MTM/CNM Teleconferences, September 2006
Below summarizes the Sept. 24, 2006, teleconference by Elizabeth Taylor, M.S., from Dr. Beggs' lab at Children's Hospital Boston.

Background: In September 2006, the Joshua Frase Foundation sponsored what is believed to be the first-ever, free, open-access teleconferences (live conferences by phone) about Myotubular Myopathy (MTM) and other forms of Centronuclear Myopathy (CNM). Families affected by MTM/CNM had the opportunity to hear directly from some of the world's leading experts on MTM/CNM and ask questions directly of the speakers.

The teleconferences were organized by Sarah Foye in New Jersey, USA, and all the fees for the conference call center and the toll-free phone lines within the USA were generously paid for by the Joshua Frase Foundation (JFF, which is a not-for-profit charity group committed to advances in MTM/CNM and similar congenital myopathies). We are also grateful to the experts, who freely gave their time to speak with us for hours on the teleconferences. We also thank the online MTM community at the Yahoo online MTM chat room, for their help in spreading the word about the teleconferences and their help in formulating the questions for the experts. That Yahoo MTM website is: http://health.groups.yahoo.com/group/Myotubular_Myopathy/

The teleconferences were a great success, generally with more than 20-25 families calling in, including callers from North America, the United Kingdom, and Australia. For some families this was the first time they had ever spoken with another family affected by MTM/CNM!

People with questions about the 2006 teleconferences, or interest in being informed of future teleconferences should contact Sarah Foye (e-mail: myopathy@optonline.net) (phone # 973-882-1996) and are also very strongly encouraged to register for free with JFF at the following website: http://www.joshuafrase.org/jff5.html , or contact Alison Frase by e-mail: alison@joshuafrase.org. Due to the rarity of MTM/CNM, is only by families joining together that we can truly make a worldwide difference for those affected by this disease. We look forward to hearing from you!

Below summarizes the September 24, 2006, teleconference by Elizabeth Taylor, M.S.

Elizabeth Taylor, M.S., Genetic Counselor and Research Study Coordinator at Dr. Beggs' Lab, at Children's Hospital Boston, part of Harvard Medical School.
Topic: "The genetics of centronuclear/myotubular myopathy: what we know and what we think we know."

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Abbreviations used within this summary:

CHB: Children's Hospital Boston, which is the site of Dr. Beggs research lab, which is actively studying MTM/CNM and similar congenital myopathies. CHB is associated with Harvard medical school.

CNM: centronuclear myopathy (the most common form of which is MTM)

MTM: myotubular myopathy

MTM1: the name of the gene responsible for X-linked myotubular myopathy.

XL-MTM (or XLMTM): X-linked myotubular myopathy

U of C: University of Chicago (the main clinical lab in the USA that does the testing for mutations of the *MTM1* gene, to confirm a suspected diagnosis of XLMTM).

Disclaimer: These teleconferences (and these written summaries) are NOT intended as medical advice and are NOT intended to be in any way a substitute for appropriate medical care from a trained health professional. Also, please note that this summary document was compiled and typed up by volunteers (MTM/CNM families) and while we believe the document to be accurate, we certainly request that interested parties should not consider this document as the final word on these MTM/CNM topics, but rather you can consider it a starting point for further discussion with you treating physicians, interested researchers, etc.

What genetic testing is available for MTM/CNM?

Currently, only one genetic test is "clinically available" for MTM. By "clinically available" we mean that it is available through a clinical/diagnostic laboratory (rather than only being available at a research laboratory). This sole test that is “clinically available” for MTM is screening for mutations in the *MTM1* gene, which is the gene affected in X-linked MTM. The child may be suspected of having MTM for a variety of reasons, including symptoms, physical exam findings, family history, or muscle biopsy showing a myotubular/centronuclear pattern [nucleus in the center of the muscle cells]. When there is a suspicion for MTM based on any of those reasons, typically a blood sample will be drawn and sent to a clinical lab to test for mutations in the *MTM1* gene.

Genes are inherited from our parents, and genes tell our body how to work. Genes can give our body information/instructions on many things, including hair color, eye color, and how our muscles work. Sometimes a person inherits a gene that does not work correctly, causing him or her to develop health problems or disease. *MTM1* is one of those genes important in muscle functioning. We all have the *MTM1* gene. Some people inherit a type of MTM1 gene that does not work properly because it contains a mutation. Like a book, a gene is made up of a specific order of letters. A change (mutation) in one of those letters (like a typo in a book) can cause the body not to understand the gene’s instructions. Mutations in the *MTM1* gene cause problems with how a person's muscles work (myopathy).

