

## ***UMDF Congressional Briefing Focuses on Potential Treatments***



***Rep. Anna Eshoo welcomes attendees to the Congressional Mitochondrial Disease Caucus briefing. Looking on are Art Estopinan, Marni Falk, M.D., and Charles A. Mohan, Jr., CEO and Executive Director of the UMDF.***

It was a crowded room filled with representatives from numerous Congressional Offices in Washington, D.C., where the United Mitochondrial Disease Foundation (UMDF) presented its third in a series of briefings on mitochondrial disease. The briefing, entitled, *"Mitochondria: Potential Treatments and Therapies That Could Benefit Many"* was presented by the Congressional Mitochondrial Disease Caucus. The caucus, created at the request of UMDF members, is sponsored by Rep. Anna Eshoo (D-Ca-18) and Rep. Tim Murphy (R-PA-18). Currently, the caucus has 16 congressmen who are members.

After welcoming remarks by Rep. Eshoo, attendees heard from Arturo Estopinan. Estopinan is the Chief of Staff for Rep. Ileana Ros-Lehtinen (R-FL-27). Estopinan told his colleagues during the presentation about his son, Arturo, Jr. When Mr. Estopinan and his wife, Olga, took their son home after his birth, the child appeared to be developing normally. After his first birthday, Arturo, Jr., began to show muscle weakness. Very soon after, he lost all of his motor skills and began losing weight. Arturo was diagnosed with a form of

*(Continued on page 5)*

*Every dollar counts! To contribute to research, go to [www.umdf.org](http://www.umdf.org) and click on "Give Today."*

## ***UMDF Launches New Caregiver and Adult Patient Support Program***

Caregivers of people affected by mitochondrial disease and adults living with mitochondrial disease are everyday heroes. The UMDF recently sponsored a new pilot program called Mito S.P.A. (Support. Play. Acknowledge.) as a way of recognizing the needs of this deserving group. The Mito S.P.A. weekend is the brainchild of Missy Knight and Joy Roeh, two Houston, TX UMDF volunteers. The weekend consisted of engaging activities that addressed the needs, hopes, and challenges of caregivers and affected adults. Time was also allowed for participants to network, share ideas, and just take a break from their very busy lives.

Thirty people attended the free weekend program which was held all day Saturday, September 14, 2013, until noon on Sunday, September 15, 2013, in the Houston Embassy Suites. The event would not have been possible without the support of some very dedicated volunteers, including Amber Ferrell, Rebecca Harkleroad, Trisha Kranz, Susan Ladd, Daralyn Phillips, and Miranda Torrey, in addition to Missy Knight and Joy Roeh.

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### ***Featured Articles:***

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# Discover the Power Transgenomic Mitochondrial Disease Genetic Testing



The complexity of diagnosing individuals with mitochondrial disease is well known, Transgenomic has launched multiple initiatives to help clinicians arrive at a definitive diagnosis for their patients. They have collaborated with leaders in the field of mitochondrial disease to understand the challenges they face and provide the testing they need for diagnostic answers.

To enhance doctors' confidence in the diagnosis, Transgenomic uses CarpeNovo, an exclusive state of the art technology that compiles data from 6 data sources (Polyphen, HGMD, 1,000 Genomes Project, dbSNP, OMIM, Exome Variant Database) and employs custom algorithms to annotate variants and improve result interpretation. Finally, a team of geneticists review every case to ensure an accurate interpretation. All of this results in an increased confidence in the diagnosis which will aid clinicians in making appropriate clinical decisions.

To ensure access to Transgenomic's specialized mitochondrial testing services, they have launched the Transgenomic ACCESS Plan (TAP). TAP is for patients with commercial insurance and is intended to limit patient out of pocket expenses. For approximately 85% of all patients, based on income levels, that "out of pocket" expense will be \$49 while all others will be protected and pay no more than 10% the price of the test up to a maximum of \$500. A TAP representative will notify the patient or family of the out of their pocket cost prior to testing. **Now that is comprehensive!**

Test	Description
Complete Genetic Evaluation	High-density SNP array (HDSA) and WES are run simultaneously.
Complete Mitochondrial Disease Evaluation	Mitochondrial whole genome assay (mtDNA). If negative, reflex to the Nuclear Mitochondrial Disease NGS panel.
Whole Exome Sequencing (Individual/Proband)	WES to detect variants in the patient's exome, including coverage of the mitochondrial genome, to determine a genetic diagnosis for disease.
Whole Exome Sequencing (TRIO/Family)	WES for the proband and up to 5 family members.

To learn more about Transgenomic, the Transgenomic ACCESS Plan, and testing for mitochondrial disease please visit [labs.transgenomic.com](http://labs.transgenomic.com)



# From the Chairman

We all know the importance of mitochondria and their role as the powerhouses of our cells. We know that when they do their jobs within our bodies, they provide the power to our organ systems that enable us to walk, talk, see, hear, and breathe. They also perform many other critical functions for each cell in our body. In addition, we know that when they aren't working properly, those same organ systems do not function as they should which leads to mitochondrial disease.



"complex life." But according to the research by Dr. Lane and by Dr. William Martin, from the University of Dusseldorf, the mitochondria may play a much larger role in the process. The research goes on to hypothesize that mitochondria were fundamental to the development of complex innovations like the nucleus and multicellular organisms because of their function as power stations in the cell.

As this one-day symposium continued, there were numerous topics such as the links between cancer progression and mitochondrial dynamics and mitochondria's relevance to Parkinson's disease. I know this is a very simple look at Dr. Lane and Dr. Martin's research, but it does tie in with what we have been saying all along. We believe simply that researching the mitochondria not only impacts our community, but has a much broader impact on human health in general. Many, many diseases create dysfunctional mitochondria and dysfunctional mitochondria from whatever cause equals sickness and even death.

If you would like to delve into this a little further, I suggest you read Dr. Lane's book, "Power, Sex, Suicide: Mitochondria and the Meaning of Life." It really is a fascinating look at how something so tiny, like our mitochondria, impacts everyone.

Energy to all,



W. Dan Wright, UMDF Chairman



HOPE. ENERGY. LIFE.

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# UMDF national

News from the national office.

## Introducing the UMDF's New Science & Alliance Department

by Philip E. Yeske, Ph.D., UMDF Science & Alliance Officer

It was July 2004 and my first daughter, Natalie, then three months old, required minor surgery to correct a throat issue that was contributing to her vague "failure to thrive" diagnosis that our pediatrician provided. Within a few hours she went from operating room to intensive care, mysteriously and dramatically reacting poorly to the general anesthetic. A Ph.D. chemist by training, it was that day that I heard the words "mitochondria" and "disease" used together for the first time in my life. I had no idea that the tiny cellular power houses of biology classes gone by had their own set of diseases associated with them. Looking back, how fortunate my family was to have a diagnosis relatively quickly, because we knew what we were dealing with (neuropathy, ataxia and retinitis pigmentosa or NARP). We were also fortunate to be living nearby to Children's Hospital of Pittsburgh, where Pediatric Neurologist Dr. Amy Goldstein was able to provide loving care and tell us about a great resource for education and support - the United Mitochondrial Disease Foundation.



*Phil with his daughter, Natalie, in 2005.*

Natalie survived her first birthday, but only by a handful of days. Her all-too-short journey as a patient affected by mitochondrial disease, and the support that the UMDF provided during that time, left a lasting impression on me. I had to find a way to help this organization with their mission. I eventually joined the UMDF Board of Trustees, bringing with me the perspective of a mito parent, scientist, businessman, and entrepreneur. About a year ago, UMDF Executive Director Chuck Mohan asked for my help in writing a position description for a new role within the UMDF that would help "connect the dots" between academic research, government agencies, and drug discovery companies. I gladly assisted, immediately seeing the potential for such a role. In fact, my excitement grew to the point of wanting to be involved in not just defining the position, but in being the person to carry the torch.



*Phil with his daughter, Celeste, his son, Nathan, and their dog, London, at the 2013 Energy for Life Walkathon in Pittsburgh, PA. Celeste and Nathan do not have mitochondrial disease.*

After spending the first six months of 2013 working for the foundation on a part-time basis, I became a full-time staff member in July. I am the Dr. Frankenstein that created my title, Science & Alliance Officer, which is meant to convey the importance of both science and business relationships in the development of treatments and cures for mitochondrial disease. It is now broadly accepted that patient groups such as the UMDF are ideally positioned to facilitate this complex process, as our singular goal is to help those affected by disease. The Cystic Fibrosis Foundation has received extensive press for the impact they had on the discovery and development of

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## **New Science & Alliance Department**

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Kalydeco, a drug marketed by Vertex Pharmaceuticals that addresses a global population of perhaps 1,000 - 2,000 patients...the very essence of rare. Rare disease drug development is a hot topic in the pharmaceutical industry, and there are many advances within the mitochondrial research community that are prepared to translate from bench to clinical applications. Some of these advances you have heard about in active clinical trials, others are dots waiting to be connected, while many others still need to be further nurtured. The take home message: it's important to have possibilities in each of these categories in order to have a well-stocked pipeline of therapeutic candidates.

So what does the UMDF need to do to ensure similar success as the Cystic Fibrosis Foundation? In short, an infrastructure needs to be created to support the development of therapeutics, and the good news is we

are already actively engaged in many of the most important components. A few of these components include funding basic and clinical research, creating patient registries as well as a clinical research network, and working to ensure a consistent, global collection of patient data.

In future articles, I will share more details about these initiatives and our plans, and I welcome your input as a member of the mitochondrial disease community. My goal in this introductory article is to share a bit about my journey to this important new role for the UMDF so that you know how important it is to me personally, but also to convey a message of hope. There is a powerful mix of science, advocacy, and business brewing that I firmly believe will lead to exciting new treatments and cures in the coming years for those affected by mitochondrial disease. Stay tuned, never give up hope, and know that the UMDF is working hard to make this vision a reality.

