

## MUSCLE BIOPSY... YES OR NO?

A Dialogue with Sumit Parikh, MD

Additional comments provided by Russell Saneto, DO, PhD and Carlos Moraes, PhD

As tests for mitochondrial disease continue to improve, the UMDF has received questions about whether there is still a need to have a muscle biopsy in order to diagnose mitochondrial disease. There is some confusion as to why a muscle biopsy may be ordered in some cases but not in others. The following dialogue is intended to provide additional information about this complex issue, bearing in mind that there may be some differences of opinion among mitochondrial disease specialists and that this information may change as technology advances.

**Question: Besides muscle biopsy, what other kinds of tests can be used to diagnose mitochondrial diseases? What can and cannot they specifically detect? For example, can both mitochondrial DNA (mtDNA) and nuclear DNA (nDNA) mutations be detected now in blood samples?**

Most mitochondrial evaluations begin with biochemical studies in blood and urine. Genetic testing (same thing as DNA testing) via blood, urine, skin, cheek swab, etc, is currently still very limited, although better than it used to be, and may soon improve dramatically if newly developed genetic tests prove to work well.

Let's remember that the nucleus of each cell has DNA (called nuclear DNA, or nDNA) that directly impacts the mitochondria, but that also the mitochondria in each cell have their own DNA (called mitochondrial DNA, or mtDNA). A genetic

defect in either the nDNA or the mtDNA can cause a mitochondrial disorder.

While non-invasive or minimally-invasive tests are already available for mitochondrial DNA mutations, their usefulness is limited by the previously-stated fact that many mitochondrial diseases are the result of mutations in nuclear DNA rather than in mitochondrial DNA. Nuclear DNA testing is laborious, costly, one gene at a time, and still very hit-or-miss unless a specific gene mutation is correctly suspected and identified. Most of the recent mitochondrial genome tests that have recently been announced via press releases by labs only test the mitochondrial DNA.

There are approximately 1500 distinct protein products involved in mitochondrial function and structure. In theory, any one of these may cause disease. Currently, we only know of approximately 70 or so nuclear genes (genes in the nucleus) involved in disease. So, as you would expect, we are not ready to sequence every gene involved in mitochondrial disease as we do not fully know the full range of genes involved in disease.

**Question: When you run blood/urine tests first, are you referring to DNA tests (and not just tests such as lactic acid levels)? If these tests indicate mitochondrial dysfunction, and/or a genetic mutation, why might the patient then need a muscle biopsy, too? What else are you looking for?**

Most of the time blood/urine testing is focused on biochemical and not genetic studies; if genetic studies are done, currently they are limited to a handful of nuclear DNA genes or mitochondrial DNA sequencing if a maternal inheritance or mitochondrial DNA syndrome is strongly

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**Energy for Life: New England Walkathon was held on May 23, 2010. "We are all worth a cure!"**

**See pages 4-7 for more events and fundraisers!**



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## FROM PAGE ONE

suspected. If a genetic diagnosis is made, a muscle biopsy is not needed. If all we have is biochemical evidence in blood/urine, then a muscle biopsy may flesh out the extent of the problem and allow for more focused genetic testing.

One of the things that we need to prove concerning a pathological mutation in a gene is whether the mutation is in fact “causal” to inducing a mitochondrial disease. In many instances, the muscle biopsy is the best tissue to use as a standard for mitochondrial function. When a defect is noted in the muscle enzyme analysis, then we have a “functional” marker (significant decrease in enzyme activity) for function. If we induce the gene mutation in

question, in let's say a yeast cell, and can reproduce the enzyme activity defect, then we have begun the process to “prove” that this mutation is “causal” for inducing mitochondrial disease.

Using other criteria, such as this mutation is not found in similar “normal” patients of the same ethnic group, then we have a direct correlate of gene mutation and function. At this point we can then use the simple, but likely very expensive gene sequencing testing to define the mitochondrial disease in others.

**Question: What other test results, besides genetic, might you be looking for in a muscle biopsy?**

At bare minimum, when evaluating for mitochondrial disease, muscle should always get sent for enzymology (ETC testing), histology, electron microscopy, mtDNA quantification, and mtDNA deletion analysis. Unfortunately, some physicians only order ETC and histology but nothing else. When possible, we also obtain CoQ levels and western blotting. Genetic testing for mtDNA mutations or mtDNA gene sequencing in muscle are rarely obtained routinely. All of the above can be obtained on frozen muscle tissue, though enzymology testing in frozen muscle might be less sensitive and prone to false negatives and positives due to difficulties with specimen handling.

**Question: Even if another doctor didn't order all the right tests, wouldn't there be some frozen muscle tissue left over that could be**

**used to run the additional tests you want to order?**

Not always. At times there is not enough muscle left. And even when there is enough muscle it has not always been stored or processed properly to allow for more testing to get added on. Lastly, when we have asked for additional testing in available muscle, we have had issues with local providers not always obtaining it.

**Question: Just for further clarification, can all necessary DNA tests be run on a muscle biopsy?**

Since many mutations are in nDNA, blood is an acceptable sample by most labs. Some mtDNA point mutation testing can now be obtained with better accuracy in blood. For mtDNA deletion and depletion testing, muscle is still a MUST. (One example is the mtDNA deletion that causes Kearns-Sayre Syndrome.)

To state it another way - nDNA testing has always been obtained in blood; mtDNA testing was limited in blood and hampered by issues of not being able to

detect low heteroplasmy (muscle was necessary) - but we now have the ability to detect low heteroplasmy mutations - so it can be obtained in blood. The point of mitochondrial disorders mostly being due to nDNA mutations still stands - so mtDNA testing (in blood or muscle) is low yield.

**Question: What is your approach to testing for mitochondrial diseases?**

My approach is to try to prove as much as reasonably possible with genetic and biochemical blood/urine testing, and only proceed with a biopsy once a) mitochondrial dysfunction is strongly suspected or seen on biochemical tests in blood/urine, b) parents/patients understand the strengths/weaknesses of the biopsy, c) and parents/patients understand that the biopsy may still leave us with partial diagnostic information and no genetic diagnosis (either because the specific mutation could not be located in the biopsy tissue, or the muscle results were falsely negative, or the full spectrum of mitochondrial testing was not obtained if sent from a local/regional hospital). It is also essential to exclude other genetic and metabolic disorders that may lead to similar symptoms.



**Dr. Sumit Parikh**



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### **UMDF MISSION**

*To promote research and education for the diagnosis, treatment and cure of mitochondrial disorders and to provide support to affected individuals and families.*



# ASK THE MITO DOC<sup>SM</sup>

**L**iving with mitochondrial disease presents many twists and turns and a maze of questions. UMDF is pleased to offer answers to some of those questions as taken from *Ask the Mito Doc<sup>SM</sup>* at UMDF.org. Please note that information contained in *Ask the Mito Doc<sup>SM</sup>* is for informational and educational purposes only. Such information is not intended to replace and should not be interpreted or relied upon as professional advice, whether medical or otherwise.

Responder for this issue: Marni Falk, MD, of The Children's Hospital of Philadelphia, PA, and Russell P. Saneto, DO, PhD, of Children's Hospital and Regional Medical Center, Seattle, WA.

### THE QUESTION IS...

I have a son with clinical manifestations (exercise intolerance, seizures, dysautonomia, global delays, ptosis, lack of cognitive development, regression with illness, migraines, pain, PDD-NOS, etc) and a mtDNA mutation of 4216 T>C. As his mother I also have similar clinical manifestations minus the learning delays, as well as lupus and mixed connective tissue disease, and am awaiting the genetic tests. We also have a strong family history on the maternal side. Would this be considered a mitochondrial cytopathy or mitochondrial myopathy?

### RESPONSE FROM MARNI J. FALK, MD:

Maternally-inherited disease manifestations affecting multiple body systems as you've described certainly raises concern for a "mitochondrial cytopathy." However, it is at times difficult to definitively understand if a particular mtDNA sequence variant is disease-causing. The sequence variant has to be interpreted in the context of the overall clinical history, family history, findings in other individuals with the same variant, understanding of whether that variant is present in only a fraction of the person's mtDNA (heteroplasmy) or all of the person's mtDNA (homoplasmy), and results of other biochemical screening studies in blood, urine, and oftentimes, muscle. Ultimately, it may even be necessary to have a researcher study the mitochondrial function of cells containing the specific mtDNA mutation in question in a common nuclear gene

background (through generation of cell hybrids, or "cybrids").

In regard to the specific mtDNA mutation you describe, recent articles have found that the T4216C mtDNA is a sequence variant that is found almost exclusively among Caucasians. Very recent research has linked it with an increased risk of multiple sclerosis, as well as poor wound healing after a serious infection, trauma, or burns. Thus, there has been suggestion this particular variant alters the function of complex I of the 5-complex mitochondrial energy-generating respiratory chain. To move toward answering whether this mutation is "the" cause of mitochondrial disease in your family, it may be helpful to not only know if it is heteroplasmic or homoplasmic, but also the activity of complex I in affected individuals' muscle and/or skin cells. If low, it would provide independent evidence of a mitochondrial disorder. It is very often necessary to also evaluate the specific "variant" in question in the background of all of the other mitochondrial DNA variants identified in your family (your haplogroup).

