## Gentetic Test for Muscular Myopathy in Labradors

by Marilyn J. Fender, Ph.D.

Muscular Myopathy is a serious disease that affects Labrador Retrievers. It is inherited, and like many recessive diseases, dogs are affected if they get deleterious alleles from both parents or they are carriers if they get the deleterious allele from only one parent. Carrier dogs may appear normal in appearance and behavior, but they can transmit the disease to their offspring. Although this disease has likely been around for over 35 years it has become a serious concern in the last 15 years, since it can be found in several of the top field trial lines. For a description of the disease, see accompanying article by Drs. Tiret & Blot, as well as

From clinical data, we could guess that numerous field pedigrees show at least one, or sometimes more, identified or potential carrier dogs. Conformation lines apparently have less known carriers; but there has been a variety of evidence that they are far from free of the disease. For example, some breeders have infused field lines into show lines and have unfortunately chosen known CNM carriers in doing so. In other cases, show lines imported from other counties have included known carriers.

Several research groups have worked on this disease in an attempt to understand its mode of inheritance and develop a genetic marker to identify carriers. The good news is that Dr. Laurent Tiret, and his clinical colleagues Stéphane Blot, of Ecole Vétérinaire d'Alfort in France, along with colleagues from their laboratories, have succeeded in finding such a marker. Dogs in Europe appear to have the same disease that we refer to as Muscular Myopathy in the USA and Canada. They call this disease CentroNuclear Myopathy (CNM). In the accompanying article by Drs. Tiret and Blot, they briefly describe why they call the disease CNM and provide the results of their research.

Dr. Tiret developed his marker from European dogs. He then evaluated and compared swabs from known Muscular Myopathy carriers in the USA through Diane Shelton, Department of Pathology, University of California San Diego. However, he wanted further verification in prominent United States and Canadian Labrador pedigrees. I located Labradors that had been positively identified as candidate to be Muscular Myopathy carriers, by having produced a litter with affected pups and sent him swabs out of a variety of well-known pedigrees. The same mutation was diagnosed in US and Canadian dogs and therefore the CNM test initially developed for French dogs was confirmed to be suitable to identify what we call Muscular Myopathy on this continent.

## Steps for a CNM Diagnostic System in the USA

In June 2005, Dr. Tiret came to the Minnesota National Amateur Retriever Club Retriever Championship Stake to present his research at their General Business Meeting. His presentation marked the beginning of a CNM Pilot Program for introduction of the testing procedures in the United States and Canada. His trip was supported by the Retriever Field Trial News through Managing Editor, Mary Knapp and RFTN Board President Dennis Bath.

During the first stage of the Pilot, Evaluation Kits were given to 150-200 dogs. The purpose of the Pilot was to:

Evaluate the prevalence of the mutated allele in our field Labrador Retrievers;

Refine the documents and directions for the CNM evaluation; and

Expand and modify the web site, www.labradorcnm.com

The second stage, beginning August 15, 2005, was designed to continue the objectives of the CNM Pilot. Another 100 to 150 dogs will be tested. It is anticipated that by mid-September 2005, availability of the testing will go beyond the Pilot stage and be open to an unlimited number of Labradors. Announcements regarding the specifics of the progress toward full implementation will appear on www.labradorcnm.com.

( Editors note: The program has been in full implementation since September 2005 )

## Implications for Breeding

Since CNM is inherited as a simple recessive trait in Labradors, the disease allele must be inherited from both parents in order to cause CNM in a pup. Whenever two carrier dogs are bred, there is a possibility of producing affected pups and more carriers. On average, a litter from two carriers would produce 25% affected pups, 50% healthy carriers, and 25% healthy non-carriers. If a carrier was bred to a non-carrier, then no pups would be affected, but 50% of the pups would be carriers. These percentages are averages from large samples. In a normal sized litter there may be a larger or smaller percentage in each category. Since affected dogs look so different physically and lack the ability to deal with cold and harsh environments compared to normal Labradors, they would never be bred, even though they are

It is widely believed that it would be a disadvantage to remove all carriers of all potential recessives from breeding programs. Narrowing the gene pool to remove all possible carriers of the many possible recessives could result in a gene pool that is significantly constricted. Narrow gene pools often allow new recessives, not prevalent or even recognized at this time, to emerge. In addition, many of the highly desirable characteristics that are sought and enjoyed in Labradors may be lost. The identification of CNM carriers should be looked at as a tool for better breeding practices, not for total elimination of those dogs from the gene pool. This point was discussed during the Stake with Dr Tiret who also strongly supported a non-elimination strategy.

It is expected that the CNM recessive will be only the first of many recessive markers available to breeders of Labradors. There will be an enormous need for public education regarding breeding practices due to the rapid changes in genetic information that will soon be available.

## Acknowledgements

Many people are helping to ensure this project will be successful in North America. I would like to thank Dr. Mark Neff of the University of California at Davis, who is very active in the Canine Genome research. He has a strong interest in getting involved in helping Labrador owners find answers to some of their genetic problems. In 2003, Dawn Walker, an occasional client of Mike Lardy's, had arranged for Dr. Neff to visit Mike's pre-National training in California to discuss Labrador breeding concerns. Mike referred Dr. Neff to me, since I had been gathering information on various research efforts.

In October 2004, Dr. Neff went to the Netherlands to an International Conference on Canine Genetics where he met with Dr. Laurent Tiret and told him of the interest in the USA and Canada in CNM research. After returning to the USA, Dr. Neff put me in touch with Dr. Tiret.

I am extremely grateful to Dr. Tiret for all his cooperation, interest and expertise in developing a diagnostic system for this disease. The Labradors in the United States and Canada will benefit greatly from his, and his colleagues, research.

I would like to thank Dennis Voigt and Mike Lardy for their assistance in this project; as well as Retriever Field Trial News and Retrievers ONLINE for their leadership in spreading the word. Mary Knapp and Dennis Bath have been extremely helpful and cooperative as this project moves forward.

Future articles will update the general public on progress. My role is to coordinate the progress of this project and communications in the USA and Canada. Questions may be sent to me at cnm@athenet.net