

## CENTRONUCLEAR MYOPATHY IN THE LABRADOR RETRIEVER

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Centronuclear myopathy (CNM) also known as Type II myopathy, or muscular myopathy is a muscle abnormality that is inherited as an autosomal recessive trait. The disorder was first recognized in the 1970's and has been diagnosed in the United States, Canada, the United Kingdom, Germany, France and Sweden. The disorder occurs with equal frequency in both male and female Labrador Retrievers. In order to be affected with this disease process the affected dog must inherit the gene for the disease from both of its parents. The parents of the affected animal appear to be normal but carry a single copy of the mutation for the PTPLA gene. When bred to a partner carrying the same mutation an affected individual can result. An affected animal bred to a carrier animal or another affected animal will also produce puppies with the disorder. However, these breedings occur with a much lower frequency that does carrier to carrier breedings.

At birth the affected puppy appears no different than its normal littermates but as the puppies begin to walk and play it becomes apparent that some of the puppies appear weak, move about awkwardly and have decreased stamina. The disease progresses with severely affected puppies walking with their lead held low and flexed toward the ground and short, stiff steps. The rear legs also have a shortened stride resulting in what looks like a bunny hop. These puppies are worse when stressed, excited or in the cold... Over time these puppies look bony although their weight is often normal. Many of them have muscle wasting of the muscles on the head and shoulders. The puppies have abnormal (decreased) tendon reflexes especially of the patellar reflexes but normal ability to hop and place their feet.

By eight months to one year of age, the signs stabilize and dogs that can still walk at that age may be functional as very inactive pets but will never be suitable as gun dogs or active family pets. Some of these dogs develop problems with the esophagus leading to aspiration pneumonia.

Clinical signs are the first clue that a Labrador may be afflicted with CNM. Routine laboratory testing on these dogs is usually normal. More sophisticated tests such as electromyography and muscle biopsy are more definitive. Histopathology of muscle tissue reveals marked difference in muscle fiber diameter with angular atrophy of muscle fiber types and ultimately myofibers with centrally placed nuclei. Outstanding research led to the discovery of the causative genetic mutation – mutation in the gene encoding protein tyrosine phosphatase-like member A (PTPLA) in 2003 by Dr. Laurent Tiret of the Alfort School of Veterinary Medicine, France. This test is now commercially available both for the definitive diagnosis of a suspicious puppy and as importantly to screen potential breeding animals for their carrier status. Visit [www.labradorcnm.com](http://www.labradorcnm.com) for more information.

Today, Labrador breeders can prevent the production of any affected puppies by simply performing the DNA test on every breeding pair. Normal to normal will only yield normal puppies. Normal to carrier will yield half normal and half carrier puppies. Carrier to carrier will yield one fourth normal, one half carrier and one fourth affected puppies. Affected to affected breedings can only yield affected puppies. The test needs only be performed once in the dog's lifetime and is very reasonably priced.