### THE QUESTION IS...

When my mito complex 1 condition and POTS presented itself when I was 27 (over 15 years ago) I developed many of the normal symptoms and became highly allergic to citric acid in any products. The citric acid attacks lowered my base weight from 165 to 125. Despite large volumes of food and supplements there was no change in my condition. I was and still am able to work full time. Three years ago we finally discovered a partial pattern to my symptoms and with a very rigid diet I was able to alter my base weight back to 152 over a 2 year period. That weight was the best since I took ill. Nearly a year ago while still showing a slow steady improvement something changed but none of the testing showed anything remarkable. I am now more prone to colds this year. Despite a very balanced diet and vitamin/mineral supplements I am slowly weakening, losing weight and feeling generally weak, loss of motor skill fine movements, muscle spasms including heart at times and a low appetite. This is unusual since my appetite is not affected by many things typically and I don't show any gastro issues. I am urinating about every hour from my high sodium intake but this had not changed since my weight increase. I don't seem to be effectively utilizing the proteins, carbohydrates and minerals. The only strange results show that my amylase is elevated; phosphorus, Vitamin D, and magnesium are all dropping, despite supplements. Calcium is normal but my bones are thin but stable.

Can you think of any chemicals, hormones or suggest anything we can test to isolate the cause of the change?

### RESPONSE FROM RUSSELL P. SANETO, DO, PHD:

I am sorry that you are backsliding in your health. It sounds like you have done a magnificent job of finding a way to put back weight and remain active. Unless you live in a bubble, you are bound to get ill, and viral illness is always a threat to exacerbate a mitochondrial disease. It might be that one (or more) of you flu-like illnesses have pushed you off your previous precarious perch of well being and health. Without knowing more detail, I can answer only in generalities. The body is pretty good at using what it is given to provide the essentials for building up what our daily lives tear down. Even in the face of a mitochondrial disease, it will function pretty well (when not ill). One of the problems in mitochondrial disease is that finding or demonstrating a subtle change in a cell/organ function is not always easy as the change is often subtle, and since multiple upstream and downstream functions are also altered, the causal change is not always detectable. What I have found with most of my patients is that sometimes we need to look at the trends but not necessarily the numbers. For example, a few percentages below or above normal range values can be misleading-lab error, just eaten meal, etc. A slow trending may be related to a new health plateau due to the disease. Sometimes we may or may not be able to alter this change. Some simple ideas to think about: 1)-Calcium, vitamin D, and phosphorous are inter-related. Your thin bones let me know that you have been using bone to maintain levels. You probably need to better balance this, so I would talk to a nutritionist about increasing vitamin D supplementation and intake of more usable calcium. Also, there is a hormone that helps regulate bone and calcium that you can have your endocrinologist check-parathyroid. 2) - giving too much magnesium can be a problem, at least from several of my self-medicating patient histories. What does your nephrologist think about what you are supplementing and why your kidneys are not regulating your magnesium?

Submitting questions to *Ask the Mito Doc<sup>SM</sup>* is a benefit of the UMDF "Energy" membership. If you are a member and would like to submit a question, log in to the UMDF website using your user ID and password. If you would like more information on becoming a member of the UMDF, email [info@umdf.org](mailto:info@umdf.org).

## CHAPTER EVENTS

### ATLANTA CHAPTER

- **April 24, 2010.** The 3rd Annual All Aboard for a Cure - One Mile Walk and Family Fun Day was a success again, despite the less-than-ideal weather. While the wet weather kept some away, over 300 people attended the event in Historic Norcross where the rain took a break just long enough to get in a one-mile walk in support and raising awareness of mitochondrial disease. Families, friends and over one hundred volunteers then gathered in Thrasher Park with live music from Poseidon and The Darin Seldes Band, performances by



Team Kirra at Atlanta's All Aboard for a Cure

Abyss, the HBO Crew, and the Gwinnett Ballet Theater, and food, a raffle, a silent auction, games and activities for kids of all ages and abilities! In all, over \$50,000 was raised this year for the United

Mitochondrial Disease Foundation, over \$150,000 raised in its first three years! Next year's event has already been announced as the City of Norcross is once again gracious enough to host the event for us on April 30, 2011 - hopefully with much better weather!

- **May 13, 2010.** The Atlanta Chapter hosted Mitochondrial Disease Awareness Night at Mutual Stadium, home of the Rome Braves. Thanks to everyone who attended and helped spread awareness about mitochondrial disease.

### CHICAGO CHAPTER

- **May 21, 2010.** The 10th Annual and final Kites for Kristen event in honor of Kristen Charleston was held at the European Chalet in Chicago, IL. The event benefited the Kristen Charleston Research Fund, which surpassed \$40,000 with this year's event. Thanks to the Charleston Family for their continued support and dedication over the past ten years.

### MIDDLE TENNESSEE CHAPTER

- **March 27, 2010.** The First Annual Jackson Culley Mito-What?5K was held at USA Stadium in Millington, Tennessee. This event drew over 500 people and raised over \$23,000 for the UMDF. It was a beautiful day and many will be returning for the second annual race next year! Thanks to everyone who helped make this day a success!

### NEW ENGLAND CHAPTER

- **May 23, 2010.** The New England Chapter held the UMDF's first ever "Energy for Life" walk at MIT's Johnson Athletic Center in Cambridge, MA. Despite having only a little over 3 1/2 months to plan, the New England chapter came together and surpassed their goals by raising over \$30,000.00 towards a cure! It was an amazing day of hope, joy, and friendship as we celebrated being part of the cure together. The New England Chapter was so encouraged by the turnout and response that they have already started planning their walk for the Spring of 2011! Hope to see you there!

### OHIO CHAPTER

- **May 8, 2010.** The Wild for a Cure Walkathon was held at the Cleveland Metroparks Zoo in Cleveland, OH. Despite cold and rainy weather over \$95,000 was raised for the UMDF. Thanks to all who helped make this walk so successful!

## OTHER NOTABLE EVENTS (CON'T)

- **March 20 - 21, 2010.** The sun was shining bright, smiles were shining even brighter and the weather was perfect! That was the atmosphere in Virginia Beach on March 20th and 21st during the 5th Annual Preston's March For Energy. Preston's March piggybacks with The Shamrock Sportsfest and raised over \$37,000. Preston's Marchers enjoyed a delicious dinner generously provided by Baker's Crust and proudly displayed bright green performance shirts, sweatshirts and gym bags from HG Apparel. Preston's family continues to be humbled by the love and support they receive from the Yeungling Shamrock Marathon, the runners and community. Through their support, Preston's March has raised over \$120,000 for UMDF. Preston's family has been fortunate enough to form everlasting friendships because of this, many of those with families affected by Mito.

- **April 3, 2010.** The St. Louis Mito Group hosted the seventh annual Mito- What? Family Fun Day at the Shrine of Our Lady of the Snows in Belleville, IL. This year the walk raised more than \$20,000 for the UMDF! A special thank you to the race committee for all your hard work.

- **April 23-24, 2010.** The fifth annual and final Caroline's On My Mind Weekend was held in Spartanburg, SC. The event, held in honor of Caroline Pulliam, went out with a bang, raising over \$160,000, reaching their goal of raising \$1 million. Thanks to everyone involved for their support!

- **April 25, 2010.** Congratulations to Emilie Pazdan of Greenville, SC who participated in the Big Sur Marathon in honor of Alex Newton. Thanks to generous donations, over \$1,000 was donated to the UMDF.

- **May 1, 2010.** The First Annual Brittany

Wilkinson Co-Ed Softball Tournament was held at Sierra Bicentennial Park in Clovis, CA. Thanks to all the teams who participated in this event, benefitting the Brittany Wilkinson Research Fund. Thanks to the Wilkinson Family for their continued support!

- **May 8, 2010.** Over 300 members of the Western PA Mito Group attended Mitochondrial Disease Awareness Night at PNC Park, home of the Pittsburgh Pirates. Thanks to all the families who attended.
- **May 13, 2010.** The Memphis Area Mito Group hosted Mitochondrial Disease Awareness Night at AutoZone Park in Memphis, TN, home of the Memphis Redbirds. Thanks to all the families who attended.
- **May 25, 2010.** The Idaho MitoGroup held a Fun'Draising Night at Red Robin in Boise, Idaho. Thanks to everyone who came out!