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## **Congressional Briefing Focuses on Potential Treatments**

*(Continued from page 1)*

mitochondrial disease called TK2. Because he lacks the enzyme needed to develop the mitochondria in his little body, Arturo is unable to move his fingers and toes and cannot breathe on his own. Estopinan credited the medical experts at Columbia University for understanding the symptoms quickly and developing a treatment plan. Arturo, Jr., who was not expected to survive, will be going home from the hospital in late October. "This is why we need to advance research and why you, as my colleagues, need to understand this disease and help us fund research," Estopinan told Hill staffers.

Also presenting at the briefing was Marni Falk, M.D. Dr. Falk, who is Assistant Professor in the Division of Human Genetics, Department of Pediatrics at The Children's Hospital of Philadelphia (CHOP) and University of Pennsylvania (Upenn) Perelman School of Medicine, was able to educate the congressional staffers about her work in developing improved diagnostic approaches for mitochondrial disease, including whole exome sequencing approaches in affected families. Dr. Falk is Chairman of the UMDF's Scientific and Medical Advisory Board (SMAB).

Anne Pariser, M.D., Associate Director for Rare Diseases in the Office of New Drugs (OND), Center for Drug Evaluation and Research, US Food and Drug Administration (FDA), shared details of her work to facilitate the research and development of investigational drug and biological products for the treatment of rare diseases. Dr. Pariser is

actively involved in numerous collaborations within FDA and with drug developers, other governmental agencies, advocacy groups and other stakeholders to further the development of products for rare diseases.

At the UMDF Symposium in Newport Beach, CA last summer, attendees learned about 22 potential therapies for mitochondrial disorders. Congressional staffers were then educated about some of the potential therapies and treatments on the horizon from Travis Wilson, President and CEO of Stealth Peptides Incorporated. Stealth Peptides is a clinical stage biopharmaceutical company developing a novel class of mitochondria-targeted peptide therapeutics for treatment of cardiovascular, renal and neurodegenerative diseases. Wilson told the staffers about Bendavia, which is Stealth Peptides lead compound in Phase 2 development. Bendavia has been shown to improve cellular ATP levels in disease, and prevent pathological reactive oxygen species (ROS) formation and opening of the mitochondrial permeability transition pore, thereby improving compromised cardiac, renal and skeletal muscle function.

The briefing was the third in a series that will be presented to Congress. UMDF is planning the next briefing for the early spring of 2014. The topic will focus on aging and age related mitochondrial dysfunction. Urge your congressman to join the caucus. Visit [www.umdf.org/legislation](http://www.umdf.org/legislation) and send a letter to your congressman today!

# UMDF support

Connecting our community.

## UMDF Highlighted Support Events

**June 29, 2013.** A Family Meet and Greet was held at Boyce Park in the eastern suburbs of Pittsburgh, PA for UMDF members in the Pittsburgh area. About 15 people attended the event. Thank you to Amanda Butler for organizing the event.

**July 14, 2013.** A family fun event was organized by the Detroit-area Support Group at the Livonia Recreational Center Waterpark. Thirty-four mito kids, adults, and their families joined in the waterpark fun and socializing with others in the area. Thanks to Julie Scott and Missy Leone for organizing it.

**July 28, 2013.** The first Meet & Greet in Kentucky was co-hosted by the UMDF and Mitoaction. Local families in the Louisville and Lexington areas met to connect and discuss future activities in Kentucky. Thank you to Melissa and Dennis Newton for organizing the event.

**September 5, 2013.** A Meet & Greet with Regional Coordinator Anne Simonsen was held in Grand Rapids, MI. Approximately 22 members & families attended to connect and discuss future activities for the Grand Rapids area. Thank you to Ashlee Senn for organizing the event.

**September 7, 2013.** The South Florida Support Group held a meeting in West Palm Beach, FL featuring Dr. Richard Newman of Melbourne, FL. Thank you Dr. Newman and South Florida Support Group Leader and Ambassador, Holly Schneider for organizing this event.

**September 8, 2013.** The Evansville, IN area members held their regular Support Group meeting to discuss support needs and future plans for the area. Welcome new Support Group leaders Kaitlin Thompson and Jamie Sterchi! Thank you to Melissa Edmondson and Lori Meyer, outgoing support group leaders, for supporting the needs of members in your area for the past year!

**September 14, 2013.** The Atlanta Chapter held it's first ever "Adult Support Meet and Greet" at an Atlanta area Starbucks. Thank you, Volunteer Mark Braitman for setting up this event.

**October 5, 2013.** The Lake Norman Little League in North Carolina held a wonderful awareness event at Bailey Park in Cornelius, NC. The event was called the 2013 Slamboree, "Strike Out Mitochondrial Diseases." A fun UMDF awareness tent was front and center and each Little League player received a practice t-shirt complete with a mito ribbon. Special thanks to Lake Norman Little league Leadership, Mike Burns and Bill Hill and UMDF Member and "Mito Dad" Jerry Yoder for organizing this event.

**Please note:** These events just represent some of the activities offered by the UMDF in June, July, August, September, and October. Over 30 support and educational events filled the UMDF calendar. Be sure to check the calendar at [www.umdf.org/calendar](http://www.umdf.org/calendar) for activities in your area. If you would like to plan a meeting in your area contact UMDF Member Services at [connect@umdf.org](mailto:connect@umdf.org) or call the UMDF, toll-free, 888-317-8633.



*Jennifer and Caitlin Yoder  
at the 2013 Slamboree.*

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## A New Resource from the Genetic Alliance

On October 10, 2013, the Genetic Alliance, in partnership with Family Voices and Parent to Parent USA, announced the launch of a new online resource for individuals with special healthcare needs and their families. The Advocacy ATLAS: Accessible Tools for Leadership and Advocacy Success, features over 250 tools and resources to help new and experienced advocates communicate their needs around services, support, and access.

These resources aim to help individuals with special healthcare needs and their families navigate a world often full of barriers and inaccessibility. The Advocacy ATLAS features resources and perspectives on youth leadership, transition to adulthood, insurance and financial assistance, communicating about your health, access to healthcare, and more. Genetic Alliance, Family Voices, and Parent to Parent USA will continue to reach out to partners for new resources to expand the utility of the Advocacy ATLAS for new and experienced advocates alike. To access the Advocacy ATLAS, go to <http://www.geneticalliance.org> and look under resources.

## UMDF Launches New Caregiver and Adult Patient Support Program

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The success of the event can be best summed up by the participants themselves.

"This weekend allowed me to interact and engage with others in a 'mito' forum where the emphasis wasn't on the medical experience of mito, but rather, the human experience of it."

"An amazing experience shared with those who walk in our same shoes."

"I felt totally comfortable and relaxed, I didn't have to think about trying to walk normal and covering up my symptoms so I could appear as normal as possible."

"It meant a lot to me to connect with other patients - to know that I am not alone in this journey through mitochondrial disease. I often feel so alone regarding my illness, so meeting others who 'get it' was amazing."

With the success of this first event, UMDF hopes to duplicate the program in other areas. The national office and the regional coordinators are reviewing future locations for this program. If you know of possible sponsors and volunteers in your area that would be interested in the Mito S.P.A. weekend, please contact the UMDF at [connect@umdf.org](mailto:connect@umdf.org).



Joy Roeh and Missy Knight  
the creators of the Mito S.P.A.



Attendees pose for a photo at the Mito S.P.A.

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## UMDF Grieving Program

The UMDF reaches out to those who recently lost a loved one affected with a mitochondrial disease. If you know of someone who passed away, and you would like to receive information from the UMDF, please call the UMDF National Office, toll-free, at 1-888-317-8633.

# UMDF Creates “Energy for Action” Across the Globe

UMDF’s volunteers made a tremendous impact across the globe during this year’s Mitochondrial Disease Awareness Week, September 15-21, 2013. The UMDF was able to support its volunteers by sending out free educational materials to raise awareness. The staff sent out nearly 70,000 pieces of collateral. Materials were requested for awareness activities that were held in the United States, Canada, and Ireland. These activities raised awareness in schools, churches, medical communities, places of employment, and to the general public as volunteers either distributed educational materials at awareness and fundraising events or to individuals, such as physicians. Volunteers also created awareness on social media sites, television news reports, and newspaper articles. Way to go UMDF volunteers!

Melissa Jo Branch of Newport, New Hampshire, participated by setting up an informational table at her workplace, Dartmouth-Hitchcock Medical Center. Branch said the booth provided valuable information and was successful. “It was shocking to me that so few physicians knew what mitochondrial disease is, despite having a world-renowned neuro-metabolic specialist working at the facility,” Branch said. “I included a board where families could write the names of the loved ones they had that are fighting or had lost the battle with mito, and I learned it was on a larger scale locally than I had imagined. Hopefully, the new awareness may help find a cure.”



Melissa Jo Branch’s Awareness Week table.

While Branch was handing out materials in Lebanon, New Hampshire, Melissa Edmondson was teaching kids about mitochondrial disease in her daughter, Aly’s first grade class in Evansville, Indiana. One of the items that Edmondson handed out was Energy for Action tattoos, which went with this year’s Awareness Week theme. Edmondson said, “Aly felt much better knowing that other kids get sick too... even if it is a different sick.”



Aly Edmondson in class.

As part of Awareness Week, the UMDF held its third Congressional Mitochondrial Disease Caucus briefing in Washington, D.C. The briefing attracted the attention of 18 Congressional Offices. It was in the briefing that elected officials learned details about therapies and potential drugs that could help mitochondrial disease patients.

## Unique Awareness Week Activities

Denise Andolino of Bloomfield, New Jersey, gave away mito tattoos and information about mito at the hospital where she works. She also sold green hair extensions at the hospital to raise money for the UMDF.

Debra Olivas in Arkansas City, Kansas, always holds a “green week” at work and one evening at her church during Awareness Week. During that week, she has something green for them to eat, but they have to read the educational materials she provided.

Barb Quinn in Ponte Vedra Beach, Florida, ordered awareness materials for her son, Eric Quinn, Jr., who started a Mito Awareness Club at his high school on behalf of his sister. He has given presentations to as many as 200 educators/physicians.