What is the benefit of having *MTM1* testing done when muscle biopsy has already been done? Why bother?

Muscle biopsy may show the classic abnormality of CNM/MTM where the nuclei are in the center of the muscle cells (rather than having that nuclear material located in the

normal location at the periphery or side of the muscle cells). This "nucleus in the center" appearance is called "centronuclear", which is how "centronuclear myopathy" gets its name. This appearance on biopsy is also referred to as "myotubular", since there is a myotubular stage during development when some immature muscle cells look this way. Some people in the past have used the terms centronuclear myopathy (CNM) and myotubular myopathy (MTM) interchangeably, whereas more recently XLMTM has been considered to be one form of CNM. Thus, the skeletal muscle biopsy showing these classic abnormalities (e.g. the nucleus in the center) can indeed give some helpful information regarding the general category of myopathy in the patient.

HOWEVER, the muscle biopsy alone does NOT reliably distinguish between the various forms of MTM/CNM. Thus, if the diagnostic workup does not continue with genetic testing after a positive muscle biopsy, then no one will know for sure the “exact” form of myopathy that the patient has (since there are multiple forms of MTM/CNM). Thus, the patient, family, relatives, treating physician, researchers, etc., need the genetic testing to precisely identify the myopathy. Identifying the specific form of MTM/CNM is important for multiple reasons. For example, some medical complications are reported to occur more commonly with certain forms of MTM/CNM than with other forms (e.g., gallbladder and liver problems with XLMTM). Knowing the specific mutation that causes a child’s MTM may also be helpful for identifying treatment options that may become available in the future.

Also, from a genetic counseling point of view, the more specific the genetic diagnosis the better the genetic counselor is able to advise the patient, family, and relatives. For example, if a child with MTM/CNM on muscle biopsy is determined by genetic testing to have a genetic mutation at the *MTM1* gene, then the child would be considered to have the XLMTM form. Since a boy's X-chromosome is inherited from his mother, the mother could be tested to see if she has that same MTM1 mutation (i.e., to see if she is a carrier). This can help with the assessment as to whether the genetic mutation was inherited (i.e., being passed down through the generations) . This is important because a family will often want to know what the risk would be of having a second child with the same myopathy. Also, if indeed the mother tests positive for being a carrier for the mutation, then it is possible (and often recommended) that female blood relatives of the mother should also be tested to see if they are carriers for the mutation. This is important, because the mother of a child with MTM may have aunts, sisters, female cousins, nieces, etc., who are at risk for having a child with MTM without even knowing it. Conversely, those aunts, sisters, female cousins, nieces might be unnecessarily fearing having a child with MTM, when really the genetics might reassure them that they are not at increased risk. The only way to know for sure is through the genetic testing. Thus, after a specific MTM1 mutation is found in a child with myopathy, the mother (and then her female blood relatives) can be screened for that same specific mutation, which is EXTREMELY helpful information so that each relative can start to understand whether she is at risk for having a child with this myopathy. If the child only had a muscle biopsy and did NOT have the genetic testing, the mother and relatives have a MUCH more difficult time evaluating their individual risks for having a child with MTM.

A number of MTM/CNM family members on the conference call reported having a MTM/CNM child that had diagnoses made mainly just based upon muscle biopsy, without genetic testing. Many of these family members indicated that they were totally unaware of the additional benefits genetic testing offers to the MTM/CNM child himself

(by clueing parents/doctors in on what medical complications to watch for in the child). Many family members also indicated they had been unaware of the benefits that genetic testing offered to relatives as well, particularly in helping with their own risk assessment and family planning decisions.

Where is the MTM1 gene testing done?

A number of MTM/CNM family members asked about where the genetic testing is done for MTM1 mutations. The answer is that in the USA the main clinical lab forming this test is the genetics lab at the University of Chicago. [The contact information for the U. of C. lab is as follows:

University of Chicago Genetic Services
5841 S. Maryland Ave., Rm. L035, MC 0077,
Chicago, Illinois 60637
Call Toll Free: (888) UC GENES
Call Toll Free: (888) 824-3637
Local phone: (773) 834-0555.
FAX: (773) 834-0556
e-mail: ucgslabs@genetics.uchicago.edu
Website: www.genes.uchicago.edu
Website info on MTM1:
http://genes.uchicago.edu/LabPDF/01MTM1.pdf

Does health insurance pay for MTM1 gene testing?