## GIFTS FROM THE HEART

- Valley Bowling in Youngstown, PA hosted a fundraiser in honor of Cooper Groves that raised \$3484.25 through a bowling tournament, 50/50 drawing, raffle and a Chinese auction.
- Michelle Meddin of Crooksville, OH held an energy band fundraiser at North Gwinnett Middle School in honor of her niece which raised over \$1,000. Thank you for all your support!
- St. Bernadette's School in Monroeville, PA participated in the Coins for a Cure program in memory of Gina Mohan. Over \$1,000 was donated to the UMDF.
- The second Annual Crop for a Cause was held on April 25th in Cumming, Georgia. A total of \$2,139.46 was raised for the UMDF. Special thanks to Robyn Gravitt for organizing this successful event!
- A bake sale in honor of Abby Kasuba of Wheaton, IL, raised \$400. Thanks to all of those who participated.
- Activision / Blizzard in Santa Monica, CA held an internal fundraiser totaling \$2,325.00. Thanks to all the employees who contributed.
- Bill Goade, CEO of Cresa Partners in Boston, MA, raised \$2,600 through an office fundraiser on behalf of his friend's son who is affected. Thanks to all who participated.
- Gulf Coast Bank in New Orleans held a fundraiser in honor of JD Blue, raising over \$1,100. Thanks to all who participated.
- Jim Lomeo held a fundraiser in Pittsburgh, PA benefitting the UMDF. It raised \$1,015. Thanks to Jim and everyone who attended!
- Joey's Annual Spaghetti Dinner Fundraiser took place in honor of Joey Ricci of Mogadore, OH. This fundraiser benefitted Joey's Wild for a Cure team. Thanks to the Ricci Family for their continued support!
- Kathleen Liao of Princeton Junction, NJ hosted an Art Exhibit in honor of Sarah Ward, where she donated ten percent of her art sales to the UMDF. Thank you, Kathleen!
- Midland Middle School in Columbus, GA held a fundraiser in honor of Ava Wilson, raising \$525. Thanks to all the students and faculty at Midland Middle School.
- Mrs. Burcham's 2nd Grade Class at Jeter Elementary School in Millington, TN conducted fundraiser that raised \$563 in honor of Jackson Culley. Thanks to all who participated.
- The annual Hefferon Recycling Party was held in McLean, VA and raised over \$6,100 this year. Thanks to the Hefferon Family and their family and friends for their continued support.
- The Candelario Family of Lorain, OH, hosted Vanessa's Vision Hope For A Cure Dinner, in honor of Vanessa Candelario. The proceeds of this fundraiser benefitted Team Vanessa in the Wild for a Cure Walkathon. Thanks to the Candelario Family for their continued support!
- The Gleannloch Farms Spring Market Show in honor of Curtis Jackson was held in Spring, TX and helped raised \$1,300 for the UMDF. Thanks to everyone who attended!
- The Kobunski Family of North Royalton, OH, hosted the Guest Bartender Evening at 82nd Street Grill & Pub, which helped raise funds in honor of Kyle Kobunski's Wild for a Cure Team. Thanks to all the guest bartenders and those who came out to support this event.
- The Precision Roof Golf Classic was held in Roswell, GA and raised \$5,175 in honor of Joseph Atchley II. Thanks to all who participated.
- The students at Fallen Timbers Middle School in Whitehouse, OH, donated \$110 to the UMDF as part of a larger fundraiser. Thanks to all the students and faculty at Fallen Timbers Middle School.
- Vicki Sendelbach of Englewood, OH, ran in the Sixth Annual Another Dam 50K in honor of Alex Sendelbach. Thanks to Vicki for taking on such a monumental task in support of Alex and the UMDF!
- William Laczko's 1st Birthday Party was held in Cuyahoga Falls, OH and yielded over \$450 in donations in honor of his cousin, Caroline Lyman. Thanks to William's friends and family for their support.
- St. Columbille School in Parma, OH raised over \$775 in a Coins for a Cure campaign in honor of the Dylan Kollin family. Thanks to all who supported their efforts.
- The Southern Ohio Bikers held a Valentine's Day dance in honor of Faith and Ayden Hingsbergen, raising \$1,400. Thanks so much to all who attended.
- Jerica Bennett of Ohio coordinated a "Crazy Sock" day at her school, Morgan West Elementary. The students made a donation to wear "Crazy Socks" to school in honor of Baylee Thompson. The students raised \$117 and presented it to the Thompson family at their event, Bet on Baylee.
- Sydney Breslow, a junior at Delaware Valley Friends School in Cherry Hill, NJ, led a mitochondrial disease awareness campaign, advocacy letter writing campaign, and a Coins for a Cure drive, raising \$1,214!! Thanks to Sydney and all of the student and faculty at Delaware Valley Friends School!
- Thanks to Morristown Elementary School in Shellyville, IN, for their Coins for a cure Drive in honor of Drake Cornn, raising \$44!
- Thank you to Michael Belcher of Blue Bell, PA, for holding a NCAA Tournament fundraiser called "Brackets for Mito" where each participant paid a \$10 entry fee. Michael and his friends raised \$200 for the UMDF!
- Pam Barmash completed the Boston Marathon in honor of Mallorie Cantwell. A total of over \$1,000 was donated to the UMDF. Way to go Pam!
- The second annual four hour spin-a-thon was held in honor of Bobby Arnold in Cleveland, Ohio. Over \$3,000 was raised for the UMDF to find a treatment and cure for mitochondrial disease. Participants received a great workout and got to enjoy food and music while helping a great cause.
- Robert Burgess of Fon Du Lac, WI, hosted a bowling fundraiser in honor of Kennedy Burgess that raised over \$1,500 for the UMDF. Thank you for your support!
- Angela & Basil Louvaris were married in Boardman, OH. They asked wedding guests to donate to the UMDF in their honor. Their generosity helped raise \$515. Thanks to Angela, Basil, and their wedding guests!
- Hoops for Hope, a 3x3 basketball tournament and luncheon, was held in Collinsville, OK in honor of Isabella Magee. Thanks to all who played, watched and organized!
- The Kaitlin Vasilich Hope Fund 7th Annual Fun Walk was held in Pittsburgh, PA in memory of Kaitlin Vasilich. Thanks to everyone who came out to the walk.
- The Mount Leadership Society Scholars held a dodgeball tournament at Ohio State University. Proceeds were donated to UMDF. Thanks so much to all that were involved!

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## MITO FACTS

- When mitochondria fail, less and less energy is generated within the cell, and cellular injury and even death follow.
- Adult onset of mitochondrial disease can result in drastic changes from active lifestyle to debilitating illness in a short amount of time.
- A child with a mitochondrial disease is at highest risk for neurological and organ damage during and two weeks after an illness.

## A MOTHER'S VENTURES

### *A Day in the Life of Sean & Susan Freiburger*

Since our son, Sean, was diagnosed with a probable mitochondrial disorder two years ago, we wanted to get involved with the UMDF. We live in Pittsburgh, so the UMDF is right in our backyard and we are so grateful for all that the UMDF does to educate, advocate, and advance research for mitochondrial disease patients. In the last couple of weeks, we've had the opportunity to participate in several activities with the UMDF, including filming a video, participating in a meeting about the mitochondrial disease research registry, and participating in the grant review allocation meeting.

considering to create this registry. The UMDF is carefully considering the costs and benefits of purchasing such a program. They are getting input from researchers and clinicians as well as families to make sure that if they build a registry, it will contain the elements needed to make it work.

I was also given the opportunity to attend the UMDF Grant Review luncheon and allocation meeting as an observer. There were 9 reviewers



**Sean Freiburger**

and 2 co-chairs participating in the grant review as well as UMDF's CFO and grants coordinator. I was very impressed with the depth, diversity, caliber, and credentials of the grant review committee. While I was not privy to the details of the grant applications due to confidentiality, it was clear that there were many strong grant proposals and that

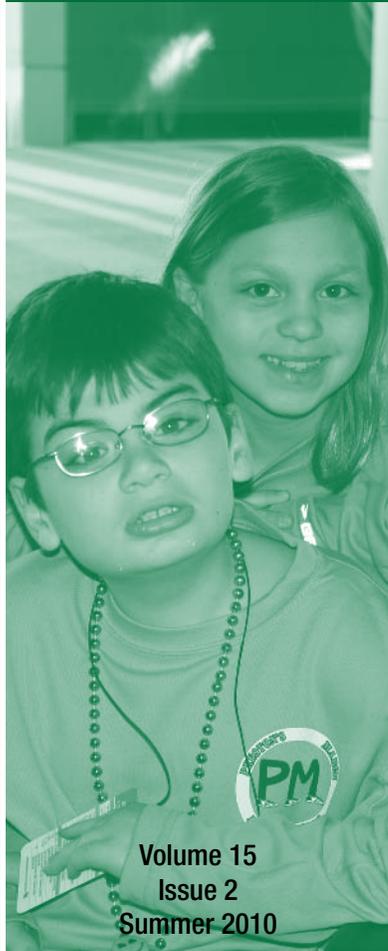
the review process was professional and rigorous. The reviewers were thoughtful in their allocation process, as there were more grants that they wanted to fund than money available. There were some excellent grants submitted that the review committee ranked highly, but unfortunately, there were not enough research funds available for the UMDF to fund them all. In order to fund as many grants as possible, the committee considered options such as shortening the term of the grant, cutting line items in the grant, and whether there were potentially other sources of grant money available to co-fund the grants. The grants committee was also looking at funding clinical as well as research programs. This process allowed the UMDF to fund the top three rated grants in one way or another. Being part of this process showed me the quality of the research that is being pursued to help find a cure for mitochondrial diseases. It also reinforced to me the importance of raising more money to fund this research. What if the research proposal that was left unfunded is the key to finding a cure? We all need to help the UMDF raise awareness, advocacy, and money to help find a cure.