## A Workout Honors Mito Patient

Michelle Palmer in Kansas City, Missouri, promoted awareness by holding a Cross-Fit Workout of the Day (WOD) in honor of her brother, Travis Palmer. The workout consisted of 4 rounds (1 in 4 chance a child is affected if parents are carriers) - 19 pull ups (Travis’ age), 90 double under jump ropes (mito effects 90% of the body’s energy), 30 kettlebell swings, 10 power cleans (every 30 minutes a child is born who will develop mito by age 10), and 20 burpees, which took an average of 30 minutes to complete. Afterward, the entire group did a wall sit, as long as they could to represent no cure for mito diseases. Palmer said, “Everyone was extremely motivated to use their muscles for those who can’t, and a few of the guys even teared up as Travis was smiling the whole time!”



Jamie Sterchi of Vincennes, Indiana, talked to the students at her son, Brady’s school about mitochondrial disease during awareness week.

# MITOCHONDRIAL MEDICINE 2014: PITTSBURGH

SHERATON STATION SQUARE HOTEL, PITTSBURGH, PA



## Scientific Program: June 4 - June 7, 2014

Course Chair: Jerry Vockley, M.D., Ph.D.

Course Co-Chair: Amy Goldstein, M.D.

CME Chair: Bruce H. Cohen, M.D.

### 2014 Platform Sessions Include:

- Architecture of Energy Metabolism
- Mitochondrial DNA Damage & Repair
- Drug Discovery/Clinical Trials
- Animal Models
- Tools for Research (Lab Oriented Session)
- Clinical Studies Updates
- Neurodegeneration

The scientific program and the invited faculty are now posted on the symposium web page. Call for abstracts is NOW open and will close on March 17, 2014.

All abstracts must be submitted electronically online at [www.umdf.org/callforabstracts](http://www.umdf.org/callforabstracts).

For updates, please visit [www.umdf.org/symposium](http://www.umdf.org/symposium). Once registration is open, please use code SCI1408 for online registration. For information on continuing education please e-mail [symposium@umdf.org](mailto:symposium@umdf.org) or call 888-317-UMDF, ext. 114. Travel scholarships are available for new researchers! For details, go to [www.umdf.org/symposium](http://www.umdf.org/symposium).

## Family Program: June 6 - June 7, 2014

### Special LHON Program: June 5, 2014

### Tentative Topics for 2014:

- Mitochondrial Disease: What is It and What to Expect?
- Current Therapies in the Treatment of Mitochondrial Diseases
- Screening and Evaluation in Patients with Mitochondrial Disease
- Ask the Mito Doc Panel Discussions for Adult and Pediatric Patients
- Palliative Care
- Exercise and/or Nutrition
- Adult Specific Topics
- Young Adult and Teen Sessions

For updates, scholarship, and hotel information, visit [www.umdf.org/symposium](http://www.umdf.org/symposium). Once registration is open, please use code FAM1409 for online registration.

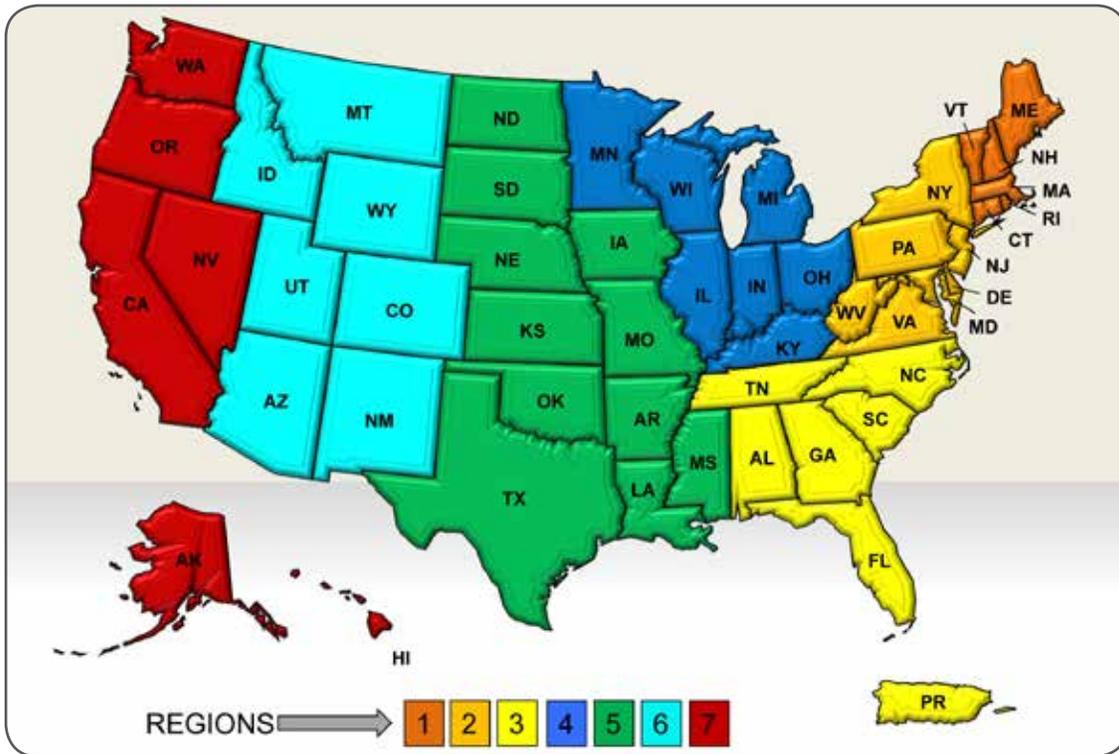
## Did You Ever Wonder How UMDF Decides Which Research Grants to Fund?

The 2014 UMDF grant cycle opened July 1st, 2013 and the deadline was September 13, 2013. Two-hundred and six Letters of Intent (LOIs) were submitted, which was a substantial increase from the numbers over the last few years. The applicants represent 21 different countries, a new record! Next, any LOIs not UMDF mission-appropriate are triaged by the grant co-chairs. Each remaining LOI is then assigned reviewers, many of whom are on the Scientific and Medical Advisory Board. Those LOIs that score well will then be invited to send a full proposal. The proposals will be reviewed and then discussed by the Grant Review Committee in April. Final grant selection recommendations will be made at the grant review meeting, and then sent to the UMDF Board of Trustees for approval.



In addition to the support staff at the UMDF National Office, help is available to you across the United States and around the world. To reach a state contact for support and/or if your state is not listed and you would still like to connect, simply e-mail [connect@umdf.org](mailto:connect@umdf.org). When sending the e-mail, please include the leader's name or city/state in the subject line for us to better serve your needs. Interested in getting involved? Visit [www.umdf.org/volunteeropps](http://www.umdf.org/volunteeropps) or call 1-888-317-8633!

**Note: The volunteer support leaders listed here have completed a Support Group Leader and/or Support Ambassador Agreement. If your name is not listed, please check with Member Services on the status of your agreement.**



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Amber Taylor, Bangor  
**VERMONT**  
MaryBeth LeFevre

**CONNECTICUT, NEW HAMPSHIRE, RHODE ISLAND**

Contact the National Office to Connect

**MASSACHUSETTS**  
Julie Gortze, North Attleboro, MA

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Kathleen Stapleford, Magnolia  
Judy Weeks, Dover

**DC** – See Virginia

**MARYLAND**  
Dawn Murphy, DC/Baltimore/  
Northern Virginia Chapter

**NEW JERSEY**  
Laurel Smith, Delaware Valley Chapter  
Carrie Mullin, Pittsgrove, NJ

**Northeast Region (2)**

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Linda Roesch, Buffalo, Western NY Support Group  
Jennifer Schwartzoff, Buffalo, Western NY Support Group  
Erica Beyea, Buffalo, Western NY Parents Support Group  
Sandy Sallaj, Buffalo, Western NY Parents Support Group  
Jacqueline Perrotta, Albany  
Lori Piccirilli, Binghamton  
Kimberly Dedrick, Utica

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Jessica Myers, Erie Mito Group  
Kim Olenderski, Central Pennsylvania  
Heather Pallas, Pittsburgh (children)  
Karen Wilson, Pittsburgh (adults)

**VIRGINIA**  
Heather Meyer, Lynchburg  
Judi Bartle, Central Virginia Support Group  
Sharon Hoffert, Central Virginia Support Group

**Northeast Region (2)**

Sharon Goldin, DC/Baltimore/Northern Virginia Chapter  
Anne Tuccillo, DC/Baltimore/Northern Virginia Chapter

**WEST VIRGINIA**  
Contact the National Office to Connect

**Southeast Region (3)**  
*Margaret Moore, Regional Coordinator*

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Marissa Benjamin, Leeds

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Garry Krueger, North Central Florida  
Joan Morris, Titusville, FL  
Denise Richardson, Fort Lauderdale  
Holly Schneider, Coconut Creek  
Jennifer Slauter, Orlando, Central FL Mito Group  
Sophie Szilagy, North East Florida  
Marla Tobia, Tampa

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Hannah Bossie, Athens  
Sebastien Cotte, Atlanta  
Mary Beth Morris, Atlanta  
Cheryl Porter, Atlanta  
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Shelly Lorenzen, Sugar Hill  
Wendy Clegg, Loyd, Columbus  
Tiffany Tuggle, Stockbridge  
Erin Willis, Valdosta

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Adriana Smith, Raleigh Durham  
Jenny Hobbs, Winston-Salem  
Terry Holeman, Fayetteville  
Christy Koury, Charlotte  
Kris Shields, Charlotte

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Group  
Karrie LaCroix, Memphis area  
Courtney Fellers, Nashville area  
Nancy Garrison, Nashville  
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Cleveland  
Chris & Alisa Rawski, Northwest Area/  
Toledo

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Appleton, Green Bay  
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Tomahawk, Minocqua Area  
Josh Bartz, Southeast Area/Madison  
Karen Loftus, Southeast Area/  
Milwaukee

## OKLAHOMA

Contact the National Office to connect

## West Region (6)

### ARIZONA

Marin Pelletier, Phoenix

### IDAHO

Jennifer Pfefferle, Boise, Idaho Support Group

### NEW MEXICO

Stephanie Cassady, Albuquerque

### UTAH

Laura McCluskey, Orem

### COLORADO AND WYOMING

Contact the National Office to Connect

## Pacific and Northwest Region (7)

### CALIFORNIA

Norma Gibson, California Chapter  
Cheryl Burge, Inland Empire  
Cory Greenlee, La Verne