Yes, usually health insurance will pay for MTM1 gene testing. But the test is expensive and you certainly should check with your health plan and check with the University of Chicago testing lab (or whatever lab you are using, e.g. if you are outside the USA). If your doctor needs help with writing a letter of medical necessity for the genetic testing, MTM/CNM families can contact to the Foye family (e-mail: myopathy@optonline.net) for a copy of a template that the treating physician can use to help get the testing authorized. Families without insurance coverage for this testing can contact the Muscular Dystrophy Association and/or the Joshua Frase Foundation for further advice and assistance.

What does "X-linked" mean?

A significant genetic difference between a man and a woman is that a woman has 2 X chromosomes and a man has a single X chromosome and a single Y chromosome. (Chromosomes are the packages that contain our genes.) If a condition is "X-linked" it means that it is carried on the X chromosome - the gene associated with the condition is on the X chromosome. Since a woman has two X chromosomes (i.e., she has a pair of X chromosomes), she could have a non-working gene on one X chromosome but still be able to compensate for it with the second “working” copy of the gene on her second X chromosome. In that case, the woman would be considered a "carrier" for the genetic mutation, i.e. she has the mutation on one X-chromosome, even though she may not manifest with symptoms of the disease herself. Conversely, if there is a non-working gene or defect (mutation) on a male’s single X chromosome, then the male generally

has health problems or disease because he does not have a second X chromosome (and a second copy of the gene) to compensate for the non-working one.

Now, getting specifically back to MTM/CNM, it is notable that the most well-known form of these myopathies is XLMTM (X-linked MTM). XLMTM is caused by a genetic mutation on the MTM1 gene, which is located on the X-chromosome. Thus, although a woman may be a carrier for a genetic mutation on the MTM1 gene (on one of her two X-chromosomes), commonly the woman will not manifest any symptoms of MTM. However, with each male pregnancy, there would be a 50% (1 in 2) chance that each of her sons will inherit the defective X-chromosome, and thus a 50% (1 in 2) chance that each of her sons would have XLMTM. Likewise, there would be a 50% (1 in 2) chance that each son will inherit the working X-chromosome and thus a 50% (1 in 2) chance that each of her sons would be healthy, without any myopathy. Meanwhile, there would be a 50% chance that each daughter would inherit the defective X-chromosome (and thus, those daughters would be carriers, but usually not clinically affected themselves), and a 50% chance that each daughter would inherit the mother's normal X-chromosome (and thus those daughters would not only be clinically healthy, but they would not even be carrying the defective gene).

[The web site for Dr. Beggs lab has some diagrams and professionally-written descriptions of X-linked inheritance and other inheritance patterns. Here is the link for that website: http://www.chb-genomics.org/hndp/research/beggs/xl.html]

Are all forms of MTM/CNM X-linked?

The answer is no. Although the most well-known form of MTM/CNM is XLMTM (which is X-linked), other forms have been discovered. In late 2005, Dr. Alan Beggs was a contributing author for the landmark paper that first reported an autosomal dominant form of CNM. Dr. Beggs (a Harvard geneticist at Children's Hospital Boston, USA) collaborated on this publication with important MTM/CNM researchers in France. This is a great example of how international researchers can work together to make advances in MTM/CNM research. Together, they found and reported that the autosomal dominant form of CNM can occur from mutations in the "Dynamin 2" gene, known as *DNM2*, on chromosome 19. Chromosome 19 is NOT the X-chromosome, and thus this is one of the forms of CNM that is not X-linked. Therefore, we all generally have two copies of the *DNM2* gene. Certain mutations in *DNM2* can cause the gene not to work properly in the muscle. Individuals who inherit one of these mutations will have CNM. In this form of CNM, a second working copy of *DNM2* is not enough to keep a person from developing symptoms. Therefore, *DNM2* CNM can be inherited directly from a parent to a child. This type of pattern is called dominant inheritance. A parent with DNM2 CNM has a 50% (1 in 2) chance of passing on their non-working copy of DNM2 (with the mutation) to each child and a 50% (1 in 2) chance of passing on their working copy of the DNM2 gene.

