Laboratory Report

Laboratory #: 142696
Order #: 63663
Ordered By: Lauren Sikkema
Ordered: July 23, 2019
Received: Aug. 15, 2019
Reported: Aug. 20, 2019

Call Name: Vesper
Registered Name: -
Breed: Australian Cobberdog
Sex: Female
DOB: May 2019
Registration #: -

Results:

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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

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. These tests were developed and their performance determined by Paw Print Genetics®. This laboratory has established and verified the tests' accuracy and precision. Because all tests performed are DNA-based, rare genomic variations may interfere with the performance of some tests producing false results. If you think these 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.