### HAWAII

Kimo Phan, Honolulu

### OREGON

Kimberli Freiling, Monmouth Support  
Group

### WASHINGTON

Mareesa Henderson, Spokane  
Joy Krumdiack, NW Washington

### ALASKA, MONTANA, NEVADA

Contact the National Office to Connect

### INTERNATIONAL

Rob Ryan, Australia  
John Carreiro, British Columbia  
Atanga Emmanuel, Cameroon  
Andrew Alexander, Hungary  
Nilam Agrawal, India  
Saijad Haider, Pakistan  
Anne Hansen, Norway  
Vidar Hunstad Vik, Norway  
Rowland Dicker, United Kingdom  
Keely Schellenberg, Winnipeg

### YOUTH AMBASSADORS

Hannah Eichner, North Carolina  
Joe Wise, California  
Emily Swinn, Georgia  
Briana Garrldo, Hawaii  
Haley Wroth, Massachusetts/Connecticut  
Alexandra Simonian, New York  
Tyler Liebegott, Pennsylvania  
Colleen Powell, Pennsylvania  
Devin Shuman, Washington  
Jordan Schmeer, Virginia

### PDCD AMBASSADORS

Melissa James, Bernardston, MA  
Michelle King, Lima, OH  
Jillian Austin, Cranberry Township, PA

### LHON AMBASSADOR

Lissa Poincenot

### CREATINE DEFICENCY SYNDROMES

Linda Cooper  
Jane Duffy  
Kathy Furtner  
Beth Robinson

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## Great Lakes and Midwest Region (4)

*Anne Simonsen, Regional Coordinator*

### ILLINOIS

Cherie Lawson, Chicago Area Support  
Group  
Vicki Ternberg, Chicago Area  
Gail Wehling, Chicago Area Support  
Group  
Luke and Leslie Kirby, Philo  
Victoria Helms, Southwest Area/St. Louis  
Support Group

### INDIANA

Melissa Edmondson, Evansville Area  
Jamie Sterchi, Evansville Area Support  
Group  
Kaitlin Thompson, Evansville Area  
Support Group  
Jackie Parrish, Indianapolis Area

### KENTUCKY

Krystena Richards, Lexington &  
Cincinnati Areas  
Mandy Salensky, Lexington &  
Cincinnati Areas

### MICHIGAN

Carrie Gervasone, East Michigan/  
Detroit Area  
Missy Leone, East Michigan/Detroit Area  
Support Group  
Julie Scott, East Michigan/Detroit Area  
Support Group  
Genevieve Angeloff, Upper Penninsula  
Holly Worden, West Michigan/Grand  
Rapids Area Support Group

## Central Region (5)

*Cassie Franklin, Regional Coordinator*

### ARKANSAS

Lacie Moore, Rogers

### IOWA

Ronda Eick, Northern Iowa  
Kim Novy, Des Moines, Iowa Support  
Group

### KANSAS

Anne Tramposh, Kansas City Area

### LOUISIANA

Nicole DeJean, Lafayette  
Rhonda Mailhos, New Orleans Support  
Group  
Mandy Poche, Baton Rouge  
Tammy Reyna, Bossier City  
Anna Stewart, Bossier City  
Chantel Wooley, Covington

### MISSISSIPPI

Tracy Shedd, Vicksburg

### MISSOURI (see also Illinois)

Matt Bishop, Kansas City Area  
Keli Stone, St. Louis Area Support Group

### TEXAS

Shamayn Kennedy, Wichita Falls  
Jamie Buryanek, Houston  
Melissa Knight, Houston  
Trisha Kranz, Houston  
Kari Richardson, Houston  
Joy Roeh, Houston  
Crystal Lopez, Dallas/Fort Worth  
Shawna McElveen, Dallas/Fort Worth  
Heather McNair, Dallas/Fort Worth  
Laura Torres, San Antonio  
Kristen Wilson, San Antonio

### NORTH DAKOTA, SOUTH DAKOTA, & MONTANA

Marty Campbell, Beach, ND

### NEBRASKA

Dana Ritterbush, Omaha

# Ask The Mito Doc<sup>SM</sup>

Living 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 [www.umdf.org](http://www.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.

Responders for this issue include:  
Fran D. Kendall, M.D., Virtual Medical Practice in Atlanta, GA and Northside Alpharetta Medical Campus in Alpharetta, GA and Mark Tarnopolsky, M.D., Ph.D., FRCP(C.), McMaster University, Hamilton, Ontario, Canada.

**The Question is...** What are the chances of a patient being misdiagnosed with mito? Is it common? How would one know of a misdiagnosis? My 2 year old son was recently diagnosed with mito through a muscle biopsy. Results came back that he has complex 1. I was concerned that maybe he was misdiagnosed because he seems totally fine. His main problem was poor weight gain and a food aversion. He also had reflux. His appearance is normal, good muscle strength and tone. He walks (runs).

## Response from Fran D. Kendall, M.D.

Varying information indicates that muscle biopsy enzymology may demonstrate a false positive result in 30% to 50% of cases. This rate may be due to a variety of reasons including secondary mitochondrial enzyme abnormalities caused by non-mitochondrial disease. Personally, I have re-diagnosed many children with various complex abnormalities identified on muscle biopsies with a wide variety of other diseases ranging from closely related energy disorders such as Rett syndrome, SCN1A or other seizure gene disorders, Lesch Nyan syndrome, chromosome abnormalities and even rickets. The cases that I have re-diagnosed have typically been in children whose clinical features were concerning for another disorder (for example, intractable seizures that are typically seen with many seizure gene disorders) and/or in those patients who had NO other laboratory abnormalities supporting mitochondrial disease (such as increased lactate or CPK, low carnitine, Krebs cycle intermediates on urine organic acid analysis). As such, I am a proponent of screening children for any and all other disorders that could cause their problems before moving towards invasive muscle testing. In my experience, this would have eliminated essentially all of the false positive cases I have seen. When I begin my evaluation for mitochondrial disorders, I typically utilize blood, urine and other non-invasive studies as a first approach to screen patients for possible mitochondrial disease. Depending on the testing results I may pursue more definitive testing using gene studies or skin and muscle biopsy samples. If you have questions about your son I would encourage you to seek a second opinion by a mitochondrial specialist to review the testing results of your child in the context of his clinical features.



**Fran D. Kendall, Ph.D.**



**The Question is...** Can a diagnosis be made simply on someone's symptoms and a finding of an unknown variant of mtDNA change, if all other things have been ruled out? For example in my case, they have ruled out many other conditions but have the finding of the mtDNA change and fatty acid oxidation levels that are outside of normal ranges. It seems that for many the diagnosis process is not straight forward so I'm trying to figure out at what point they can say it is mitochondrial disease? How many tests need to show positive or is there some other standard that indicates mito? Many patients are struggling for years to get a diagnosis and I'm not clear how definitive the testing needs to be for the doctors to be able to say that it's mito? Another aspect to my question is, if an unknown variant is found in testing (in my case, a novel variant on the CYTB gene that is homoplasmic) and there is no one else to compare to, can it be diagnosed it as mito?

## Response from Mark Tarnopolsky, M.D., Ph.D., FRCP(C)

There are many answers to this question and it is very complex. The diagnosis of mitochondrial disease is very similar to many others with categories as; definitive, probable, possible and unlikely often being used (i.e., in polymyositis). The difficulty in such disorders (mito included) is that there are diseases that look like mito and can have many positive "mito" tests but are actually another disease (i.e Rett, chromosomal copy number variations with complex I deficiency- these are false positive disorders). On the other hand there are people with true mitochondrial disorders who are negative on all standard testing (i.e., a boy 15 with 11778 LHON mutation who will become blind in a year but has neither symptoms nor abnormal screening tests (lactate, CK, alanine, etc.- false negative disease testing).

We and others have found homoplasmic mutations in genes (including cytochrome b) that lead to mitochondrial changes and symptoms and have proven that they are pathogenic with specialized tests (hybrids) BUT this type of testing is not often available outside a research lab. In time we will have a better idea of what mtDNA sequence variants are associated with true disease and what are benign and what are "risk factors" - Most doctors are too stuck on the one gene mutation = one idiosyncratic disease model; however, for mitochondrial disease, it is likely that heteroplasmy, multiple combination sequence variants and the variants themselves are on a spectrum from 100 % pathogenic and 100 % penetrant to 0 % - consequently, your cytochrome b sequence variant can lie anywhere along this spectrum.

Usually I use a set of rules/guidelines based upon: symptoms + mutation + lactate, + alanine + urine organic acids + CK + neurological exam + MRI/MRS + muscle biopsy (light and electron microscope) + mtDNA deletion and depletion to put people into a definite, probable, possible, or unlikely category.

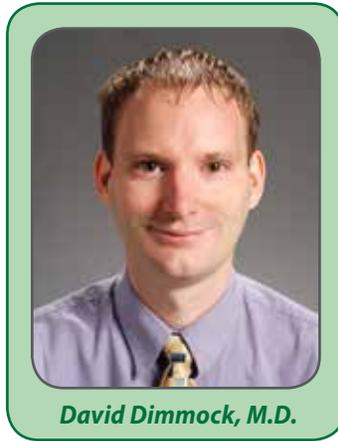
It would be illogical and irresponsible to diagnose someone from symptoms alone as having mitochondrial disease, and most prudent to be careful and use some version of the above tests so that the wrong diagnosis is not made (i.e. calling another disease mito and missing proper treatment, calling "fatigue" and muscle aches as mitochondrial disease when it is just life/aging, missing a true diagnosis of mitochondrial disease, etc).



**Mark Tarnopolsky, M.D., Ph.D., FRCP(C)**

# In the Clinic

The Genetics Center at the Children's Hospital of Wisconsin is a regional referral center that provides ongoing care for patients with mitochondrial diseases. Our care consists of extensive consultative services to establish the underlying cause of disease for patients with a variety of muscle, liver, and neurological problems that might result from a mitochondrial disease. These services include state-of-the-art muscle histology, electron microscopy, and molecular testing. In addition, the physicians at the center have pioneered the use of whole genome and whole exome sequencing for clinical diagnostics. The physicians maintain an active research program, including over 15 clinical trials into new therapies for rare genetic disorders. We are also the primary site for two large trials into new molecular tests for mitochondrial disease. As a health care team, we aim to excel at providing family-focused care which supports individuals in all stages of mitochondrial disease.