**Sean and Susan at the Pirates Video Taping**

Sean was included in the production of a short awareness video that was shown on the Jumbotron at the UMDF night at a Pittsburgh Pirates game. He was so excited about going to the game and being able to see himself on the Jumbotron. At this filming, we met Tyler Liebegott who is a youth ambassador for UMDF. His story is inspirational and he took the time to talk to Sean about how tough it can be to take all the medicines, but how important they are so he can stay healthy and strong. I was grateful for the opportunity to meet other local mitochondrial disease families, to talk with Chuck Mohan and the rest of the dedicated UMDF staff, and to get a tour of the UMDF office.

Chuck Mohan and I spoke about a project that the UMDF is working on – the creation of a research registry for mitochondrial disease patients. This registry has the potential to connect researchers with patients that meet the criteria for upcoming clinical trials and research studies. It is so important for mitochondrial disease patients to consider participating in these studies to help advance research and get us all closer to a cure. I was invited to attend a demo of a program that the UMDF is



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## FROM THE CHAIRMAN

In just a few short days, the world's best mitochondrial medicine experts, affected adults and parents of affected children will gather for what is considered to be one of the top meetings for mitochondrial medicine in the world. The UMDF Symposium – Mitochondrial Medicine 2010, begins with sessions for the scientific and medical community on June 16th and continues with sessions for affected individuals and families on June 18th and 19th. This year, we will be the guests of the beautiful JW Marriott Camelback Resort in Scottsdale, AZ.



Typically, the symposium enables patients and their families to learn the latest information available about the many different forms of mitochondrial disease. It is also the place where many scientists, clinicians and medical experts gather to share information and ideas. When we held our first symposium 12 years ago, I would never have imagined that our symposium would have evolved into what it is today. I have attended 11 of the past 12 meetings. Each and every time, I come away with increased knowledge about mitochondrial disease. I hope you have had the same experience.

This year, we are actually able to present information about current clinical trials and other very important information that is being offered. If you read the last issue of this newsletter, you probably saw the information about the Mayo Clinic Mitochondrial Disease Bio Bank. The Bio Bank will be on site, collecting blood samples from interested participants. The samples will be used to aid researchers who are looking for a cure. In addition to that, the North American Mitochondrial Disease Consortium will staff an information table that will provide

details on their plans to begin a patient registry. Those are just a few of the organizations on hand to provide information and to gather patients for current and potential clinical trials.

I am always excited about the presentation of the Scientific and Medical Research Grants and our awards for volunteers. For 2010, the UMDF will fund over \$300,000 in research projects. This year, we had more than 175 inquiries about the funding but just fewer than 40 were asked to submit full grant proposals. The grant review committee recently spent two days in Pittsburgh, PA reviewing the proposals and, based on their criteria, selected three projects for funding this year. I encourage you to read about the 2010 research grant recipients in this newsletter. They will be recognized during our symposium banquet on June 18, 2010.

I also look forward to handing out UMDF awards to volunteers. Without all of you, none of this would be possible. I have been told we have a record number of nominations for our LEAP and Heartstrings Awards this year. Deciding the winner will be very difficult, but you will learn about them in our next newsletter or online at [www.umdff.org](http://www.umdff.org) after the symposium.

If you are coming to Scottsdale, I look forward to seeing you there. If you can't make it this year, consider attending our 2011 symposium in Chicago.

Energy to all,



W. Dan Wright, UMDF Chairman

## ENERGY FOR LIFE WALKATHONS

The New England Chapter of the United Mitochondrial Disease Foundation has done something no other chapter has done --- they held the first ever 'Energy for Life' Walkathon. The walk, which drew more than 200 participants, has raised more than \$30,000 to date.

Planning an 'Energy for Life' Walkathon typically begins more than eight months from the actual walk date, but the New England Chapter pulled the walkathon together in four months. The credit for the success of the walk goes to co-chairs Stefani Bush and Lindsay Knopps and their team of volunteers. In addition to securing the teams, they all worked very hard to coordinate the location, which was at the Massachusetts Institute of Technology (M.I.T.) Johnson Athletic Field. They also secured the sponsorship of M.I.T., Senator John Kerry, and Milestones.

"This walk was successful because of the support and attention given by the volunteer co-chairs, Stefani Bush and Lindsay Knopps," said Carol Milsovic, the UMDF's National Events Manager. "Stefani came to the Energy for Life Walkathon training in Pittsburgh back in January,

and look what she and her team did in just four short months." Milsovic thinks the New England Chapter Walkathon will only continue to get larger each year.



Energy for Life - New England's starting line

The fall of 2010 is shaping up to be a busy season for 'Energy for Life' Walkathons. Organizers in Minneapolis are busy planning their walkathon at Normandale Park on September 11th. Also scheduled on that day is a walkathon in the Buffalo area at Island Park. On September 18th, volunteers with our Carolina Foothills Chapter have scheduled an 'Energy for Life' Walkathon at Freedom Park in Charlotte. Our Chicago Chapter is also planning a walk that will be held in Saint Charles, IL., on September 19th. That is also the day that our Kansas City Chapter will hold their 'Energy for Life' Walkathon at T-Bones. 'Energy for Life' Walkathons in September wrap up with the DeVal Chapter walk at Campbell's Field Stadium in Philadelphia and with a walk scheduled by our Erie Mito Group, both on September 25th.

"Because we want the walks to be successful, we like to start

*(continued on page 16)*

**UNITED MITOCHONDRIAL DISEASE FOUNDATION**

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**SUPPORT UMDF THROUGH GIFT CARDS AND LOYALTY POINTS**

You can now donate funds to The United Mitochondrial Disease Foundation (UMDF) by using Charity Choice Gift Cards, a national giving program that enables purchasers and redeemers of charity gift cards to designate UMDF to receive the value of the card. Charity Choice ([www.mycharitypoints.org](http://www.mycharitypoints.org)) also enables you to convert your credit card points and employee rewards into a cash donation to UMDF.

Hundreds of companies across the nation use the Charity Choice program to enable their customers to convert their loyalty points and rewards into a donation to their favorite charity—and now you can designate UMDF to receive the funds.

UMDF's special code for the Charity Choice program is 437 which can be used to automatically pre-select UMDF as the

“featured” charity, especially when ordering charity gift cards. When giving gift cards using that code, you will designate that UMDF will receive up to 75 percent of the value of the card when it is redeemed by the recipient. You can suggest that the recipient of the gift card can designate the remainder to UMDF or select up to three other charities from a list of charitable organizations to get the remaining 25 percent of the card's value.

You can find UMDF listed as a potential beneficiary of your points and rewards on the Charity Choice web site under the “Local and Additional National Charities” section of the “List of Over 100 Charities” tab.

Go to [www.mycharitypoints.org](http://www.mycharitypoints.org) for unique charity gift-giving opportunities to benefit UMDF for birthday, business, holiday, sympathy, or wedding gift ideas.

*Following is a sampling of the companies listed on the Charity Choice site that allow credit card points and rewards to be donated to UMDF.*

- |  |                         |                                       |
|--|-------------------------|---------------------------------------|
| AAA                                      | Congoleum               | Mitsubishi                            |
| Alegent Health                           | Continental Airlines    | Monsanto                              |
| American Airlines                        | Cooper Tire             | Motorola                              |
| AmeriHealth                              | Deloitte                | Nationwide Better Health              |
| Anheuser-Busch Companies                 | DuPont                  | Nissan                                |
| ARAMARK                                  | Ernst & Young           | Nokia                                 |
| Autodesk                                 | Education Management    | Olympus                               |
| Avery Dennison                           | Energizer Holdings Inc. | Panasonic                             |
| Blue Cross Blue Shield of Florida        | ESPN                    | Physicians Plus Insurance Corporation |
| Blue Cross Blue Shield of Michigan       | Federal Reserve Bank    | PNC Merchant Services                 |
| Blue Cross Blue Shield of North Carolina | Fifth Third Bank        | Providence Health & Services          |
| Blue Cross Blue Shield of South Carolina | General Motors          | Resolution Health                     |
| Boston Scientific                        | Gestetner               | Ricoh                                 |
| Bowe Bell + Howell                       | GN Netcom               | Safeway                               |
| BP                                       | Graybar                 | SC Johnson                            |
| Bridgestone/Firestone                    | Guaranty Bank           | Scott & White                         |
| British Airways                          | HBO                     | Seagate                               |
| Broadridge Financial                     | Health Net              | Sharp                                 |
| Capital One                              | IBM                     | Siemens                               |
| Carlton-Bates                            | Independence Blue Cross | Sony                                  |
| Charter Communications                   | Juniper Bank            | The Container Store                   |
| CIBA Vision                              | Kaiser Permanente       | Toshiba                               |
| ConAgra                                  | Kellogg                 | WCI Communities, Inc.                 |
|  | L-3 Communications      | WebMD                                 |
|  | LaSalle Bank            | WellPoint                             |
|  | Merck                   | Wells Fargo                           |
|  | Microsoft               |                                       |



*Matthew Slauter of Orlando, Florida*

## CLINICAL TRIALS

**Stanford University is seeking volunteers with MELAS for a seasonal flu vaccine study.** The Stanford-LPCH Vaccine Program is seeking healthy kids and adults to participate in an influenza (commonly known as “the flu”) vaccine research study to better understand effects of immunizations for people with mitochondrial disorder. We will enroll 15 healthy male and female adult participants (18-50 years old) along with 15 otherwise healthy children and adults (13-50 years old) with a metabolic disorder called MELAS syndrome.

