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Insiders Guide to Muscle Biopsies

Go to our Resource tab on our website to download a 15 pg guide to biopsies that covers numerous areas of consideration with a list of typical questions a person considering a biopsy can ask the provider.

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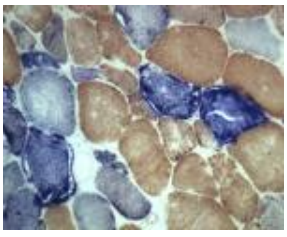
Muscle Biopsies: What are their limitations?

When patients are suspected of having mitochondrial disease, muscle biopsies are often obtained for respiratory chain enzymology, histology studies and some gene testing. Abnormal results often come in the form of certain complex deficiencies such as a complex I defect or a combination of complex deficiencies.

In most cases, though, a specific gene defect is NOT identified. Because abnormalities in mitochondrial enzyme complex assays can be due to a true disease process in the mitochondria (due to a mitochondrial or nuclear gene mutation) or a secondary process that essentially makes the mitochondria sick when they were initially programmed to function normally (examples include Parkinson's disease, Alzheimer's disease and other disorders that "poison" the cell environment and make the mitochondria sick) many practitioners will no longer rely just on enzymology to diagnosis a patient with a mitochondrial disease, particularly if their clinical symptoms, such as hypotonia (low tone) can be caused by many diseases.

In recent years, we have re-diagnosed a number of patients who were thought to have a primary mitochondrial disorder with another disease including chromosome abnormalities. Conversely, we have found mitochondrial DNA changes in some patients who had many mitochondrial disease related symptoms and negative biopsy enzymology. As such, we are extremely cautious about labeling patients with or without mitochondrial disease if an extensive evaluation prior to biopsy was not completed (for example did not obtain chromosome microarray studies) or if no gene mutation is identified in those biopsied and labeled abnormal when they have more "generic" symptoms that can be seen with many disorders.



If you have questions about your evaluation, you should discuss your work-up with your provider.

VMP is available for second opinion consults if requested. On our website we have a downloadable 15 pg insiders guide to biopsies that covers numerous areas of consideration with a list of typical questions a person considering a biopsy can ask the provider. More information on biopsies is available on our website at www.virtualmdpractice.com/musclebiopsy.html.

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Lectures on YouTube and Our Website

We have placed on YouTube snippets (about 9 minutes) of Dr Kendall's various presentations. The full 40 minute long presentations are located on our website under the Resource tab.

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Expansion of Mitochondrial Disease Gene Testing

Primary mitochondrial disease affects the intrinsic ability of the mitochondria to function properly by altering how it's many components, like pieces to a puzzle, come together to create energy. Primary mitochondrial disorders are caused by changes in mitochondrial or nuclear genes. Genes are our genetic blueprint. They are units of heredity that determine everything about us including how tall or short we are, the color of our hair and eyes and whether or not we make all of the many proteins that ultimately come together to make our energy packets known as ATP.

For about twenty years we have known a lot about the mitochondrial genes or those inherited exclusively through our mothers. These 37 mitochondrial genes are housed inside the egg cells of our moms. The remaining genes involved in energy production are inherited or passed on to us through both parents, are locating in the center of our body cells known as the nucleus, and consist of hundreds of genes. Current technology allows us to look at only a handful of these nuclear genes.

A number of studies suggest that 75% to 90% of mitochondrial disease in pediatric patients is due to changes in the nuclear genes inherited through both of our parents. As such, the DNA testing available to date does not allow us to find the gene causing mitochondrial disease in all patients.

Several laboratories are currently working on testing development that will identify essentially all genes causing mitochondrial disease. Once available these noninvasive studies will all but eliminate the need for more invasive testing, including biopsies, in most cases. This will also negate the ambiguity that can come with those often not-so-clear testing results. Updates on availability of this testing will be posted on our website.

Yearly Visit and Multi-System Screens

It is time again for your/your child's annual visit and customary blood and urine studies. What are the recommended tests and how do they help manage your/your child's disease? Most mitochondrial specialists will obtain a battery of blood and urine studies yearly to help determine if the problems facing a given mito patient are contained to those known and addressed at your visit or include others that have not yet caused you/your child problems. These tests typically include a standard blood count to check for anemia, complete metabolic panel to look for things such as sugar levels, kidney and liver function tests, CPK, a muscle enzyme that can be elevated if your/your child's muscle is showing mitochondrial stress, and thyroid function tests for low thyroid activity. Others like carnitine profile and CoQ level help determine if adjustments are required in medication doses. Depending on the given patient additional studies such as hearing and vision screen, EKG and Echocardiogram may be warranted. Like testing such as cholesterol levels done on aging adults, the goal of the multisystem screen is to detect and treat issues before they become problematic. Approach varies somewhat between practitioners but don't hesitate to ask your provider what he/she recommends and why. We are happy to discuss our recommendations and the reasons

why with you.