Since *DNM2* mutations have only recently been implicated as the cause of autosomal dominant CNM, testing for these mutations is not yet available at "clinical labs". Thus, practically the only way a patient/family can be tested to see if a dynamin 2 mutation is causing their myopathy would be for the patient/family to enroll with a research laboratory that is specifically studying MTM/CNM. There are some specific protocols about how a "research" lab can distribute the results (e.g., in coordination with the

treating physician) and whether those "research" results can officially be considered "diagnostic" (often a positive finding by a research lab would need to be confirmed by specifically repeating just that portion of the test at a "clinical" lab). It is important to realize that, unlike clinical testing, research testing is generally a long-term process, and there is often no guarantee on when a family may get results from research testing, if ever.

Thus, scientists so far have identified an X-linked form of MTM/CNM (XLMTM, with mutations on the *MTM1* gene), and have also identified an autosomal dominant form of CNM (dynamin 2 CNM, sometimes abbreviated *DNM2*-CNM). Meanwhile, researchers are still looking for the gene mutation responsible for the autosomal recessive form of CNM. It is an area of active research to find the responsible gene mutation in those cases where the *MTM1* gene testing is normal. Thus, it is particularly important for families to enroll in research when a standard testing (*MTM1*) fails to identify the genetic cause of the MTM/CNM.

How can females manifest MTM? (How can a female manifest symptoms of an X-linked disorder like XLMTM?)

Females have two X chromosomes, and theoretically any cell within her body could get by with just one of those X chromosomes (the other one serving as a backup). Thus, one of the two X chromosomes will be "turned off" in any given cell within a woman. Statistically, it is 50-50 chance (50% chance) as to which of the two X-chromosome gets turned off within a given cell. This is called "x-inactivation", since one of the X-chromosomes essentially gets inactivated, while the other one is expressed. Thus, on average, a woman will have 50% of her cells expressing one X-chromosome and 50% of her cells expressing the other X-chromosome. In some females, it is not a 50-50 split, but rather one X-chromosome is expressed more than the other. For instance it can be 90-10 (i.e., 90% of the cells expressing one X-chromosome and only 10% of the cells expressing the other X-chromosome). In some females, the one that is being significantly expressed is the one that has the genetic mutation. Thus, although on average X-linked disorders like XLMTM only cause significant symptoms in males, some females can have symptoms. Although it is uncommon for women carrying a *MTM1* mutation to actually have clinical symptoms of myopathy, it is certainly possible and indeed female cases like this have already been documented within the medical literature. If there is a male with XLMTM (*MTM1* mutation) who has female relatives with symptoms of possible myopathy, they should probably consult with both a neurologist and a genetic counselor, to discuss undergoing testing to see if they not only carry the *MTM1* mutation but may actually be a "manifesting carrier". ("Manifesting" meaning that the person actually has symptoms due to this dictation.) Again, this is one more reason why the genetic testing is so important for families affected by MTM/CNM.

Is there a test for a mom who is technically not a MTM carrier?

Usually mothers are either a carrier or non-carrier, but in some cases there is what is called mosaicism, where the mother has the mutation in some of her cells, but not others. For instance, the mutation may be in a woman’s blood cells but not in her muscle cells. In this situation, the mutation most likely occurred for the first time in the mother (i.e., the mutation occurred spontaneously years ago, when the boy's mother was herself developing in utero, prior to her birth, without the mutation having been present in her

parents/ancestors). In a case like this, it would be expected that there would be little or no risk of this mutation being present in the mother's sisters, nieces, etc.. But again the genetic testing plays a critically important role in determining whether this scenario is present in any given family. Without genetic testing, the family/relatives would have no real idea whether the boy's mother was a typical "carrier" (with her siblings, cousins, nieces, etc. being at risk) versus whether the boy's mother (or the boy himself) was the first person in the family to have that mutation. Such information is important for families/relatives so that they may understand their potential risks or lack thereof.

If one MTM/CNM child presents with symptoms at a young age, what are the chances that other children in the same family would present at a similar age?

XLMTM typically presents in infancy. In most families, if the affected child expresses at a young age, the other children would also express at a young age (if affected). Thus, if one boy has XLMTM and he had symptoms since birth, and his brother is 10 years old and never had any symptoms at all, then most likely the 10-year-old is totally unaffected and does not have the mutation. Thus, most likely the 10-year-old will not develop XLMTM later on. Of course, if there are any concerns in a specific case than the family should probably consult a neurologist and a genetic counselor for a more personalized analysis.

Are MTM1 mutations the only form of X-linked MTM?