The flu vaccine used in this study is licensed by the FDA and not experimental. This is the same seasonal flu vaccine used for the public during the 2010-2011 flu season. The research will be conducted at Stanford University by Dr. Gregory Enns, Associate Professor of Pediatrics and the Director of the Biochemical Genetics Program and Dr. Cornelia L. Dekker, Professor of Pediatrics at Stanford.

The purpose of this study is to compare specific metabolic responses and immune biomarkers before and after the administration of the seasonal flu vaccine. Preliminary studies show those with mitochondrial disorders may respond differently as compared to healthy participants. The information collected by this study will help us better understand the safety and efficacy of influenza vaccination in volunteers with mitochondrial disease.

To qualify you should be:

- An adolescent or adult volunteer 13-50 years of age with a diagnosis of a mitochondrial disorder known as MELAS, or a healthy volunteer 18-50 years of age.
- Able to attend 4 clinic visits at Stanford within a one month period (September 2010- January 2011)
- Able to provide 4 blood samples

Website: [http://vaccinesvaccines.stanford.edu/clinical\\_trials.html](http://vaccinesvaccines.stanford.edu/clinical_trials.html)

Contact: Stanford-LPCH Vaccine Program at (650) 498-7284

Email: [vaccines\\_program@stanford.edu](mailto:vaccines_program@stanford.edu)

*This study is funded by the National Institutes of Health*

**The University of Florida** is continuing to recruit patients for a clinical trial to investigate the safety and effectiveness of Coenzyme Q10 (CoQ10) as a treatment for children with specific mitochondrial diseases. CoQ10 is a mitochondrial cofactor and antioxidant in the process that cells use to convert food and oxygen into energy. To be considered for the trial, patients must be 12 months to 17 years of age and have a biochemical or molecular diagnosis of a deficiency of complex I, III or IV of the respiratory chain. For additional information contact Tracie Kurtz, RN, at [Tracie.Kurtz@medicine.ufl.edu](mailto:Tracie.Kurtz@medicine.ufl.edu) or 352-273-9016.

**Bascom Palmer Eye Institute at the University of Miami Health Center** is recruiting LHON patients. The major aim of this study is to identify and clinically characterize a group of patients and carriers with LHON due to G11778A mutation in preparation for a future gene therapy study. The intent is to eventually choose subjects for the future phase 1 or phase 2 of the gene therapy study from the participants in this preparatory phase of the study. Study doctors will be happy to answer any questions that you may have regarding this study plan. You may call Byron L. Lam M.D. 305-326-6021 or Alexis Morante, study coordinator, at 305-202-4731 to have any questions regarding this study answered.

**Baylor College of Medicine and Texas Children's Hospital** are recruiting subjects with MELAS (mitochondrial myopathy, encephalomyopathy, lactic acidosis, and stroke-like episodes) syndrome for a clinical study. The purpose of this study is to measure nitric oxide in subjects with MELAS syndrome and to see if giving arginine or citrulline will increase the formation of nitric oxide. Nitric oxide is thought to be helpful in improving and preventing strokes. Therefore, if arginine and/or citrulline are shown to increase the formation of nitric oxide, they could be used to prevent and improve the strokes in patients with MELAS syndrome. Adults or children affected with MELAS syndrome and carrying the A3243G mutation can

participate. Adults without MELAS disease will be recruited to participate as control subjects. Subjects with MELAS will be admitted twice to the General Clinical Research Center (GCRC) at Texas Children's Hospital and nitric oxide production will be measured by stable isotopes infusion which is a safe procedure. The principal investigator is Dr. Fernando Scaglia. Subjects interested in participation or getting more information can contact the co-investigator Dr. Ayman El-Hattab at email: [elhattab@bcm.edu](mailto:elhattab@bcm.edu), phone: 832-822-4289 or pager: 832-824-7243 (5523).

**The University of Texas Southwestern Medical Center** is recruiting patients for this research study. Adult or pediatric patients afflicted by the mitochondrial disease MELAS (mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes) associated with mitochondrial DNA mutation A3243G will be subject to physical examination, bloodwork and a type of MRI named 7T MRS (magnetic resonance spectroscopy), performed on the leg muscle, to understand how muscle MRS results correlate with disease severity and mutation abundance in blood. Patients with any type of metal implants in the body or unable to receive an MRI are not eligible to participate. There is no treatment or other intervention associated with this study at this time. Patient participation will last less than 3 hours for each visit, for a maximum of three visits. Patients interested in participating or in obtaining more information should contact the principal investigator Juan M. Pascual, MD, PhD at [Juan.Pascual@UTSouthwestern.edu](mailto:Juan.Pascual@UTSouthwestern.edu) or call us at 214-456-2768

**Columbia University in New York City** is seeking study participants for a double-blind, placebo controlled clinical trial of idebenone in MELAS (Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like Episodes). The Phase IIa study will compare two different doses of an experimental medication, idebenone, administered over a one month period to determine the efficacy of the drug. People with MELAS and the 3243 mutation, aged 8-65 years, may be eligible. The main goal of the clinical trial is to determine if idebenone has an effect on brain lactate as measured by magnetic resonance spectroscopy (MRS). MRS is done in an MRI scanner, and is safe and typically well tolerated. An additional goal is to study the safety and tolerability of idebenone in people with MELAS. If you or a family member would like more information regarding this study please see our website (<http://giblinlab.org/studies.html>) or contact the Research Coordinator, Kris Engelstad, at 212-305-6834.

**Effects of Exercise on Mitochondrial Function and Quality of Life in Patients with Mitochondrial Myopathies due to Mitochondrial DNA Mutations.** University of Texas Southwestern Medical Center is recruiting patients for this NIH-supported study. We are attempting to define the effects of regular exercise training and the consequences of a lack of regular exercise on mitochondrial function over 2 years of observation. We are recruiting patients who have defined mitochondrial DNA defects to undergo detailed muscle and heart evaluation and then will undergo supervised exercise training with repeat detailed evaluation at three additional time points over the 2 years of study. Patients interested in participating or in obtaining more information should contact the research coordinator, Marta Newby, R.N. at 214-345-4655 or email [MartaNewby@texashealth.org](mailto:MartaNewby@texashealth.org)

**University of Texas Height and Weight Data Collection for Mitochondrial Patients.** If you have a mitochondrial disorder or are the parent of someone with a mitochondrial disorder, Dr. Mary Kay Koenig at the University of Texas Houston Mitochondrial Clinic is seeking information on heights and weights of persons with mitochondrial disorders. For more information on this study, email [ut.mito@uth.tmc.edu](mailto:ut.mito@uth.tmc.edu) or call 713.500.7164

**Metabolic Consequences of Primary Mitochondrial Disease.** This is an NIH-sponsored study at the Children's Hospital of Philadelphia designed to primarily evaluate previously collected muscle, skin and blood from individuals with definite mitochondrial disease. For more information on this study, call 267-426-9650.

## PLANNED GIVING OPTIONS SUPPORT UMDF AND HELP YOU TAKE CHARGE

The UMDF is pleased to continue this series of articles on helping you take charge of your finances. Here are a number of ways that a planned gift and careful estate planning can help you and UMDF.

- Planned gifts offer many financial advantages,
- Reduce estate taxes,
- Provide a life income stream,
- Enable you to make a much larger gift than you thought possible,
- Receive a current income tax deduction,
- Reduce or avoid capital gains tax, and
- Support the vital work of the UMDF.

### Wills

One of the most common ways to remember the UMDF and help us carry on our programs is to leave a bequest through your will. The following is suggested language to use in wills and a variety of other estate planning:

Making a gift to the United Mitochondrial Disease Foundation, a 501 (c)(3) nonprofit organization incorporated in the Commonwealth of Pennsylvania "I give and bequeath the sum of \$\_\_\_\_\_ (or \_\_\_\_\_% of my estate) to the United Mitochondrial Disease Foundation, 8085 Saltsburg Road, Suite 201, Pittsburgh, PA 15239 to be used for general purposes."

You may also give a particular asset ("\_\_\_ shares of XYZ stock...") or a portion of the remainder of your estate after other bequests have been paid ("50% of the rest, residue and remainder of my estate").

### Trusts

There are many different types of trusts that can serve a variety of purposes. The advice of an attorney and qualified financial planner is necessary to assess your situation and decide which trust might best serve your goals. Please know, however, that it is easy to include a gift to UMDF through your trust by using the language as shown above.

- Charitable Remainder Trusts permit you to transfer highly appreciated, low yielding assets to an irrevocable trust managed by a bank or other trustee you may select. The trust pays you fixed or variable income of at least five percent for life or a period of years then passes the principal to UMDF. Such gifts avoid capital gains tax, reduce estate taxes and allow the immediate sale and diversification of the gifted assets.
- Charitable Lead Trusts are the opposite of Charitable Remainder Trusts. Instead of having income payments directed to the donor or other beneficiaries, payments are made to UMDF for a specified number of years. After the term expires, the remaining principal is distributed to the beneficiaries designated by the donor. The Charitable Lead Trust is a popular tool for supporting charity, reducing estate taxes and passing assets to children or grandchildren.

