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

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Laboratory Report

Laboratory #:	484431	**Call Name:**	Shimmer
Order #:	220325	**Registered Name:**	Empress Shimmer of Hope
Ordered By:	Dawn Nacey	**Breed:**	Labrador Retriever
Ordered:	Feb. 14, 2025	**Sex:**	Female
Received:	Feb. 28, 2025	**DOB:**	May 2022
Reported:	March 28, 2025	**Registration #:**	SS34238601
		Microchip #:	992000000963529

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*	M/M	X-Linked Female Two Copy Carrier
Copper Toxicosis (Labrador Retriever Type) ATP7B	*ATP7B*	WT/WT	Normal (Clear)
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/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 28 mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 28 mutations.

In addition, we identified two mutant copies of the DNA sequences for *ATP7A*. Thus, this dog carries two copies of the protective mutation for Copper Toxicosis (Labrador Retriever Type) ATP7A.

Recommendations:

This dog was 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 two copies of the *ATP7A* gene mutation. Therefore, this dog may have a lesser risk of copper toxicosis than the risk associated with the inheritance of the *ATP7B* gene mutation alone. In addition, 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. 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. Dogs that did not inherit the *ATP7B* gene mutation are not affected positively or negatively when they inherit one or two copies of the *ATP7A* gene mutation. 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.

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Coat Color and Trait Certificate

Call Name:	Shimmer	**Laboratory #:**	484431
Registered Name:	Empress Shimmer of Hope	**Registration #:**	SS34238601
Breed:	Labrador Retriever	**Microchip #:**	992000000963529
Sex:	Female	**Certificate Date:**	March 28, 2025
DOB:	May 2022		

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

Coat Color/Trait Test	Gene	Genotype	Interpretation
A Locus (Agouti)	*ASIP*	a^t/a	Tricolor, black and tan (carries bicolor/solid)
A^S Locus (Saddle Tan)	*RALY*	N/A^S	Saddle tan/creeping tan (non saddle tan carrier)
B Locus (Brown)	*TYRP1*	B/b	Black coat, nose and foot pads (carries one copy 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 one copy of $\mathbf{a^t}$ and one copy of **a** 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 $\mathbf{a^t}$ to 50% of its offspring and **a** to 50% of its offspring.

This dog carries one copy of an $\mathbf{A^s}$ allele and one copy of **N**, the wildtype sequence. This combination of alleles is also found in dogs with a saddle tan coat color. However, this dog's coat color is also dependent on the E, A, and K loci. Saddle tan is found only in dogs that are also E/E or E/e at the E locus, k^y/k^y at the K locus, and a^t/a^t or a^t/a at the A locus. This dog will pass the **N** allele to 50% of its offspring and the $\mathbf{A^s}$ allele to 50% of its offspring, which can produce saddle tan dogs.

This dog carries one copy of one of the b mutations and has a B locus genotype of B/b. 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. This dog will pass one copy of B to 50% of its offspring and one copy of b to 50% 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). Depending on the breed, b/b dogs may be referred to as brown, chocolate, liver or red.

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.

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.

This dog carries two copies of **h** and will not have a harlequin coat color. The dog will pass on **h** to 100% of its offspring.

This dog does not carry any copies of the rh^1 or rh^2 mutations and has a genotype of HR/HR which does not result in hairlessness in the Scottish Deerhound. This dog will pass one copy of HR to 100% of its offspring.

This dog carries two copies of the recessive **hr** allele which results in a coated dog. This dog will pass on the **hr** allele to 100% of its offspring.

This dog does not carry a copy of the i mutation and has an I locus genotype of **I/I** which does not result in the lightening of the light, phaeomelanin pigments that produce the dog's coat color in an e/e dog. This dog will pass one copy of **I** to 100% of its offspring and cannot produce i/i dogs.

This dog carries two copies of **IC** and will therefore have no furnishings (improper 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 **IC** (improper coat) on to 100% of its offspring and can produce puppies with improper coat if bred with a dog that carries one copy (**F/IC**) or two copies (**IC/IC**) of the mutation for improper coat.

The K locus genotype for this dog is $\mathbf{K^B/K^B}$ which prevents expression of the agouti gene (A locus) and allows for solid eumelanin (black pigment) production in pigmented areas of the dog. However, this dog's coat color is also dependent on its genotypes at the E and B loci. This dog will pass on $\mathbf{K^B}$ to 100% of its offspring.

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

This dog carries two copies of **m**, the non-merle, wild-type allele of the *PMEL* gene, and, therefore, does not have a merle coat color/pattern. This dog will pass on one copy of the **m** allele to 100% of its offspring.

This dog carries two copies of the *LMBR1* **pd** allele which is found in dogs with normal, typical toes and likely no hind dewclaws. However, polydactyly can result from variants in other genes. This dog will pass one copy of **pd** to 100% of its offspring.

This dog carries two copies of the *ALX4* **pd** allele which is found in Great Pyrenees dogs without bilateral double dewclaws. However, polydactyly can result from variants in other genes. This dog will pass one copy of **pd** to 100% of its offspring.

This dog does not carry a copy of R or R^{Ti} and has a genotype of r/r which does not result in roan or ticking in the white areas of the coat. This dog will pass one copy of r to 100% of its offspring.

This dog carries two copies of **S** which results in a solid coat with no white spotting, flash, parti, or piebald coat color. This dog will pass on one copy of **S** to 100% of its offspring.

This dog carries one copy of **sd** and one copy of **SD** which has been associated with moderate shedding. However, the overall degree of shedding for this dog is dependent on the combination of this dog's genotypes at the SD and IC loci. This dog will pass **SD** on to 50% of its offspring and **sd** on to 50% of its offspring.

This dog does not carry a copy of ST and has a genotype of N/N which does not result in a screw or kinked tail in the bully breeds. This dog will pass one copy of **N** to 100% of its offspring.

This dog carries two copies of **t** which results in a tail of normal length (no bobtail). This dog will pass on **t** to 100% of its offspring.

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.

Paw Print Genetics® performed the testing on the dog listed on this certificate. The genes/traits reported here were selected by the client. Normal results do not exclude inherited mutations not tested in these or other genes that may cause variation in traits, 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.