David Dimmock, M.D.

We have used genetic engineering to stop the DGUOK gene working in a rat model. We are studying this rat model to evaluate what things make the disease worse in individuals with mitochondrial DNA depletion. We are also working to see if a specific therapy we have makes the disease better; promising preliminary results have led us to try this treatment in a single person. However, we are still a very long way off from a larger clinical trial. The rat is a useful model animal for our blood-based tests for mitochondrial DNA depletion.

## 6. How many mitochondrial disease patients do you see?

We have about 50 patients with confirmed mitochondrial disease that we take care of at any given time, and many more with clinical mitochondrial disease who do not yet have DNA-confirmed disease. We see about 100 new patients per year with suspected mitochondrial disease.

## 7. Are there openings for new patients?

Yes, we can typically see patients within a few weeks of requesting an appointment, depending on insurance predetermination and the specific physicians and genetic counselors the patient requests to see during their visit.

## 8. What are your plans and goals for the future at your clinic?

We strive to provide excellent holistic care for our patients with mitochondrial disease and their families. We will continue to provide the best available clinical diagnostic testing and compassionate care while increasing the number of active clinical trials. I see that mitochondrial medicine is on the verge of a revolution, much the way that childhood cancer was in the 1960s. The key growth areas that I see in the next five years are:

1. A dramatic improvement in the ability to diagnose the specific defect present in an affected individual.
2. The emergence of mitochondrial disease specific treatments, such as our small molecule therapy for DGUOK and EPI-743.
3. Evaluation and establishment of the benefit or lack of benefit of some of the current supportive therapies, such as oral arginine or citrulline for MELAS.
4. The use of medications such as PTC124 and RTC13 that work for many genetic diseases to provide significant benefit to patients with nuclear inherited mitochondrial disease.

## 9. Tell us who works on your team?

We have a core Metabolic Genetics Clinic Team caring for our mitochondrial disease patient population. At each visit, patients meet with a geneticist and genetic counselor. The genetic counselor acts as their case manager. We offer other resources to our patients including special needs care coordination team for those requiring five or more specialists involved in their care, as well as neurology and palliative care services. We also have a large Genetics Center Department with additional physicians, a nurse practitioner, and genetic counselors. If interested, individuals can call the Central Scheduling Department, toll-free, at 1.877.607.5280 to schedule an appointment. We do have an Access Program to assist in scheduling more than one appointment if traveling from a distance.

### Mitochondrial Physicians:

Dr. William Rhead  
Dr. Gunter Scharer  
Dr. Esperanza Font-Montgomery  
Dr. David Dimmock

### Other Geneticists and providers at our center (non-mitochondrial disease):

Dr. David Bick  
Dr. Donald Basel  
Dr. Esperanza Font-Montgomery

### Mitochondrial Genetic Counselors:

Amy White, MS, CGC  
Linda Carey, MS, CGC  
Jeff Kopesky, MS, CGC

### Nurse practitioner:

Stephanie Offord

### Mitochondrial Muscle Pathologist:

Dr. Mike Lawlor

## 1. Tell us how you became interested in mitochondrial disease patients?

I became actively involved in caring for patients with mitochondrial disease at Baylor College of Medicine. One memorable occasion was when I was on call with Dr. Scaglia and we had a patient with a MELAS stroke-like episode. We decided to use IV arginine for the first time. It was very exciting to try a new treatment.

When I was at Baylor I began research with Dr. Lee-Jun Wong. We were focused on improving the diagnosis of mitochondrial disease using DNA tests. As a result of this we were able to better understand the range of symptoms seen in the mitochondrial DNA depletion disorders. We were able to uncover that some patients with liver failure got better with supportive care. Following this experience I have spent the last eight years working on ways to improve the DNA testing for mitochondrial disease and have focused specifically on understanding what goes wrong with DGUOK deficiency and how we might be able to treat it.

## 2. Does your clinic accommodate children and adults?

We see individuals of all ages with suspected mitochondrial disease.

## 3. Does your clinic diagnose patients and provide follow up care?

**Diagnosis:** We are happy to see patients to try and work out the cause of their healthcare problems and to provide second opinions on patients from anywhere in the world.

**Follow up care:** We work very closely with specialists that the patient has chosen or that work at our pediatric and adult hospitals to provide coordinated care. We enjoy working closely with primary care doctors with whom patients have established medical homes.

## 4. Do patients need referrals?

Some patients' insurance companies will require referrals to see a specialist. In general, receiving a referral from a primary care doctor or specialist with information on their specific concerns is helpful. However, as a clinic we do not require a referral. If patients wish to see a specific physician, we are happy to accommodate that request.

## 5. What types of mito research are you doing right now? And have you made any discoveries in mito research?

My laboratory is focused on two specific areas: we are working on DNA-based testing to improve the diagnosis of individuals with mitochondrial disorders.

Specifically, we are evaluating the role and timing for consideration of whole genome and whole exome sequencing as a diagnostic test in patients with suspected mitochondrial disease prior to muscle biopsy.

We have spent many years working on a specific test for mitochondrial DNA depletion. We have made significant strides to have this test work well on liver samples. We have preliminary data suggesting that a new approach may be almost as useful but will avoid the need for biopsy since it can be done on blood.

## UMDF Highlighted Education Events

**July 25, 2013.** A Patient and Family Meeting was held in conjunction with the Texas Parent 2 Parent Annual Conference in San Marcos, TX featuring speakers Dan Wright, UMDf Board Chair, and Dr. Bruce Cohen of Akron Children's Hospital. Dr. Cohen presented and answered questions from the families and patients in attendance. Approximately 50 attendees came from across Texas and from as far away as Oklahoma. A special thanks to local volunteer, Becky Raiber, Dr. Mary Elizabeth Parker and the staff of Texas Parent 2 Parent for making this event possible!

**August 7, 2013.** Over 40 families and members from the Minneapolis/St. Paul, MN area attended a family meeting with special guests Chuck Mohan and Dr. Bruce Cohen. Mohan presented Decade of Difference, highlighting UMDf efforts in the past 10 years in the areas of research, clinical trials, gene sequencing, advocacy, membership, educational and doctor outreach. Dr. Cohen presented an overview of mitochondrial disease. Attendees had the opportunity to ask questions to both presenters. Atom Wolff of Minneapolis, MN said, "I'm actively following the progress of two drugs (EPI-743 and Bendavia) as they move through phase II clinical trials. Both of these drugs might be helpful for some types of mito patients (such as myself). Dr. Cohen was able to provide accurate, knowledgeable, and current information about the progress of these pharmaceuticals. This information was extremely helpful to me."



*Dr. Bruce Cohen talking to attendees at the Family Meeting in Minneapolis/St. Paul, Minnesota.*

**August 8, 2013.** Following the August 7th Minneapolis/St. Paul meetings, Chuck Mohan and Dr. Cohen traveled to Rochester, MN to present for southern Minnesota members and families. There were 12 in attendance at the meeting. Thank you to the Mayo Mitochondrial Disease Biobank for volunteering at the event. Andrew Nesseth of Jackson, MN said, "It was so refreshing to talk with an expert in the field of mitochondrial medicine, face-to-face. Opportunities to speak with someone with the breadth, knowledge, and wealth of experience doesn't come along every day."

**September 29, 2013.** Dr. Richard Boles visited New Orleans, LA for a Patient/Family Meeting and Symposium Synopsis. Symposium 2013 attendees from Louisiana had agreed unanimously that a

local meeting with Dr. Boles for his symposium presentation on "Dysautonomia and Autonomic Issues" would be especially helpful for New Orleans and greater Gulf area patients and families. This Symposium Synopsis meeting was made possible by grant funding from the Joe T. and Dorothy Dorsett Brown Foundation in Metairie, LA.

**October 5, 2013.** UMDf Dallas members that attended the UMDf Annual Symposium earlier this year presented a Symposium Synopsis Webinar. Sara Brown, Shawna McElveen, and Heather McNair shared their favorite sessions from the Family Meeting while Dan Wright gave a recap of the Science Meeting. If you were unable to join, this Symposium Synopsis Webinar is available as a recording in the Multimedia Library of the UMDf website.

**October 8, 2013.** A Patient and Family meeting featuring Dr. Bruce Cohen was held at Nemours Children's Hospital in Orlando, FL. Over 50 people attended this event. It was a great evening of education and fellowship! Thank you to volunteers Luz Oquendo, Micaela Merrills, and Jennifer Slauter for organizing this event!



*Micaela Merrills, Jennifer Slauter, and Luz Oquendo pose at the Patient and Family Meeting in Orlando, FL*

**Please note:** These events just represent some of the education events offered by the UMDf in July, August, September, and October. For meeting details, visit the UMDf calendar on the UMDf website: [www.umdf.org/calendar](http://www.umdf.org/calendar).

### Save the Date!

**UMDF Great Lakes Regional Symposium**  
 March 21-22, 2014 ~ Indianapolis, IN

**Target Audience – Clinicians, Nurses,  
 Allied Health, Patients, Families, and Caregivers.**

**E-blasts will be sent with more  
 information in November/December.**

# Challenges and Triumphs of Hosting Educational Activities

by Kara Strittmatter, Director of Chapter and Membership Services

As the UMDF's oldest employee (not in age but years in service – smile), I have personally witnessed many changes in our mitochondrial community. The most significant has been the increase in educational outlets – not only for patients but for the medical community we serve. The UMDF alone has gone from hosting one international symposium and a handful of patient meetings to organizing more than 15 physician Grand Rounds and dozens of patient meetings each year. Since 2003, the UMDF has reached more than 4,000 clinicians and even a larger number of patients through its educational efforts. This is one of our triumphs!