### Life Insurance

Life insurance can be a valuable tool in estate planning. By naming beneficiaries on policies, the proceeds can be paid directly to that person or organization without having to go through the probate process. Another option is to make the proceeds payable to a person's estate so it can be included in the total when specific or percentage bequests are made.

Life insurance also offers a wonderful way to make a charitable gift. It is possible to make gifts with "paid up" policies, policies with premiums still due, policies where you can retain the right to a policy's cash value, or by assigning the dividends in a participating policy. Check with your agent to see which option would be best for you.

### Gift Annuities

The UMDF can offer you a charitable gift annuity program that can provide you with payments for the rest of your life, while ultimately making a gift to support vital programs helping those affected by mitochondrial disease.

### Pay on Death or Transfer on Death Accounts

This estate planning tool can be an effective way to quickly transfer assets,

such as bank accounts, to a beneficiary because it avoids that asset going through the probate process. It also allows you to change the beneficiary at any time.

When establishing the account, tell your banking representative that you wish it to be a "Pay on Death" account (some banks call these Transfer on Death accounts). They will ask you for the name of the person or charitable organization you wish to receive the property upon your death.

### Gifts of Securities

While a gift of securities is not strictly an estate planning tool, there are advantages to this type of donation that have allowed many donors to make gifts that will live on after they are gone.

If you have owned stock for at least one year that has increased in value, you can donate that stock to a charitable organization without having to pay capital gains tax on the increase. Additionally, there is an income tax charitable deduction equal to the full current market value of the securities (up to 30 percent of the donor's adjusted gross income). Using appreciated stock to fund a gift annuity offers added tax benefits to that gift.

NOTE: In order to receive the most favorable tax treatment, you must donate the securities to UMDF – you cannot sell the stock and donate the proceeds. If you would like more information on how to make this transfer, please contact us.

### Retirement Plan Assets

With the increase in the variety of retirement plan assets that people own, an important aspect of your estate planning should be making sure that the money invested in these accounts goes to the people or organizations you wish to receive them.

Examples of these different retirement plans include IRAs, pension plans, Keough and 401 (K) accounts. You should be aware that proceeds from these plans may be taxed up to 70 percent of their value if proper planning is not done. If you are contemplating a charitable gift in your estate plans, using assets such as those in retirement plans can maximize your donation while allowing other property that is not subject to some taxes to be passed to your beneficiaries.

Regardless of the size of your estate, proper estate planning is important to ensure that those assets you have spent a lifetime accumulating are distributed according to your wishes.

By informing us of your donation to include UMDF in your estate plans, we are better able to plan for the future. Of course, once you inform the Foundation we would respect a wish to keep it anonymous.

### How to make a Planned Gift

A trained legal or financial advisor can help you develop a plan for making charitable gifts that complements your current investment and estate plans. He or she can help you determine

- The appropriate gift level based on your circumstances
- The type of gift (bequest, trust, mutual fund, etc.)
- The tax implications of your gift
- How to best structure the gift to benefit you and the United Mitochondrial Disease Foundation

### MITOCHONDRIAL DISEASE FOUNDATION

To discuss your planned gift, call UMDF at 412-793-8077 or contact Nick Nicholson directly at [nnicholson@monteverdegroupp.com](mailto:nnicholson@monteverdegroupp.com). Nick is a senior financial advisor with The Monteverde Group ([www.monteverdegroupp.com](http://www.monteverdegroupp.com)) and is available to provide free, no obligation, expert assistance coupled with complete confidentiality to all UMDF members and potential donors. Even if you currently have a financial advisor, Nick can provide a second opinion on your financial situation just to ensure your financial comfort and stability. You have nothing to lose because the service is free with no obligation. Take charge of your finances today.

## UPCOMING EVENTS

### ATLANTA CHAPTER

- **September 25, 2010.** The fourth annual Music for Megan and Family Fun Fest in honor of Megan Sheridan will be held in Alpharetta, GA. Visit [www.musicformegan.com](http://www.musicformegan.com) for more information.

### CAROLINA FOOTHILLS CHAPTER

- **September 18, 2010.** The Energy for Life Walk: Carolina Foothills will be held at Freedom Park in Charlotte, NC. Please visit [www.energyforlifewalk.com/carolinafoothills](http://www.energyforlifewalk.com/carolinafoothills) for more information.

### CHICAGO CHAPTER

- **September 19, 2010.** The Energy for Life Walk: Chicago will be held at Pottawatomie Park in St. Charles, IL. Please visit [www.energyforlifewalk.org/chicago](http://www.energyforlifewalk.org/chicago) for more information.

### DELAWARE VALLEY CHAPTER

- **August 23, 2010.** Join the Delaware Valley Chapter for Mitochondrial Disease Awareness Night at Campbell's Field. For more information, contact Laurel Smith at [zoeymom1@gmail.com](mailto:zoeymom1@gmail.com) or (609) 714-0091.
- **September 22, 2010.** Join the Delaware Valley Chapter for Mitochondrial Disease Awareness Night at Citizens Bank Park. For accessible seating, please contact Kaitlyn Finley in the Philadelphia Phillies Group Sales Office at 215-463-5000 ext. 5110.
- **September 25, 2010.** The Energy for Life Walk: Del Val will be held at Campbell's Field Stadium in Camden, NJ. Please visit [www.energyforlifewalk.org/delval](http://www.energyforlifewalk.org/delval) for more information.

### HOUSTON CHAPTER

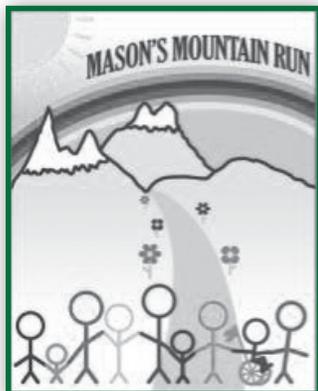
- **October 28, 2010.** The second Gleannloch Farms Spring Market Show in honor of Curtis Jackson will be held in Spring, TX. Visit [www.gleannlochfarmsmarketshow.com](http://www.gleannlochfarmsmarketshow.com) for more information.

### MINNEAPOLIS/ST. PAUL CHAPTER

- **September 11, 2010.** The Energy for Life Walk: Minnesota will be held at Normandale Lake Bandshell in Bloomington, MN. Please visit [www.energyforlifewalk.org/minnesota](http://www.energyforlifewalk.org/minnesota) for more information.

### AROUND THE COUNTRY

- **June 5, 2010.** The second annual Landon's Hope 5k walk and family fun day will be held at Eastman Park in Windsor, CO. Enjoy kids' activities, face painting, food, drinks and much more. Register at [www.umdf.org/landonshope](http://www.umdf.org/landonshope).
- **June 8, 2010.** Millicent Gottesman is hosting a Mito Luncheon in honor of her granddaughter, Rina Goldberg. All proceeds will be donated to the UMDf. Thanks for all your hard work and dedication Millicent!
- **June 12, 2010.** Join us for the first ever Mason's Mountain Run in South Red Lodge, Montana. You can sign up for a challenging 10K trail run or participate in a family fun trail walk. For more information and to register please visit [www.umdf.org/masonsmountainrun](http://www.umdf.org/masonsmountainrun).



- **June 12, 2010.** The Energy for Life Walk: Iowa will be held at the corner of Viking Road & Highway 58 in Cedar Falls, IA. Please visit [www.energyforlifewalk.org/iowa](http://www.energyforlifewalk.org/iowa) for more information.
- **June 19, 2010.** Conner's Concert for a Cure will be held in Rocky Mount, NC in honor of Conner Armstrong. For more information, contact [events@umdf.org](mailto:events@umdf.org).
- **July 31, 2010.** The 4th Of July Fun Run will be held in Kelso, WA. This is organized by Cowlitz Valley Runners. Visit [www.cowlitzvalleyrunners.org](http://www.cowlitzvalleyrunners.org) for more information.
- **July 31, 2010.** The second annual MitoMania will take place at The Farm in Cincinnati, OH. For more information, visit [www.umdf.org/mitomania](http://www.umdf.org/mitomania).
- **August 7, 2010.** The third annual Run4Raley will take place at the Philo Ball Diamond in Philo, IL. For more information, visit [umdf.org/run4rale](http://www.umdf.org/run4rale).
- **August 8, 2010.** The second annual Aubrey's Cut-A-Thon in honor of Aubrey Neely will take place at Harmony Salon in Wexford, PA. Contact Patrice DeShantz for more information by email at [patrice\\_37@hotmail.com](mailto:patrice_37@hotmail.com).
- **August 21, 2010.** The Western PA Mito Group will host the fourth annual Brew at the Zoo at the Pittsburgh Zoo. Visit [www.thezoobrew.com](http://www.thezoobrew.com) for more information.
- **September 11, 2010.** The Energy for Life Walk: Western New York will be held at Island Park in Williamsville, NY. Please visit [www.energyforlifewalk.org/westernnewyork](http://www.energyforlifewalk.org/westernnewyork) for more information.
- **September 18, 2010.** The Savannah's Hope Walk will be held in Des Moines, IA in memory of Savannah Bagby. For more information, contact [events@umdf.org](mailto:events@umdf.org).
- **September 18, 2010.** Race for Rennes will be held in honor of Rennes Thompson in Lake City, FL. For more information, contact [events@umdf.org](mailto:events@umdf.org).
- **September 25, 2010.** The Idaho Mito Group will host the Energy Bowl-a-Thon. Contact [mitogroupofidaho@yahoo.com](mailto:mitogroupofidaho@yahoo.com) for more information.
- **September 25, 2010.** The Energy for Life Walk: Northwestern PA will be held at Sugar Grove AMVETS Post 50 in Sugar Grove, PA. Please visit [www.energyforlifewalk.org/northwesternpa](http://www.energyforlifewalk.org/northwesternpa) for more information.



