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

220 E. Rowan, Suite 220
Spokane, Washington 99207
www.pawprintgenetics.com
(509) 483-5950

Laboratory Report

Laboratory #:	142696	**Call Name:**	Vesper
Order #:	63663	**Registered Name:**	-
Ordered By:	Lauren Sikkema	**Breed:**	Australian Cobberdog
Ordered:	July 23, 2019	**Sex:**	Female
Received:	Aug. 15, 2019	**DOB:**	May 2019
Reported:	Aug. 20, 2019	**Registration #:**	-

Results:

Disease	Gene	Genotype	Interpretation
Copper Toxicosis (Labrador Retriever Type) ATP7A	*ATP7A*	M/M	Two Copy Carrier Female
Copper Toxicosis (Labrador Retriever Type) ATP7B	*ATP7B*	WT/M	At-Risk

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

Interpretation:

Molecular genetic analysis was performed for two specific mutations reported to be associated with copper toxicosis in dogs (one deleterious mutation and one protective mutation). We identified one normal copy and one mutant copy of the DNA sequences for *ATP7B*. Thus, this dog is at risk for Copper Toxicosis (Labrador Retriever Type). 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:

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 most 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 speed of disease progression are variable, most affected dogs will present in 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 including abdominal swelling, jaundice, and neurological dysfunction. Dogs found to have one or two copies of the mutation may benefit from certain therapies. When a dog that has inherited a single copy of this mutation is bred with another dog with a single copy of the same mutation, there is risk of having affected pups. For each pup that is born to this pairing, there is a 25% chance that the puppy will inherit two copies of the mutation and a 50% chance that the puppy will inherit one copy of the mutation 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.

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. 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, The *ATP7A* gene mutation is more effective at decreasing the risk of copper toxicosis in male dogs than females and 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. 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.

Christina J Ramirez, PhD, DVM, DACVP
Medical Director

Casey R Carl, DVM
Associate Medical Director