A challenge typically faced during our meetings is our diverse audience. We have patients newly diagnosed who are starting from scratch with very little mitochondrial disease knowledge and then we have “the veteran” mitochondrial caregiver who could be mistaken for a doctor with the amount of knowledge they have gained over the years. Regardless of their level of knowledge, they are all eager to gain more information, and we are happy to bring speakers to them that do their very best to talk to both audiences. As I watched Dr. Douglas Wallace speak to the patients and parents at this Philadelphia meeting, there were a lot of “aha” moments filling the room. He



Dr. Douglas Wallace speaking to the attendees at the meeting in Philadelphia, PA.

is quite engaging as a speaker and yes, there was laughter as well! This challenge ended in a triumph for those seeking knowledge – everyone walked away with at least one bit of information they didn't have before.

However, for those attendees who may have heard it all before, the networking and socializing during these meetings is wonderful. It is not just about the education...we take pride in bringing patients, parents, and caregivers together who share the same interests. Witnessing someone overcome with joy after realizing they were not alone in their struggles is priceless. TRIUMPH!!!

There will always be challenges in educating the world about mitochondrial disease and the UMDF is ready to face those head-on. With your help, we will turn challenges into triumphs with every activity. We will continue to raise awareness in our medical and patient communities through distribution of literature and encouraging physicians and families to become part of the UMDF. We will continue educating the decision-makers at the National Institutes of Health through our advocacy efforts. We will expand the Grand Rounds program and introduce regional symposiums. Regional symposiums will include a full day of CME for the medical community and a half day program for patients and families in select regions. Become part of the UMDF's volunteer network for such activities and visit [www.umdf.org/volunteeropps](http://www.umdf.org/volunteeropps) today!



Ladies networking at the meeting.

## Last Grand Rounds and Patient and Family Meetings for 2013!

**Grand Rounds**  
November 21, 2013 ~ Noon  
Univ. of Alabama ~ Birmingham, AL

**Patient and Family Meeting**  
November 21, 2013 ~ 6:00 pm - 8:00 pm  
Lakeshore Foundation ~ Birmingham, AL

**Grand Rounds**  
November 22, 2013 ~ Noon  
Univ. of Alabama ~ Birmingham, AL

*For complete meeting details,  
visit the UMDF calendar on the  
UMDF website: [www.umdf.org](http://www.umdf.org).*

## Fundraisers Benefiting the UMDF

**July 20, 2013.** Greg and Carolyn Martin organized the ninth annual Carter Classic golf outing at the Prairie Creek Golf Course in Maquoketa, IA. Each July, family, and friends are invited to the event in honor and memory of Carter Martin. Thank you Greg and Carolyn!

**August 3, 2013.** The sixth annual Run4Raley event was held at the Philo Ball Park in Philo, IL. The annual event is held in honor of Raley Kirby. The event raised nearly \$22,000. Thank you Leslie and Luke Kirby!

**August 4, 2013.** Heidemarie Chernushin participated in the Cleveland Triathlon as 'Moving for Mito' in honor of her friends Alexander & Lauren Lindemann. Heidemarie raised over \$340 for the UMDF this year! Thank you Heidemarie for supporting the UMDF!

**August 10, 2013.** Team Slippery When Wet participated in this year's Cincinnati Mudathon in support of the UMDF! The team of 10 got down and dirty and raised over \$200 for the UMDF! Thank you to David Murphy for organizing the team!



*Team Slippery When Wet at the Cincinnati Mudathon.*

**August 16, 2013.** An art gallery show was held at Gallery Nucleus in Alhambra, CA where artists auctioned off their work and donated all proceeds to the UMDF. Over \$9,000 was raised. Thank you to Francesca Esquenazi and Riot Games for organizing this event! You can still purchase artwork at this web address: [www.umdf.org/riotgames](http://www.umdf.org/riotgames).

**August 16-18, 2013.** Thank you to all of the teams that came out in support of Baylee's Ball Bash this year! The annual softball tournament held at Cannelville Softball Field in Crooksville, OH raises money for the UMDF in honor of Baylee Thompson! Thank you to Jody for all of your hard work!

**August 17, 2013.** The second annual Swing for a Cure for Alex Golf Outing was held in Tiffin, OH in honor of Alex Sendelbach. The outing featured door prizes, a basket raffle, and kids activities. The outing raised over \$3,000. Thank you to the Sendelbach family for organizing this event!

**August 24, 2013.** Shoppers supported the UMDF by purchasing a \$5.00 coupon to Macy's Shop for a Cause and enjoyed a day of spectacular discounts. The event raised \$2,665.

**August 24, 2013.** AMVETS Rider's Chapter 88 hosted the 1st annual Cruize for a Cure in Bronson, FL. This motorcycle ride/ auto poker run included raffles, auctions, food, music, and fun! Over \$2,000 was raised. A special thanks for Jenna Clifton for organizing this event!



*AMVETS Rider's Chapter 88 at the 1st annual Cruize for a Cure.*

**September 2013.** Baxter Pharmaceutical Solutions in Bloomington, IN held month-long events in honor of Jack Thomas Edwards. Along with a UMDF Light Bulb Campaign, they also held a bake sale and other various events to raise funds for Jack's research fund! Thank you Carol and everyone who helped organize these events which raised over \$3,550!

**September 2013.** CDM Smith Inc. held a month long fundraiser in Maitland, FL in memory of Dalton Liam Benjamin and raised over \$1,100! Thank you to David Rasmussen, Benjamin Pernezny, Aaron Rogge, and Michael Cahill for organizing this event!

**September 7, 2013.** The 3F Nursing Staff at Nemours Al DuPont Hospital for Children held their first Bowling for Mito event in Wilmington, DE! The wonderful event raised over \$890 for the UMDF! Thank you to Corrie Scarberry for organizing this great event!

**September 14, 2013.** Applebee's of Lorain, OH hosted a pancake breakfast during mitochondrial disease awareness week in honor of the Candelario family. Thank you Selena Candelario for organizing this event.

**September 14, 2013.** The third annual Lizzy's Walk for KSS made its debut in the fall this year! The event, which was held in Norwalk, CT raised over \$16,000 for the Elizabeth Piro Research Fund with the UMDF! Thank you to Keelin and Cindy Walsh for all of your help again this year!

**September 14, 2013.** The seventh annual Mito Bowl was held in Meridian, ID by the Mito Group of Idaho. The annual event raised nearly \$1,000 for the UMDF! Thank you to Jennifer Pferrerle for organizing this event!

**September 14, 2013.** Faith McColl held her third annual lemonade stand in Marietta, GA and raised over \$2,200! You are amazing, Faith! Thank you for being such a wonderful, young fundraiser!

**September 15, 2013.** The Brittany Wilkinson UMDF Golfing for a Cure outing was held in Clovis, CA and raised over \$1,000 for the Brittany Wilkinson Research Fund with the UMDF. Thank you to the Wilkinson Family for your hard work and dedication!

**September 19, 2013.** A 'Give Back' Night was held at Chili's Restaurant in Cummings, GA. Various other restaurants in the area held similar nights during awareness week in which a percentage of sales were donated to the UMDF! Thank you to Lisa Tatum and your crew for all of your hard work!

**September 20, 2013.** The Pittsburgh Group of the UMDF attended a Mitochondrial Disease Awareness Night at a Pittsburgh Pirates game! Our word got out to a sold out crowd during pre-game! Over \$440 was raised by ticket sales to our members!

**September 21, 2013.** The sixth annual Savannah's Hope Mito Walk was held at the Adventure Life Reformed Church in Altoona, IA. The event included family-friendly activities, and it raised \$52.00 for the UMDF.

**September 21, 2013.** The fifth annual Ayden and Faith Hingsbergen bike run was held in Cincinnati, Ohio. Nearly sixty bikers showed up and raised \$2,000 for the Ayden and Faith Hingsbergen Research Fund. Thank you for your generous contribution!

**September 21, 2013.** The Samuel Cutliff Research Fund held their first annual Walk and Awareness Day in Anderson, SC. Three additional families joined the Cutliff's as they celebrated the life of sweet Samuel and raised over \$2,000. Thank you for your support!

**September 22, 2013.** The annual Brittany's Walk toward Energy for Life was held in Clovis, CA in memory of Brittany Wilkinson. The event supports the Brittany Wilkinson Research Fund with the UMDF. Thank you to the Wilkinson Family for keeping Brittany's mission going!

**September 23, 2013.** The third annual "Life for Lila" Golf Classic, supporting the UMDF and presented by UNUM, was held on Monday, September 23, 2013, at The Golf Club at Ballantyne, in Charlotte, NC. More than \$63,000 was raised for the UMDF. Thank you Dan and Eaddy Richardson!

**September 28, 2013.** The Breyton Senn Golf Classic and Charity Dinner was held at Whitefish Lake Golf Course in Pierson, MI to benefit the Breyton Senn Research Fund with the UMDF. The first annual outing raised nearly \$2,000! Congratulations to the winning team Roger & Donnell Ralph and Justin & Heather Ralph! Thank you to all of the supporters who loved Breyton!

**September 28, 2013.** The first annual Hope for Brionna Run/Walk was held at Fairbanks Park in Terre Haute, IN. This successful event raised \$12,000 for the UMDF in honor of Brionna Myers. Thank you to Karen Myers and the Terre Haute community for your support!

**September 29, 2013.** The thirteenth annual Olivia Steele Memorial Golf Outing was held at Chapel Hill Golf Course in Mt. Vernon, OH. Thank you JR Steele for organizing this outing and raising nearly \$1,500 in support of the UMDF.

**October 1, 2013.** Krista De La Vega of Shiloh, IL hosted a Party for a Cure at a 31 Gifts Party. A portion of her sales of product that featured a green ribbon to symbolize mitochondrial disease was donated to the UMDF! Thank you Krista for hosting this event in honor of the Schnitzler family!

**October 8, 2013.** Precision Roofing held its annual Golf Classic at Lakewood Oaks Golf Club in Lee's Summit, MO. The UMDF received a portion of the proceeds from the outing. A special thank you to Deidra Atchley for helping to organize this event!

**October 12, 2013.** The UMDF was the charity of choice for the Glow Run that was held in Lawrence, KS! 700 runners gathered in the evening to light up the city with their bright colors and black lights! Thank you to Kim Johnson for all of your hard work and for connecting the UMDF with the Glow Run!

