

COPPER TOXICOSIS LABRADOR RETRIEVER ATP7B (CT-LAB-B)

Copper Toxicosis (Labrador Retriever Type) is an inherited metabolic disease in Labrador Retrievers that can potentially lead to chronic liver failure. Dogs suffering from the disease will have a reduced ability to excrete dietary copper from the body resulting in excessive copper storage in organs such as the liver and tissues which can result in damage and eventually liver cirrhosis. The age of onset and disease progression can vary. Many dogs will exhibit symptoms around middle age consisting of weight loss, weakness, lethargy, vomiting, diarrhea and abdominal pain. As the disease progresses, affected dogs may develop signs of liver failure including abdominal swelling, jaundice and neurological dysfunction.

It has been determined that a mutation in the ATP7B gene is one of the causes of Copper Toxicosis in Labrador Retrievers. Dogs that inherit two clear copies of the gene lacking the mutation are more likely to maintain normal levels of copper in their liver and tissues. Dogs that inherit one copy of the mutation will typically display slightly to moderately elevated levels of copper in their liver and tissues. Dogs that inherit two copies of the mutation and hence lack a normal copy of the gene, will typically exhibit the highest levels of copper accumulation in the liver and tissues. Although dogs inheriting two copies of the mutation are at higher risk for disease progression, it is important to note that there have been reports of dogs with one copy of the mutation that exhibit copper levels similar to dogs with two copies of the mutation.

In addition to the disease-causing mutation in the ATP7B gene, there also exists a second mutation in the ATP7A gene that acts in an opposite fashion to minimize copper accumulation in the liver and tissues. The presence of the ATP7A mutation can be thought of as potentially decreasing the effect of the ATP7B disease mutation. The ATP7A gene is located on the X chromosome (i.e., sex-linked) which means male dogs can inherit only a single copy of the ATP7A gene while females inherit two copies of the ATP7A gene. Males that inherit one copy of the ATP7A mutation see reduced copper levels, while females require inheritance of two copies of the mutation to see a benefit in copper level reductions. Females are more commonly diagnosed with this disease due to the mode of sex-linked inheritance for the ATP7A mutation.

Because there are multiple factors contributing to Copper Toxicosis in Labradors, dogs inheriting the ATP7A mutation may still be at risk of Copper Toxicosis if they have also inherited the ATP7B gene mutation or other unknown mutations.

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