Patient Advocacy

We believe that good medicine does not end with the consult. Patient Advocacy services assist patients and families to access necessary care and resources to improve their quality of life. Whom better to explain how this area of service changed a family's life than to post a letter from a mother who utilized this service:

Raising a child with special needs has offered many opportunities to see the need for a patient advocate. With IEP's, section 504 plans, homebound school etc., there was always the requirement for doctor recommendation and approval. However, we've recently learned that the transition to adulthood offers equal or even greater challenges.

Dr. Kendall has provided outstanding medical care to our son for the past 3 ½ years and she has recently filled a critical need with her role as a patient advocate.

I am the mother of a 19 year old (almost 20) young man with a mitochondrial disease diagnosed via muscle biopsy in August 2006. It wasn't until the past year that we came to really understand the need for a doctor who would not only provide medical care, but would also be our child's advocate, who understands his condition, how his body works and how this illness effects day to day living. Along with the physical problems associated with his illness, our son also suffers from severe behavioral, cognitive, and learning problems. But, when he walks in a room, he looks like a relatively healthy 19 year old.

In May of 2008 (shortly after our son's 18th birthday), we started the application process for social security disability and medicaid. In June 2009, our son graduated from high school (not without a lot of assistance and support) and we began the transition to adulthood. Our top priority and major concern was health insurance. If our son was not a full time student, he was no longer covered under our family plan.

In June 2009, after more than a year of waiting, answering questions, filling out forms and attending appointments, we were facing our second rejection from social security. People (the doctors involved) simply do not understand the devastating effects of mitochondrial disease. We realized we needed help.

Shortly before, we had learned that Dr. Kendall had opened Virtual Medical Practice and was offering consultation and patient advocacy services in addition to direct patient care. Within 2 weeks of requesting Dr. Kendall's involvement (at very reasonable prices), we had a detailed letter from her explaining mitochondrial disease, our son's symptoms and his limitations. Within 3 months of that, we had an approval for SSI and Medicaid. A huge burden was lifted.

At the same time, we were also applying for our son to remain on our insurance as a disabled child. Again, a rejection. When we got Dr. Kendall involved, she again filled out paperwork, wrote a letter (or tailored her original letter), and within a month or so, another approval. So, within two months of

applications, we had both private and public insurance. This was no small feat as our medical bills would be unaffordable without insurance. In addition, we had our son registered for four classes at the community college. He was set up for failure which of course added to his frustration and exhaustion.

We can see how this assistance continues. For our local community college, we needed paperwork to qualify John for disability services. Again, Dr. Kendall quickly completed the required documents (my son's disability counselor was shocked by the support!). And, we recently met with our local department of social services to obtain a case worker and learn about public services. The case worker and psychiatrist reviewed Dr. Kendall's documentation and we already have a verbal approval for services.

For those of you that suffer from mitochondrial disease or are a parent of a child with mitochondrial disease, you can relate to the fact that every day is a challenge. Having a doctor that is truly an advocate has made a huge difference in our lives. My son now takes one class, and works 8 to 10 hours per week. That is the schedule that provides him the highest quality of life. On Dr. Kendall's website, she lists the service as "Patient Advocacy – assisting patients and families to access necessary care and resources to improve their quality of life". Dr. Kendall has done just that for our child and our family and we are grateful.

PM

Mitochondrial and Its Association to Autism Lecture

The question of whether or not mitochondrial disease is a cause of or trigger for disorders in the autistic spectrum has been a topic of hot debate and concern over recent years. Several recent studies have indeed linked mitochondrial disease and autism spectrum disorders (ASD) in a small but significant number of cases (4 out of 100 cases of ASD have mitochondrial disease). Despite this association, many critical questions remain unanswered. Among those questions is which patients with ASD should be evaluated for mitochondrial disease. Although mitochondrial disease has proven to be one of the most commonly identifiable genetic causes of ASD the numbers are still small and may only warrant investigation under certain circumstances. While this topic was discussed in a previous newsletter, a recent lecture by Dr Kendall to the North Fulton County Autism group explores this general topic in more detail and can be viewed in its entirety on our website through the following link: www.virtualmdpractice.com/Lectures.html or a snippet on YouTube: www.youtube.com/watch?v=tx_bNykd3Pc