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

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Laboratory Report

Laboratory #:	291848	**Call Name:**	BB
Order #:	132433	**Registered Name:**	Empress the Bee's Knees
Ordered By:	Dawn Nacey	**Breed:**	Labrador Retriever
Ordered:	Feb. 17, 2022	**Sex:**	Female
Received:	March 1, 2022	**DOB:**	Oct. 2020
Reported:	March 9, 2022	**Registration #:**	SS228323/01
		Microchip #:	992000000412232

Results:

Disease	Gene	Genotype	Interpretation
Centronuclear Myopathy	*PTPLA*	WT/WT	Normal (clear)
Congenital Myasthenic Syndrome (Labrador Retriever Type)	*COLQ*	WT/WT	Normal (clear)
Copper Toxicosis (Labrador Retriever Type) ATP7A	*ATP7A*	M/M	Two Copy Carrier Female
Copper Toxicosis (Labrador Retriever Type) ATP7B	*ATP7B*	WT/WT	Normal (clear)
Exercise-Induced Collapse	*DNM1*	WT/WT	Normal (clear)
Hereditary Nasal Parakeratosis	*SUV39H2*	WT/WT	Normal (clear)
Macular Corneal Dystrophy (Labrador Retriever Type)	*CHST6*	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)
Retinal Dysplasia/Oculoskeletal Dysplasia 1	*COL9A3*	WT/WT	Normal (clear)
Skeletal Dysplasia 2	*COL11A2*	WT/WT	Normal (clear)
Stargardt Disease	*ABCA4*	M/M	At-Risk/Affected

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

Interpretation:

Molecular genetic analysis was performed for 12 specific mutations reported to be associated with disease in dogs (11 deleterious mutations and one protective mutation). We identified two normal copies of the DNA sequences for 10 of the deleterious mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 10 mutations. However, we identified two mutant copies of the DNA sequences for *ABCA4*. Thus, this dog is at risk for/affected with Stargardt Disease. 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).

Recommendations:

Stargardt Disease is inherited in an autosomal recessive fashion. Based on this, and the fact that this dog showed a mutation in both copies of the *ABCA4* gene, this dog is at risk for/affected with this disease. Affected dogs present prior to 10 years of age with signs of vision loss including dilated pupils and decreased response to light. On a veterinary eye exam, affected dogs will have changes in reflectivity and appearance of a structure behind the retina called the tapedum and, may show thinning of the retinal blood vessels. Dogs may not completely lose

their vision during their lifetime, but will develop significant loss of vision, especially in well-lit environments. Breeding of this dog is not recommended if you wish to eliminate this mutation from your lines because 100% of the offspring from a breeding between an *ABCA4* at-risk/affected dog (M/M) and an *ABCA4* normal dog (WT/WT) will be carriers (WT/M) of the mutation for Stargardt Disease and approximately half of the offspring from a breeding between an *ABCA4* at-risk/affected dog (M/M) and an *ABCA4* carrier dog (WT/M) will be at risk for/affected with Stargardt Disease. 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 two copies of the *ATP7A* gene mutation. 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. 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

Coat Color and Trait Certificate

Call Name:	BB	**Laboratory #:**	291848
Registered Name:	Empress the Bee's Knees	**Registration #:**	SS228323/01
Breed:	Labrador Retriever	**Microchip #:**	992000000412232
Sex:	Female	**Certificate Date:**	March 9, 2022
DOB:	Oct. 2020		

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

Coat Color/Trait Test	Gene	Genotype	Interpretation
D Locus (Dilute)	*MLPH*	D/D	Non-dilute (does not carry dilute)
E Locus - e (Apricot/Cream/Red/Yellow, Common Variant Found in Many Breeds)	*MC1R*	E/e	Black (carries yellow/red)
L Locus (Long Hair/Fluffy) - Lh^1 (Common Variant Found in Many Breeds)	*FGF5*	Sh/Sh	Shorthaired (does not carry long hair)

Interpretation:

This dog does not carry any copies of the d^1 or d^2 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 one copy of **E** and one copy 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 50% of its offspring and **e** to 50% of its offspring, which can produce a yellow/red coat (including shades of white, cream, yellow, apricot or red) if inherited with another copy of **e**.

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

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Blake C Ballif, PhD
Laboratory & Scientific Director

Christina J Ramirez, PhD, DVM, DACVP
Medical Director