**October 13, 2013.** Team Hope Energy Life participated in the 2013 Chicago Marathon and collected funds for the UMDF! Thank you to Gary Moberly, Christina Brown, Mariah Rettenmeier, Meghan Rettenmeier, and Patrick Kelley for running for the UMDF!



*Gary Moberly, Meghan Rettenmeier, Mariah Rettenmeier, and Christina Brown of Team Hope Energy Life at the 2013 Chicago Marathon.*

# Energy for Life Walkathons

**August 17, 2013.** The fourth annual Energy for Life Walkathon: Minnesota was held at Normandale Lake Bandshell in Bloomfield, MN. Over 330 participants came together to help raise over \$61,000. Thank you to Kalynn Wendt and Stacey Pieper who have helped plan the walk for the past six years. You will be missed as co-chairs! Also, thank you to the rest of the committee for your help with the walk! We couldn't do it without all of you!



*Mary Beth Towell and Carrie Benner at the Energy for Life Walkathon in Akron, OH.*

**August 24, 2013.** The second annual Energy for Life Walkathon: Akron was held in Akron, OH at the beautiful Lock 3! Thank you to our walk committee and to all of our teams in helping to raise over \$36,000 for the UMDF!

**September 7, 2013.** The second annual Energy for Life Walkathon: Detroit was held at Dodge Park in Sterling Heights, MI. With a move from Ann Arbor this year, over 320 participants attended bringing the walk total to over \$35,000! A big thank you to the Energy for Life Walkathon: Detroit Committee who helped to make this another successful year. We are so grateful for all your hard work!



*Cindy and Zoe Kavan with Zoe's aunt, Sally Brown, at the Energy for Life Walkathon in Kansas City, KS. Photo by Cindy Kavan.*

**September 14, 2013.** The Kansas City Group held an Energy for Life Walkathon at the CommunityAmerica Ballpark (Kansas City T-Bones Stadium) in Kansas City, KS. The walk raised more than \$31,000 for the UMDF. Thank you to the walk committee for all of their hard work!



*Chelsea Loyd and Dr. LoDuca stand behind Noah Ramirez at the Energy for Life Walkathon in Columbus, GA.*

**September 14, 2013.** The First Annual Columbus Energy for Life Walk was held in Columbus, GA at Golden Park Baseball Field/Riverwalk. Families came together for a morning of awareness and raised over \$19,000. Thank you to everyone who helped make the event possible.

**September 15, 2013.** The fourth annual Energy for Life Walkathon: Chicago was held again this year at Katherine Legge Memorial Park in Hinsdale, IL. Even in the rain, over 260 participants endured the weather to help raise over \$54,000! Thank you to our amazing Energy for Life Walkathon: Chicago Committee - you have done a great job!

**September 21, 2013.** The New York Chapter held the fourth annual Energy for Life Walkathon at Cheektowaga Town Park in Cheektowaga, NY. The event raised more than \$27,000. Thank you to the wonderful walk committee for all of your support!



*Attendees at the Energy for Life Walkathon in Cheektowaga, NY.*

**September 21, 2013.** The second annual Energy for Life Walkathon: Central Texas was held at Old Settlers Park in Round Rock, Texas. Attendance was up over 300 participants bringing the walk total to up over \$28,000! Thank you to the walk committee who did an exceptional job. Congratulations on a job well done!



*Mimi's Walk 'N' Rollers at the Energy for Life Walkathon in Round Rock, Texas.*

**September 28, 2013.** The fourth annual Energy for Life Walkathon: Delaware Valley brought together over 400 walkers at Campbell's Field in Camden, NJ and raised over \$33,000! Thank you to our committee, volunteers, and teams who came out to support this great walk!

**September 28, 2013.** For a second year, walkers came out in support of the Energy for Life Walkathon: Milwaukee at Greensfield Park! Teams from all over Wisconsin came together and raised over \$23,000 for the UMDF! Thank you to the walk committee and our wonderful walkers for your support!

**October 12, 2013.** The Carolina Foothills Chapter held an Energy for Life Walkathon in Freedom Park in Charlotte, NC. The event raised more than \$90,000 for the UMDF. Thank you to the amazing walk committee!



*Team Walk for Aiden at the Energy for Life Walkathon in Camden, NJ.*



*Kaden's Kure at the Energy for Life Walkathon in Milwaukee, Wisconsin. Photo by Nic Photography.*



*Ben's Brigade at the Energy for Life Walkathon in Charlotte, NC.*

# Gifts from the Heart

**June 2013.** Autumn Hoekstra's wish for a new swing-set was granted by Make-A-Wish of Iowa and so Autumn's mother, Nickkie Hoekstra of Rock Valley, IA hosted a "Wish Party" and raised \$783. Nickkie raised the money by selling flowers in honor of Autumn and putting out a donation box at the "Wish Party" with UMDF informational materials. Thank you Nickkie and Autumn!

**July 2013.** Sophi Edwards had a lemonade stand at her mom's yard sale in Solsberry, IN. About \$250 was raised towards the Jack Edwards Research Fund. Thank you to the Edwards family for making a difference within the mito community!

**August 2013.** The Mohans held their seventh annual fundraiser in Bronx, NY which honored Michelle Mohan and celebrated her 22nd birthday; the fundraiser was also held in memory of Isabelle "Izzy" Sherman. The event raised \$9,016. A portion of the funds went to the UMDF research fund for Brittany Wilkinson. We are deeply saddened by the passing of

Michelle Mohan on October 6, 2013. Our hearts go out to all of those touched by her beautiful life.

**August 2013.** Matthew Calhoun, from Oxford, GA sold artistic inspirational cards and raised \$100. He was inspired by a local artist at the UMDF Christmas Party two years ago. Thank you Matthew!

**September 2013.** Lillian DeJean, an 11-year-old from Lafayette, LA who is affected by mitochondrial disease, organized a fundraiser called "An Evening of Spoons" based on the "Spoon Theory." Prior to the event, over 100 spoons were donated and Lillian's friends and family decorated them. The spoons were sold at the event, which raised \$1,622. About 50 people attended the event. Thank you Lillian for such a successful and creative event!

**September 2013.** Noah Taylor of East Lansing, MI held a Lacrosse Clinic in honor of his brother Simon Taylor. Thank you so much for raising \$442 and bringing us closer towards a cure!

## Coins for a Cure & Light Bulb Campaigns

All of the people listed below have recently held Coins for a Cure Campaigns.

- Tammy Naranjo – Pueblo, CO
- Lora Pryor – Winnabow, NC
- Shawna Barton – Metairie, LA
- Melissa Newton – Shelbyville, KY
- Sandra Holt-Smith – Oakley, CA
- Kathleen Connors – Hidden Valley Lake, CA
- Vicki Eberly – Blue Ball, PA
- Tina Scanio at Southeaster Fishing Tackle – Tampa, FL

All of the people listed below have recently held Light Bulb Campaigns.

- Nunzio Galipo – Mayfield Heights, OH
- Antoinette Ingrassia – Mayfield Heights, OH
- Tina Scanio at Southeaster Fishing Tackle – Tampa, FL
- Danielle Goldberg – Long Island, NY
- Melissa Newton – Shelbyville, KY
- Melinda Edwards – Bloomfield, IN
- Vicki Eberly – Blue Ball, PA

**Thank you all for your support!**



### Become an Energy for Life Walkathon Volunteer!

*We need you! As we are gearing up for the spring walk season, we are currently looking for enthusiastic volunteers who want to be a part of one of our amazing Energy for Life Walkathon Committees. Volunteering for an Energy for Life Walkathon is a wonderful way to make a difference in the lives of the adults and children who are affected by mitochondrial disease. We can't have successful Energy for Life Walkathons without YOUR help! To see if there is an Energy for Life Walkathon near you, visit [www.energyforlifewalk.org](http://www.energyforlifewalk.org) or contact [events@umdf.org](mailto:events@umdf.org).*

# Upcoming Events to Benefit the UMDF

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**November 3, 2013.** A cut-a-thon will be held at Clyde St. Amand's Hair Design studio in West Hartford, CT. Make a \$25 donation and receive a haircut with all proceeds being donated to the UMDF. For more information, please visit [www.clydestamandshairdesign.com](http://www.clydestamandshairdesign.com).

**November 6, 2013.** Join us for Margaritas for Mito at a fundraising day at Chili's Restaurant in Metairie, LA where 10% of your sales will come to the UMDF! In addition to the fundraising day, the group will be gathering twice that day! Go to [www.umdf.org](http://www.umdf.org) and click on our event calendar to get all of the details and to download the flyer! Contact GiGi Calatayud at [gigicat1@gmail.com](mailto:gigicat1@gmail.com) for more information!

**November 6, 2013.** Fight Mito for Maddix will be holding a fundraising night at Buffalo Wild Wings in Peoria, IL and 15% of the sales will come to the UMDF! You will need a to present a flyer to your server, so please visit our event calendar at [www.umdf.org](http://www.umdf.org) to download your copy! For questions, contact Christine Carter at [ccarter@dentalartslab.com](mailto:ccarter@dentalartslab.com)!

**November 9, 2013.** The DC-Baltimore/Northern Virginia Chapter invite you to the fifth annual Fall Into a Cure Silent Auction & Cocktail Party to be held at Belle Haven Country Club in Alexandria, VA. This elegant evening will feature dinner, amazing silent auction items, dancing and much more. Tickets can be purchased at [www.umdf.org/dcgal](http://www.umdf.org/dcgal) for \$90 per person.

**February 1, 2014.** Join us for the Energy for Life Walkathon: Houston by forming a walk team for your family, company or school! The walk will be held along Allen Parkway at Sam Houston Park in downtown Houston, TX. If you aren't able to join us that day, why not register as a virtual walk team or make a donation to your favorite team! Visit us at [www.energyforlifewalk.org/houston](http://www.energyforlifewalk.org/houston) today!

**Ongoing.** Christina Brown, a UMDF member who ran the Chicago Marathon, will continue to run one marathon per month and continue to raise funds for the UMDF. Upon completion, Christina will run her very first ultra-marathon. Thank you for taking on this challenge on behalf of the UMDF!