The short answer is that we don't know for sure yet. *MTM1* mutations are the only form of X-linked MTM that has been identified so far. Saying that another way: in a patient with MTM, if the mutation is on the X-chromosome, the only place on the X-chromosome where we currently know to look is at the *MTM1* gene. However, a significant number of patients with biopsy-proven myotubular/centronuclear myopathy will end up testing negative for mutations at the *MTM1* gene. It is possible that the current genetic test may fail to detect 100% of the mutations that really are occurring at that *MTM1* gene (since no test is 100% perfect). However, it is likely that most patients who test negative for *MTM1* mutations have mutations elsewhere, in other genes. Some of these "other" responsible genes might theoretically be located elsewhere on the X-chromosome (i.e., still on the X-chromosome, but not at the *MTM1* gene). But it is perhaps more likely that the "other" responsible genes are likely located on chromosomes other than the X-chromosome. A chromosome that is neither an X-chromosome nor a Y-chromosome is called an "autosomal" chromosome. Thus, some forms of MTM/CNM are X-linked while others are autosomal, but this distinction basically depends on whether the mutation is on the X-chromosome or not.

If there is a different x-linked MTM than *MTM1* could they express later?

It is theoretically possible. At this time there is only one x-linked mutation (*MTM1*) known to cause MTM/CNM, but there may theoretically be other forms also located on the X-chromosome. If a condition is x-linked, a female could have mild symptoms (as discussed elsewhere within this teleconference). Generally, if that *MTM1* mutation is ruled out, then we can think that that form is probably not present in the family. (If the MTM1 gene testing is normal, then "most likely" that person does not have an X-linked form of MTM/CNM, but admittedly this is not 100% certain.)

How can a mutation occur? (How does a mutation first start?)

When the sperm and egg unite in utero, they create a single cell that must multiply millions and millions of times to create an infant. This process of multiplying and copying is not 100% perfect. Occasionally, an error (genetic mutation) occurs. In fact, ALL people have some genetic mutations in their DNA. Often, the genetic mutations do not produce clinical disease in the person, because the mutation is not affecting a critical function, or the mutation can be compensated for by having other genes that are normal. In addition, some new mutations occur during the development of eggs and sperm.

In XLMTM, the mutation at the MTM1 gene can occur spontaneously during the development of the egg, the boy in utero, or perhaps one generation earlier when his mother was developing in utero. However, most commonly it is thought to have been passed down through the prior generations. "Stillbirth" and neonatal deaths in prior generations might potentially have been due to XLMTM that was never diagnosed at the time. Ancestors and relatives who appear healthy might be carriers of the mutation. The only way to know for sure is by genetic testing of the affected child and potentially the family members and relatives (depending upon the results of the genetic testing in the affected child).

What about a sister who has a sibling with MTM, should she be genetically tested?

If the child affected by MTM has a genetic test that identifies the responsible mutation, then there are certainly benefits to testing relatives, including the sister described in this question. Ggenetic testing may help the girl and her family to understand whether or not she is a carrier (which has implications for her own eventual family planning, etc.). Many genetic specialists recommend waiting for carrier testing of females until they reach the age of 18 years and can make their own decision about testing. However, if the sister has some neurologic symptoms (weakness, fatigue, muscle aching) then consultation with a neurologist and a genetic counselor may be helpful. In fact, if neurologic symptoms like these develop in any relative (whether male or female) of someone with MTM/CNM, consultation with a neurologist and a genetic counselor may be helpful to help determine whether the symptoms are related to MTM/CNM or not.

When will testing for the Dynamin 2 be available for families?

The lab in France and the Beggs lab are working to understand what the gene does and how to screen for it. Since it was first reported in the literature less than one year ago (late 2005), currently there are no "clinical labs" offering the testing yet. Further research is being done on this autosomal dominant CNM, so that eventually we hope there will be a clinical lab test available for the dynamin 2 mutations, just as there currently is a clinical lab test available for *MTM1* mutations. We do not know the exact time frame for such clinical lab testing for dynamin 2 mutations. Meanwhile, as noted in one of the other answers above, the testing for dynamin 2 mutations is being conducted within research labs that are specifically studying MTM/CNM.

Why are some diagnosed with “Centronuclear” Myopathy and others are diagnosed with “Myotubular” Myopathy?

Sometimes it is just a matter of the term that a given pathologist (the doctor who analyzes the muscle biopsy under a microscope) is most familiar with using. The literature is now using Centronuclear Myopathy as the broader/inclusive term, and MTM is used to describe the x-linked form. Thus, CNM would be the umbrella term under which a number of subcategories would be included, such as XLMTM, dynamin 2 CNM, etc. It is still being worked on to change the name to Centronuclear for the broad term and MTM as a sub-group. In 2006, there was a review article published on MTM/CNM, where Dr. Beggs and Dr. Pierson (also at Harvard) clarified this point. But this new terminology may take years to catch on within the medical community.

