 22, 2025

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

Coat Color/Trait Test	Gene	Genotype	Interpretation
A Locus (Agouti)	*ASIP*	a /a^{tt}	Tricolor, black and tan
A_S Locus (Saddle Tan)	*RALY*	N/N	No saddle tan/creeping tan
B Locus (Brown)	*TYRP1*	b/b	Brown coat, nose and foot pads (carries two copies of brown)
Brachycephaly	*BMP3*	BR/BR	Likely medium to long muzzle
Chondrodysplasia (CDPA)	*CFA18 FGF4*	cd/cd	No Leg Shortening Associated with CDPA
Co Locus (Cocoa, French Bulldog Type)	*HPS3*	CO/CO	Black coat, nose and foot pads (does not carry cocoa)
Cu Locus (Curly Hair)	*KRT71*	Cu/Cu	Straight coat
D Locus (Dilute)	*MLPH*	D/D	Non-dilute (does not carry dilute)
E Locus	*MC1R*	E/E	Black
H Locus (Harlequin, Great Dane Type)	*PSMB7*	h/h	No harlequin
Hairlessness	*SGK3*	Rh/Rh	Coated
Hr Locus (FOXI3 Hairless Gene Test, Mexican Hairless, Peruvian Hairless and Chinese Crested Type)	*FOXI3*	hr/hr	Coated
I Locus (Intensity)	*MFSD12*	I/I	Normal intensity
IC Locus (Improper Coat/Furnishings)	*RSPO2*	IC/IC	No furnishings, improper coat
K Locus (Dominant Black)	*CBD103*	K^B/K^B	No agouti expression allowed
L Locus (Long Hair/Fluffy)	*FGF5*	Sh/Sh	Shorthaired (does not carry long hair)
M Locus (Merle)	*PMEL*	m/m	Non merle
Polydactyly (Common Variant)	*LMBR1*	pd/pd	Normal (typical) toes (likely no hind dewclaws)
Polydactyly (Great Pyrenees Type)	*ALX4*	pd/pd	Normal (typical) toes (likely no double hind dewclaws)
R Locus (Roan/Ticked)	*USH2A*	r/r	No roan or ticking
S Locus (White Spotting, Parti, or Piebald)	*MITF*	S/S	No white spotting, flash, parti, or piebald
SD Locus (Shedding)	*MC5R*	sd/SD	Moderate shedding

ST Locus (Screw Tail, Bulldog and Terrier Type)	*DVL2*	N/N	No kinked or screw tail
T Locus (Natural Bobtail)	*T*	t/t	Normal tail

Interpretation:

This dog carries two copies of **a^t** which results in tan points and can also present as a black and tan or tricolor coat color. However, this dog's coat color is also dependent on the E, K, and B genes. The tan point coat color is only expressed if the dog is also E/E or E/e at the E locus and k^y/k^y at the K locus. This dog will pass on **a^t** to 100% of its offspring.

This dog carries two copies of the **N** allele, which is not associated with a saddle tan coat color. This dog's coat color is also dependent on the E, A, and K genes, among others. This dog will pass **N** to 100% of its offspring.

This dog carries two copies of one of the same b mutation and has a B locus genotype of b/b. Thus, this dog typically will have a brown coat, nose and foot pads. Depending on the breed, b/b dogs may be referred to as brown, chocolate, liver or red. However, this dog's coat color is dependent on the genotypes of many other genes. This dog will pass one copy of b to 100% of its offspring. This dog can produce b/b offspring if bred to a dog that is also a carrier of a b mutation (B/b or b/b).

This dog carries two copies of the BR Allele which is found in dogs with medium to long muzzles. However, the actual muzzle length of the dog is a result of a combination of factors including multiple variants in other genes. This dog will pass one copy of BR to 100% of its offspring and can produce dogs with medium to long muzzles.

Two genetic mutations are associated with shortened legs in dogs. Both mutations consist of copied sections (duplication) of the canine *FGF4* gene (called an *FGF4*-retrogene) that have been inserted into two aberrant locations in the genome; one in chromosome 12 (*CFA12 FGF4*; associated with CDDY and IVDD risk) and one in chromosome 18 (*CFA18 FGF4*; associated with chondrodysplasia [CDPA], but not associated with IVDD). Appropriate breeding decisions regarding dogs which have inherited the *CFA12 FGF4* mutation (WT/M or M/M) need to address both the potential loss of genetic diversity in a population which would occur if dogs with this mutation were prohibited from breeding as well as the loss of the short-legged appearance that is a defining physical characteristic for some breeds. In breeds which inherit both mutations, breeders may use genetic testing results to selectively breed for the CDPA (*CFA18 FGF4*) mutation while breeding away from the CDDY and IVDD risk (*CFA12 FGF4*) mutation to reduce IVDD risk and retain the short-legged appearance. However, the frequency of each mutation varies between breeds and, in some cases, may not be conducive to such a breeding strategy. For example, breeds with extreme limb shortening (e.g. Basset hound, Dachshund, Corgi) typically develop their appearance due to inheritance of both the *CFA12 FGF4* and *CFA18 FGF4* mutations. In addition, depending on the breed, offspring born without either the *CFA12 FGF4* or *CFA18 FGF4* mutations may display longer limbs than cohorts and, therefore, not meet specific breed standards.

This dog carries two copies of the **cd** allele which does not result in leg shortening. However, the actual leg length of the dog is a result of a combination of factors including the mutation associated with CDDY and IVDD risk (*CFA12 FGF4*) as well as variants in other genes. This dog will pass one copy of **cd** to 100% of its offspring.

This dog does not carry any copies of the co (cocoa) mutation and has a Co Locus genotype of **CO/CO**. Thus, this dog typically will have a black coat, nose, and foot pads. However, this dog's coat color is dependent on the genotypes of many other genes including the B Locus (Brown). This dog will pass one copy of **CO** to 100% of its offspring and cannot produce co/co (cocoa) dogs.

This dog carries two copies of **Cu** which results in a straight coat. However, the overall coat type of this dog is dependent on the combination of this dog's genotypes at the L, Cu, and IC loci. This dog will pass **Cu** on to 100% of its offspring.

This dog does not carry any copies of the d_1, d_2, or d_3 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 two copies 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 100% of its offspring.