Aubrey Neely

If you are having, have held a fundraising event or are in need of assistance, we want to talk to you!  
Email [events@umdf.org](mailto:events@umdf.org) or call 888-317-UMDF.

## 2010 UMDF RESEARCH GRANT RECIPIENTS

- **\$130,348 Cornelius Boerkoel, MD, PhD** - University of British Columbia, Vancouver

*“Spinocerebellar ataxia with axonal neuropathy: defining the mitochondrial component”*

Spinocerebellar ataxia with axonal neuropathy (SCAN1) is an inherited degenerative disorder of the nervous system. Affected persons are wheelchair bound by early adulthood but retain normal cognitive function. We have shown that SCAN1 is caused by mutation of the DNA repair enzyme tyrosyl-DNA phosphodiesterase 1 (Tdp1). Nonetheless, how this mutation causes disease has eluded delineation. So far studies have focused on the role of Tdp1 in the nucleus; however, Tdp1 is highly expressed in the cytoplasm of neurons that die in SCAN1. We have now found that Tdp1 is in the mitochondria of cells and that Tdp1 deficient mice develop blindness from retinal degeneration, a common complication of mitochondrial dysfunction. Therefore, we hypothesize that Tdp1 maintains the mitochondrial genome and that SCAN1 is a mitochondrial disorder arising from damage to the mitochondrial genome.

To investigate this, we will pursue 4 objectives. First, we will characterize the function of Tdp1 in the mitochondria. Second, we will characterize problems of the Tdp1-deficient mouse eyes and the relationship of these problems to poor mitochondrial function. Third, we will characterize the effects of antioxidants on improving mitochondrial function in the eyes of mice deficient for Tdp1. Fourth, we will see if Tdp1 inhibitors affect mitochondrial function. In summary, delineating the function of Tdp1 in the mitochondria will identify a new mechanism for maintenance of the mitochondrial genome and show whether SCAN1 is a mitochondrial disease. The pathophysiology of SCAN1 is important as a paradigm for understanding the neurodegeneration that results from defective DNA repair.

**Lay Description:** He will investigate the role played by a mutated mitochondrial DNA-repair enzyme in the development of an inherited disease that causes progressive loss of coordination and mobility in humans. The project will also assess the efficacy of antioxidant therapy in reversing the effects of the enzyme mutation.

- **\$110,000 Robert Jensen, PhD** - Johns Hopkins School of Medicine, Baltimore, MD

*“DCMA and Barth syndromes—similar diseases caused by defects in mitochondrial protein import?”*

DCMA disease is a mitochondrial disorder characterized by early-onset heart problems (dilated cardiomyopathy), growth retardation, and uncoordinated movement (ataxia). Strikingly, DCMA patients have virtually identical problems to those with Barth Syndrome (BTHS), caused by defects in the cardiolipin-remodeling enzyme, Tafazzin. We find that DCMA patients carry mutations in a gene encoding a protein homologous to a subunit of the yeast TIM23 protein import machinery. The focus of this proposal is to determine how a defect in protein import and a defect in cardiolipin metabolism can cause virtually the same disease. In our first aim, we propose to identify the cellular defect in common with DCMA and BTHS. Using several different strategies, we will ask if cells or mitochondria isolated from DCMA and BTHS patients show similar defects in the import of proteins into their mitochondria. In addition, several observations suggest that abnormal mitochondrial metabolism may underlie both DCMA and Barth diseases. In our second aim, we will examine the composition of cultured cells from patients to determine if the level of one or more metabolites is altered in DCMA and BTHS. We will also probe how a defect in mitochondrial protein import and cardiolipin deficiency impacts cellular metabolism.

**Lay Description:** He will compare two different conditions marked by similar types of cardiac dysfunction resulting from abnormal mitochondrial metabolism. Discovering the cellular defects that the two diseases have in common will provide important insights into metabolic impairments that may be common to a number of mitochondrial diseases.

- **\$75,000 Ingrid Tein, MD** - Hospital for Sick Children, Toronto, Canada

*“Pilot study to investigate the efficacy of L-arginine therapy on endothelium-dependent vasodilation & mitochondrial metabolism in MELAS syndrome”*

MELAS patients suffer from exercise intolerance, weakness, poor vision or blindness, poor growth, developmental delay, and deafness. They also have unique ‘stroke-like’ episodes (SLEs) which are not due to blockages of large or medium arteries. These ‘strokes’ are thought to be due to energy failure of very small brain blood vessels combined with energy failure in the mitochondria (cell battery) of the brain cells, especially in the back region of the brain in the vision centre. This leads to visual loss and paralysis. The overall goal of this grant is to better understand the mechanism of these SLEs at the level of the brain cells and small blood vessels. To do this, we will study a family of 4 siblings, each with different severities of MELAS, using safe, non-invasive tests. We will determine whether there is a decrease in the ability of small brain blood vessels to increase blood flow by dilating in response to certain strong stimuli such as increased blood carbon dioxide levels or in response to brain cell activation in the vision centre by strong visual stimuli. We will use a technique called BOLD-fMRI which can detect changes in brain blood flow. As exercising muscle also depends on increased blood flow and mitochondrial energy, we will study different measures of aerobic energy metabolism in exercising muscle using cycle exercise testing and special phosphorus-magnetic resonance spectroscopy which measures the changes in the major chemicals of muscle energy metabolism. The dietary amino acid L-arginine is known to dilate blood vessels increasing blood flow and to decrease toxic free radicals that are generated by dysfunctional mitochondria. We will determine the effect of a single dose and a 6 week trial of L-arginine, on brain blood vessel reactivity, brain cell activation and muscle aerobic function to see how useful this would be in the treatment of these patients and other mitochondrial disorders which present with strokes.

**Lay Description:** She will investigate the underlying vascular pathology of the stroke-like episodes experienced by individuals with the mitochondrial disease known as MELAS. Using non-invasive imaging, she will be able to detect impaired blood flow to specific brain regions and to determine whether blood flow improves with oral doses of the amino acid L-arginine.

## ADVOCACY IN ACTION

### UMDF MEMBERS RALLY TO SECURE CO-SPONSORSHIP

As this newsletter goes to print, the UMDF is happy to report that we are almost half way towards our goal of securing the co-sponsorship of 100 members of the U.S. House of Representatives for H.R. 3502. Currently, 45 members have signed onto the bill. The companion bill in the senate, S.2858, carries seven cosponsors. It is because of the direct contact of UMDF members and supporters nationwide that our elected officials are agreeing to add their names to this measure. We are highlighting some of the efforts to serve as an inspiration for all to help us gain support for these very important pieces of legislation.

In February, UMDF Ohio Chapter (North East) member Debra Fleming scheduled a meeting with her congressman Dennis Kucinich (D-10th). Debra participated in the UMDF's 'Call to Action' week which urged our members

to arrange meetings, send letters or make a phone call seeking co-sponsorship of H.R. 3502 and S.2858 while Congress was in recess. Fleming educated the congressman about mitochondrial disease and her struggles as an affected adult. "While



*Debra Fleming with Dennis Kucinich*

I sat with him at his desk, he phoned Washington, D.C., and told his office to sign him on as a co-sponsor of H.R. 3502," Deb told the UMDF. Congressman Kucinich joined the list of those who are supporting the bill. He also directed a staff member to attend the "Run Wild for a Cure" event held by the chapter at the Cleveland Zoo on May 8, 2010.

In southern California, Beth Andrews has been very busy lining up meetings and gathering co-sponsors for the legislation. Beth participates in the Mitochondrial Medicine Guild at Children's Hospital in Los Angeles. She and her network of friends and family members have managed to secure co-sponsorship for H.R. 3502 from a number of House members. Beth has secured a meeting with Rep. Henry Waxman

(D-30th). Representative Waxman is chairman of the House Energy and Commerce Committee. This meeting is important because the committee has jurisdiction over H.R.3502.

In the San Francisco Bay area, UMDF Youth Ambassador Joe Wise has been instrumental in securing co-sponsorship. Joe, 17, of Menlo Park, recently met with Rep. Jackie Speier (D-12th). Shortly after the meeting, Rep. Speier signed on as a cosponsor. On May 5, 2010, Joe testified before the California Senate Health Committee.

