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

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Laboratory Report

Laboratory #:	434785	**Call Name:**	Merle boy 1
Order #:	199108	**Registered Name:**	-
Ordered By:	Mindy Goss	**Breed:**	Australian Cobberdog
Ordered:	Feb. 4, 2024	**Sex:**	Male
Received:	Feb. 5, 2024	**DOB:**	Jan. 2024
Reported:	March 4, 2024	**Registration #:**	-

Results:

Disease	Gene	Genotype	Interpretation
Copper Toxicosis (Labrador Retriever Type) ATP7A	*ATP7A*	WT/Y	Normal/Clear Male
Copper Toxicosis (Labrador Retriever Type) ATP7B	*ATP7B*	WT/M	At-Risk
Degenerative Myelopathy (Common Variant)	*SOD1*	WT/WT	Normal (Clear)
Episodic Falling Syndrome	*BCAN*	WT/WT	Normal (Clear)
Exercise-Induced Collapse	*DNM1*	WT/M	Carrier
Familial Nephropathy (Cocker Spaniel Type)	*COL4A4*	WT/WT	Normal (Clear)
Glycogen Storage Disease VII, PFK Deficiency	*PFKM*	WT/WT	Normal (Clear)
Microphthalmia (Soft Coated Wheaten Terrier Type)	*RBP4*	WT/WT	Normal (Clear)
Muscular Dystrophy (Golden Retriever Type)	*DMD*	WT/Y	Normal/Clear Male
Neonatal Encephalopathy with Seizures	*ATF2*	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Cone-Rod Dystrophy 4	*RPGRIP1*	WT/WT	Normal (Clear)
Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration	*PRCD*	WT/WT	Normal (Clear)

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. We identified two normal copies of the DNA sequences in 10 of the mutations tested. Thus, this dog is not at an increased risk for the diseases associated with these 10 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. In addition, 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.

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.

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.

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.