Is it true that Dr. Beggs' lab (at Harvard/Children's Hospital Boston) is considered a major collecting point for MTM/CNM families?

Yes. Although Elizabeth Taylor was very humble in her answer, most of us families on the call were in agreement that at least within the USA, most of the current research on MTM/CNM is being done at Dr. Beggs' lab in Boston. One of the family members indicated that within the medical literature specifically on MTM/CNM, Dr. Beggs is among the most published researchers in the world. He also does substantial work on other, similar congenital myopathies. Those family members on the conference call who have met with or spoken with Dr. Beggs indicated that they have been inspired by his compassion and dedication to MTM/CNM patient/families worldwide.

MTM/CNM families are encouraged to register with research centers that are specifically focusing on MTM/CNM. This is extremely important since the rarity of MTM/CNM means that it is challenging for researchers to obtain a large enough sample size for meaningful research. If all of the MTM/CNM cases are spread out across the country (only a couple of cases at any given hospital in any given state) it would be extremely difficult for any researcher to make substantial progress, because he/he would only ever see a couple of cases of MTM/CNM. The most effective way to move MTM/CNM research forward is by actively researching many dozens of families with MTM/CNM, so that the researchers can analyze these similarities, differences, etc.

MTM/CNM families have a crucial role to play in helping move the research forward to help those affected by MTM/CNM. Without involvement from patient/families, advances cannot be made. The involvement typically requires filling out some paperwork (e.g., consent forms allowing the researchers to examine the medical records and examine the tissue from any muscle biopsy that was already done). Typically, the pathology department in a hospital (where the biopsy was performed) will freeze the muscle biopsy tissue and may hold it there for a decade or more. Thus, many families have muscle tissue sitting in a hospital freezer somewhere. A small sample of that tissue could be very helpful to researchers trying to help cure the disease. DNA samples can also be obtained, either during a time when the patient/family would be having some blood drawn for some other reason anyway, or perhaps a DNA sample obtained just from saliva (spitting into a container).

If my family was in Gail Herman's study years ago, has my research involvement automatically transferred over to Dr. Beggs lab?

No. Typically the patient or family must make a specific request to become involved with research at a new lab. During the previous decade, Dr. Gail Herman made significant

advances in MTM/CNM research and many families were involved. Some families indicated that they thought there materials (records, DNA samples, muscle biopsy slides, etc.) would automatically be transferred to whatever research center is currently doing the latest MTM/CNM studies. But that is not how it works. The family generally needs to initiate contact with the new lab that they want to be involved with. In fact, it is perfectly fine to be involved with MULTIPLE research centers, as long as the family lets each research center know which other sites are registered at. (That way if the two different research centers are ever collaborating, they will know when they are talking about the same person/family, rather than thinking they have stumbled across two totally different individuals/families with the same findings.)

Do you need more MTM/CNM families to join research studies?

Yes! Definitely! Involvement is actively encouraged from families affected by ALL forms of MTM/CNM (whether the MTM1 test is abnormal or not), worldwide. Generally, families must initiate the contact with the research labs (you can reach out to the research labs, but generally they can not reach out to you).

Please contact Elizabeth Taylor, M.S., Genetic Counselor and Research Study Coordinator at Dr. Beggs' Lab, at Children's Hospital Boston, part of Harvard Medical School.

Telephone: 617-919-2169
Email: etaylor@enders.tch.harvard.edu
Website: www.childrenshospital.org/research/beggs

Who can families contact to be included in additional, upcoming free teleconferences on MTM/CNM?

People with questions about the 2006 teleconferences, or interest in being informed of future teleconferences should contact Sarah Foye (e-mail: myopathy@optonline.net) (phone # 973-882-1996) and are also very strongly encouraged to register for free with JFF at the following website: http://www.joshuafrase.org/jff5.html , or contact Alison Frase by e-mail: alison@joshuafrase.org. Due to the rarity of MTM/CNM, is only by families joining together that we can truly make a worldwide difference for those affected by this disease. We look forward to hearing from you!

Also remember the Yahoo MTM website for chatting with other MTM/CNM families worldwide: http://health.groups.yahoo.com/group/Myotubular_Myopathy/