They recommended that the measure be sent to the California Senate for a full vote. On May 10, 2010, the California State Senate approved the measure 32-0. The meetings and approval of SJR 23 came because of Joe's meetings with



*Joe Wise with James Everitt, Principal of Sacred Heart Preparatory School and California State Senator Joseph Simitian*

former Senator and now California Lt. Governor Abel Maldonado, SJR was introduced. SJR 23 is important because it urges President Obama and the Congress of the United States to take action on supporting H.R. 3502 and to amend the federal statutes to support federal research for mitochondrial disease. Joe said approving the measure will send a strong message to Washington, D.C. "Together we can save my life, my brother's life and the more than 17,000 residents who have been diagnosed and the 180,000 California residents who may have this disease and not even know it," Joe testified. Both Joe and his brother Jack have mitochondrial disease.

#### **H.R. 3502 AND S.2858 STATUS**

Over the summer, the UMDF is working to try to secure a hearing for both H.R. 3502 and S. 2858. Congress will adjourn for this session on December 31, 2010. If we are unable to have a hearing and secure passage by that time, we will have to reintroduce both bills in 2011.

## MEMBER RESOURCES

### **SUMMER-TIME TRAVELS**

Where to go this year? Walt Disney World or Land? The beach or the lake? The Grand Canyon or Niagara Falls? Hollywood or New York City? Wherever your summer travel plans take you, there are some important things you need to consider when traveling with a disabled loved one. Below are a few resources to help ease the stress of travel.

#### **Access-able Travel Source**

Provides "information about: disabled travel, wheelchair travel, disabled holidays, disability travel, handicapped travel, accessible travel, vacations for disabled, mature travel." Website: [www.access-able.com](http://www.access-able.com)

#### **Accessible Journeys**

Accessible wheelchair vacations, tours, equipment rental, travel tips, networking, and more. Website: [www.disabilitytravel.com](http://www.disabilitytravel.com)

#### **Adaptive Adventures**

Plans adaptive adventure trips. Website: [www.adaptiveadventures.org](http://www.adaptiveadventures.org)

### **Children's Hemiplegia and Stroke Association**

Disney vacation description & links to many other adaptive vacation, camp & recreation resources around the world. Includes airline and other transportation links. Website: [www.chasa.org/vacations.htm](http://www.chasa.org/vacations.htm)

### **Wilderness Inquiry**

"Providing outdoor adventures for people of all ages, backgrounds, and abilities." Website: [www.wildernessinquiry.org](http://www.wildernessinquiry.org) Phone: 1-800-728-0719

### **Quality Mall**

"This department has information and resources for individuals with disabilities who want to plan vacations or who are making plans to travel. It includes information on companies that provide supervised vacation packages, recreational opportunities and travel hints that will make your plans more enjoyable." Also has links for products and other resources. Website: [www.qualitymall.org](http://www.qualitymall.org) (search under 'vacation')

**MESSAGE FROM THE AACT CHAIRMAN...**

**S**pring has sprung! This time of year seems busy, busy, busy with the theme focused upon clearing away the clutter and planning ahead. This could be true domestically and I know it's true in the work force! The UMDF is in a time cycle now in which we have grown as an organization in every area. This brings both benefits and challenges. The more that we do, the more that we know still needs to be done!

This month I wanted to pay tribute by way of an inspirational poem. I want to acknowledge and thank: ALL of the UMDF Staff in Pittsburgh for "going the distance" and blazing a trail, so that we have a path to walk down. Our UMDF Board of Trustees for your leadership and your guidance through our strategic planning process and tough decisions that get to be made. ALL Chapter leaders and Volunteers for caring out our mission and giving of your precious time and energy! Our Doctors, Researchers & Scientists working diligently on our behalf to figure this thing out. ALL of the Moms, Dads, Sisters, Brothers, Family, neighbors, caretakers that help ease our hearts and days. And lastly to YOU ALL that brave each hour of each day, like a warrior, living with Mitochondrial Disease.

Look where we've come, look what we've done...

*Sharon Shaw*

Sharon Shaw  
 Vice Chairman of the UMDF Board of Trustees  
 Chairman of The Adult Advisory Council AACT

**Adult Advisory Council Team (AACT)**

- Sharon Shaw, AACT Chair, Arizona
  - Gail Wehling, AACT Co-Chair, Illinois
  - Bob Brief, New York
  - Marge Calabrese, Arizona
  - Linda Cooper, California
  - Rev. David Hamm, Maryland
  - Etan Harmelech, Connecticut –  
 Young Adult Subcommittee
  - Pam Johnson, MD, Missouri/Kansas
  - Deb Makowski, New York – Adult  
 Liaison Coordinator - West Coast
  - Beate Pohlig, Pennsylvania – Adult  
 Liaison Coordinator - East Coast
  - Erica Schwartz, Connecticut – Young  
 Adult Subcommittee
  - Gregory Yellen, Maryland
- Medical Advisors:  
 Bruce H. Cohen, MD  
 Amy Goldstein, MD

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**PURPOSE OF AACT**

*To represent and serve the unique needs of the affected adult community and to ensure that those needs are adequately represented to UMDF resulting in enhanced services to the affected adult population. AACT is a liaison to the UMDF Board of Trustees and will assess, provide advice and guidance, and make recommendations to UMDF on adult related issues.*

Somebody said that it couldn't be done,  
 But he with a chuckle replied  
 That "maybe it couldn't," but he would be one  
 Who wouldn't say so till he'd tried.  
 So he buckled right in with the trace of a grin  
 on his face. If he worried he hid it.  
 He started to sing as he tackled the thing  
 That couldn't be done, and he did it.

Somebody scoffed: "Oh, you'll never do that;  
 At least no one ever has done it";  
 But he took off his coat and he took off his hat,  
 And the first thing we knew he'd begun it.  
 With a lift of his chin and a bit of a grin,  
 Without any doubting or quiddit,  
 He started to sing as he tackled the thing  
 That couldn't be done, and he did it.

There are thousands to tell you it cannot be done,  
 There are thousands to prophesy failure;  
 There are thousands to point out to you one by one,  
 The dangers that wait to assail you.  
 But just buckle in with a bit of a grin,  
 Just take off your coat and go to it;  
 Just start to sing as you tackle the thing  
 That "cannot be done," and you'll do it.

Edgar A. Guest - ( 1881 - 1959)

**WE WANT YOUR STORIES!**  
 Please consider submitting an article on your experiences with a specific topic that would be of interest to other adults with mitochondrial disease. Contact the AACT council at [AACT@umdf.org](mailto:AACT@umdf.org) with your story! We look forward to hearing from you!

# ANNOUNCEMENTS

working with the volunteer groups several months ahead of the actual walkathon date,” Milsovic said. “We have a step by step process in planning the walk to help our volunteers with their collateral needs, sponsorship information, banners, t-shirts and donation information. We also provide websites for the individual Energy for Life Walkathons. We want to make it a complete, easy and fun experience.”

The UMDF Special Events department will be working all summer long to help with the ‘Energy for Life’ Walkathons and the other events planned around the nation by organizers. “It is our hope that members receiving calls from our volunteers with a request to help will join us in this initiative. We are excited with the success in New England and we hope that excitement carries across the nation by enabling us to hold many more Energy for Life Walkathons,” Milsovic said. “All proceeds from the walkathons help the UMDF provide research that brings us all one step closer to a cure.”



## **SUBMISSION DEADLINE FOR VOLUME 15 ISSUE 3 IS JULY 30, 2010!**

### **UMDF MERCHANDISE**

The UMDF Store is open 24 hours a day, 7 days a week! Simply go to <http://umdf.promoshop.com> and let the shopping begin! UMDF merchandise is a great way to support the UMDF, gain awareness of mitochondrial disease and provide fundraising support. The store is currently running clearance specials on last season’s merchandise as we make room for new items! Don’t forget, Energy Members receive a 10% discount and Life Members receive a 20% discount on all merchandise. Simply follow the directions when ordering to receive the discount.

### **H.R. 3502 AND S. 2858 UPDATE**

Just before press time we learned that H.R.3502 and S.2858 have both received additional co-sponsors. Rep. Emanuel Cleaver II (D-5th/MO) and Rep. John Garamendi (D-10th/CA) have added their names to H.R. 3502. The additional co-sponsors brings the number of members of the House of Representatives who have signed onto the bill to 47. We are hoping to have 100 by December 31, 2010. Senator Maria Cantwell (D-WA) became a co-sponsor of S.2858. We are seeking 25 senate co-sponsors by the end of the year.

On a related note, the UMDF New England Chapter used their successful Energy for Life Walkathon to promote advocacy. During the walk, they signed up more than 250 people to send letters to their Massachusetts representatives in support for H.R. 3502 and S.2858. Great Work!

If you would like to send a letter to your elected official, simply visit the UMDF Action Center at [www.umdf.org/advocate](http://www.umdf.org/advocate).



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DISEASE  
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### **IN THIS ISSUE:**

**Muscle Biopsy, Yes or No?  
Read page 1!**

**Is it mitochondrial cytopathy or  
mitochondrial myopathy?  
See page 3!**

**What’s new in my area?  
Check out pages 4-7!**

**2010 Research Grant Recipients.  
On page 9!**