

Laboratory Report

Laboratory #:	392998	Call Name:	Sparkle
Order #:	180525	Registered Name:	Empress Sparkle Like A Diamond
Ordered By:	Dawn Nacey	Breed:	Labrador Retriever
Ordered:	March 23, 2023	Sex:	Female
Received:	March 24, 2023	DOB:	April 2021
Reported:	April 5, 2023	Registration #:	SS259202/02
		Microchip #:	992000000877007

Results:

Disease	Gene	Genotype	Interpretation
Centronuclear Myopathy	<i>PTPLA</i>	WT/WT	Normal (clear)
Congenital Myasthenic Syndrome (Labrador Retriever Type)	<i>COLQ</i>	WT/WT	Normal (clear)
Exercise-Induced Collapse	<i>DNM1</i>	WT/M	Carrier
Hereditary Nasal Parakeratosis (Labrador Retriever Type)	<i>SUV39H2</i>	WT/M	Carrier
Macular Corneal Dystrophy (Labrador Retriever Type)	<i>CHST6</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/M	Carrier
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 10 specific mutations reported to be associated with disease in dogs. We identified two normal copies of the DNA sequences in seven of the mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these seven 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 *SUV39H2*. Thus, this dog is a carrier of Hereditary Nasal Parakeratosis (Labrador Retriever Type). In addition, we identified one normal copy and one mutant copy of the DNA sequences for *PRCD*. Thus, this dog is a carrier of Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration.

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.

Hereditary Nasal Parakeratosis (Labrador Retriever Type) is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *SUV39H2* 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.

Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in one copy of the *PRCD* 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.

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.



Helen F Smith, PhD
Associate Laboratory Director



Christina J Ramirez, PhD, DVM, DACVP
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.

Laboratory Report

Laboratory #: 392998	Call Name: Sparkle
Order #: 180589	Registered Name: Empress Sparkle Like A Diamond
Ordered By: Dawn Nacey	Breed: Labrador Retriever
Ordered: March 23, 2023	Sex: Female
Received: March 24, 2023	DOB: April 2021
Reported: April 5, 2023	Registration #: SS259202/02
	Microchip #: 992000000877007

Results:

Disease	Gene	Genotype	Interpretation
Copper Toxicosis (Labrador Retriever Type) ATP7A	<i>ATP7A</i>	M/M	Two Copy Carrier Female
Copper Toxicosis (Labrador Retriever Type) ATP7B	<i>ATP7B</i>	WT/WT	Normal (clear)
Degenerative Myelopathy	<i>SOD1</i>	WT/WT	Normal (clear)

WT, wild type (normal); M, mutant; Y, Y chromosome (male)

Interpretation:

Molecular genetic analysis was performed for three specific mutations reported to be associated with disease in dogs (two deleterious mutations and one protective mutation). We identified two normal copies of the DNA sequences in the two deleterious mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these two mutations. However, we identified two mutant copies of the DNA sequences for *ATP7A*. Thus, this dog is Copper Toxicosis (Labrador Retriever Type) ATP7A. 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:

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. 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 two copies of the *ATP7A* gene mutation. 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. 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.

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Blake C Ballif, PhD
Laboratory & Scientific Director



Christina J Ramirez, PhD, DVM, DACVP
Medical Director

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Coat Color and Trait Certificate

Call Name:	Sparkle	Laboratory #:	392998
Registered Name:	Empress Sparkle Like A Diamond	Registration #:	SS259202/02
Breed:	Labrador Retriever	Microchip #:	992000000877007
Sex:	Female	Certificate Date:	April 5, 2023
DOB:	April 2021		

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

Coat Color/Trait Test	Gene	Genotype	Interpretation
B Locus (Brown)	<i>TYRP1</i>	B/b	Black coat, nose and foot pads (carries one copy of brown)
D Locus (Dilute)	<i>MLPH</i>	D/D	Non-dilute (does not carry dilute)
K Locus (Dominant Black)	<i>CBD103</i>	K ^B /K ^B	No agouti expression allowed

Interpretation:

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 does not carry any copies of the d¹ or d² 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.

The K locus genotype for this dog is **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 **K^B** to 100% of its offspring.

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Blake C Ballif, PhD
Laboratory & Scientific Director



Christina J Ramirez, PhD, DVM, DACVP
Medical Director

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