**Ongoing.** Stroll for UMDF is a year round fundraiser where participants can "stroll" outside and raise awareness and funds for the UMDF. If you would like to make a donation, please visit [www.umdf.org/stroll](http://www.umdf.org/stroll).

If you are having or have held a fundraising event, or are in need of assistance, we want to talk to you!

Contact the Special Events Department via e-mail at [events@umdf.org](mailto:events@umdf.org) or call them, toll-free, at 1-888-317-UMDF.

## UMDF Welcomes New Board Member

Charles A. Mohan, Jr., UMDF Chief Executive Officer/Executive Director is pleased to announce a new addition to the Board of Trustees: Brent Fields. Fields and his family live in Central Texas where he works as the Chief Executive Officer of Big Brothers Big Sisters (BBBS), a nonprofit mentoring organization that serves over 1,000 youth. Prior to his current role, he was an Administrator in the healthcare arena and then a Vice President with the American Heart Association. He has over 25 years of executive leadership experience in various industries ranging from education and counseling to healthcare and nonprofit services. His educational background includes a Bachelors Degree in Communications, a Masters Degree in Education, a Clinical Residency, and a Certification in Health Promotion Management. Fields currently serves on a number of advisory boards and, in his five years at BBBS, he's led the organization to a position of national recognition, including the honor of National Board of Directors of the Year in 2012. He and his wife, Suzette, have actively supported local UMDF efforts in the greater Austin area for the past couple of years, including their involvement in the local Energy For Life Walk. They have three children, and their youngest, Chloe, has a mitochondrial disease.



*Brent Fields*

## UMDF Member Shares How Gene Sequencing Has Changed Her Family's Life

by Linda Ramsey of East Aurora, New York

I have been involved with the UMDf since my son, Kevin, passed away 16 years ago from an unspecified mitochondrial disorder. At the advice of a good friend, I held a fundraiser in 1998 to not only raise money for the UMDf for research but also to raise awareness of mitochondrial dysfunction within the medical community so other families might find an earlier diagnosis and of course, treatment. Since that time, witnessing the growth of the UMDf and the support it provides to thousands of families, and the medical advances that have been made in research, diagnostics, and treatments of mito has truly been my "silver lining," the name of my Energy for Life Walk team. I am thrilled and inspired by the hundreds of ongoing mitochondrial-related clinical trials, a clear link between mito dysfunction and the more common diseases people are familiar with such as Alzheimer's, Parkinson's and diabetes, the hundreds of mito-docs now in existence, and the Grand Rounds that now take place to inform and teach medical professionals about the diagnosis of mito disorders.

There is one other development, however, that has just recently been of tremendous significance to me and my family. When my son passed away, one of the only things doctors were fairly certain of was the mutation was genetic and specifically on the X chromosome. My older daughter, Kathleen, completely healthy, is now 19 and obviously, I have always been concerned if she, too, is asymptomatic. I am thrilled to announce that with all the research, a genetic test was recently developed that has just diagnosed my family's defective mitochondrial gene. So we now know what to look for in my daughter with a simple blood test!

In 1998, gene sequencing was just a dream. Today it is a reality, identifying areas where mutations occur and targeting potential treatments. The ramifications of tests like this one will affect thousands of families who, like me, never gave up hope. How great is that? The tireless work the UMDf promotes to help find treatments, and ultimately a cure, for mitochondrial dysfunction is momentous and continues to bring Hope, Energy, and Life!



Linda Ramsey with her son, Kevin



Kathleen and Kevin Ramsey

**Would You Like to  
Share a Story?**

**Feel free to submit stories to  
[news@umdf.org](mailto:news@umdf.org) for consideration  
in future UMDf newsletters.**

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## Teen with Mito Wins Title of World Champ

Christopher Adkins of Danville, Illinois, who has mitochondrial disease, recently won World Champion in Taekwondo for Sparring at the ATA World Championship in Little Rock, Arkansas. He is also the 2012 and 2013 Illinois State Champion for Forms and Sparring, as well as the Mid Atlantic 2013 District Champion. The district covers the states of Illinois, Indiana, Michigan, Wisconsin, Ohio, and Kentucky. He competes in the Special Abilities-Physical-Black Belt Division ages 13 to 17. Way to go Christopher!

# Pat and Russ Charleston Meet Mito Researcher

by Pat Charleston of Chicago, Illinois

My daughter, Kristen, was born healthy and became very sick at the age of 5. By Christmas that year, we received her diagnosis of Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like Episodes (MELAS). We never heard of mitochondrial disease so when we got her diagnosis we were devastated. We joined the United Mitochondrial Disease Foundation (UMDF) and joined the University of California San Diego (UCSD) DCA study in 1999. We feel the drug helped keep her lactic acid lower so that helped prevent seizures for awhile.

I attended my first UMDF Symposium in 2000 in San Diego, California. As I was flying home on the plane, I decided that I had to raise awareness and get money for research. I was brainstorming and came up with a fundraising event called, "Kites for Kristen." I launched the event at St. Daniel the Prophet, my children's school. The kids donated money and in return decorated paper kites and won prizes. The class that donated the most money won a hot dog lunch. After several years of doing this, I thought it was time to reach out to other churches in the community. I ended up doing this for 10 years. It was my therapy and my way of helping Kristen and everyone else suffering with mitochondrial disease. Some of the money that I raised went into a research fund in Kristen's name.

I started up the fund so that we could designate where we wanted funds to go. I thought it would be great to see the research work that is being done. Some of Kristen's Research Funds went to Rajesh Ambasadhan, Ph.D., a researcher at Sanford-Burnham Medical Research Institute who is doing stem cell research with skin cells from MELAS patients.



Russ and Pat Charleston are in the front row with Dr. Rajesh Ambasadhan while Russ and Pat's son, Dave Charleston, and Dr. Richard Haas stand behind them.

On October 15, 2013, my husband, Russ, and I flew to San Diego, California, and on October 17, we toured the Sanford-Burnham Institute for Medical Research. We got to meet Dr. Rajesh Ambasadhan and Dr. Stuart Lipton with Sanford-Burnham Medical Research Institute and Dr. Richard Haas from the UCSD. They are a great group, and we learned that the research that is being done is very promising for mitochondrial disorders. In 1998, when Kristen got sick there really wasn't too much out there. I think this stem cell research will really help out one day. I can only hope it will be in my lifetime.

Kristen fought MELAS for 13 yrs. and passed away in August 2011. I promised her that I will continue

to fight for a cure because no family should go through what we had to.

Rajesh Ambasadhan, Ph.D., received a research grant at the UMDF Symposium in June of 2013. Through his research, he hopes to gain insights into mitochondrial dysfunction in MELAS and other mitochondrial diseases.

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## **Edison Updates Patient Community on EPI-743**

In an important development in the progress of EPI-743, officials from Edison Pharmaceuticals have announced that they have successfully recruited all of the patients needed for two of several ongoing clinical trials. According to Matthew Klein, M.D., Edison's Chief Medical Officer, the Mountainview California based pharmaceutical company has enrolled the required amount of patients needed for its double blind placebo controlled trial for Leigh's disease and for a trial for patients with Friedreich's Ataxia. Klein made the announcement during the second in a series of joint conference calls for the patient community sponsored by the UMDF and MitoAction.

"This is a very big step forward as we move to complete these trials and move through the regulatory process", Klein said in making the announcement. Klein said he understands the sacrifices that are made by the patients and their families through their participation in the clinical trial process. They have to "leave home, go to study sites, and fight through all of the very challenging logistics and barriers to get into a trial. And we are truly appreciative of your efforts and support in bringing us to this very successful moment of having fully identified all of the patients for both of these trials." Klein and Edison CEO Guy Miller, M.D., are also very much aware and mindful of the patients who wanted to enroll and those who may not ever have been eligible for these studies. "Here at Edison, we continually review ideas and approaches that we can take to try to get as many of you access to these adequate and well controlled trials as possible."

During the conference call, Dr. Miller reported that Edison has treated over 120 patients with an exceedingly favorable safety profile. The longest participant in the trial recently passed a milestone of four years of cumulative dosing on the drug. Dr. Miller went on to report that the team is highly encouraged with the progress being made with the drug and within the regulatory community. With the progress that is being made, Edison will soon be announcing a natural history study for patients with Leigh's. Such a study will allow Edison to follow Leigh's patients in a regulated and consistent

manner over time. The study will help Edison in the regulatory process required in bringing a drug to market and ultimately to patients. However, participation in a future natural history study does not mean patients will receive EPI-743. It does, however, help with future clinical trials in which patients will receive the drug.

While the Leigh's trial was only open to patients meeting specific age criteria, Klein made it clear that Edison is committed to mitochondrial disease patients of all ages. Klein said Edison will be carefully considering future trials that broaden the enrollment to include patients of all ages, including adult patients.

During the call, Klein announced that Edison will launch a clinical trial for those who suffer from the mitochondrial disease, Pearson's syndrome. He and Dr. Miller anticipate that the study will begin in early 2014. Dr. Miller highlighted some of the other developments underway with EPI-743. He talked about studies currently underway in Rome for patients who have a rare disease closely related to mitochondrial disease. He also announced that Edison is working on a trial sponsored by the National Institutes of Health for children who lack a genetic diagnosis but who appear to have the signs and symptoms of mitochondrial disease. Dr. Miller says the trial is actively recruiting patients and he believes that trial should be underway in the next 30 to 60 days. He also updated listeners on a trial underway for patients with Rett's syndrome, and a trial in Tokyo for patients with MELAS.

"We are very excited about where we are," Dr. Miller said. "We believe we are in the beginning of the final legs of what hopefully will be the first approved drug for inherited respiratory diseases of the mitochondria."

If you would like to listen to the entire conference, visit [www.umdff.org/epi743conferencecall](http://www.umdff.org/epi743conferencecall). For more details about studies underway, visit [www.clinicaltrials.gov](http://www.clinicaltrials.gov) or [www.edisonpharma.com](http://www.edisonpharma.com).

### **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.**

UMDF's intent is to keep you informed. We ask that you always discuss any diagnoses, treatments or medications with your personal physician. UMDF assumes no liability for any information in UMDF Connect.