

UMDF Launches Patient Driven Community Registry

The creation and launch of the Mitochondrial Disease Community Registry (MDCR) marks a significant milestone for those affected by mitochondrial disease. For the first time in the brief known history of mitochondrial disease, our community will be collecting and sharing data in a manner that will positively impact the development of treatments and cures. The registry is meant to be a community asset that all parties interested in improving the lives of patients affected by mitochondrial disease can rally around. The UMDF is committed to long-term financial sponsorship, serving as the guardian of the data, and to overall coordination of the project.

We selected the Genetic Alliance as a partner for this initiative because of their patient-focused philosophy. We share a vision with the Genetic Alliance that registry data belongs to no one other than the person who provided that data, and that a disease advocacy organization such as the UMDF is the ideal steward of such data. Whereas large academic, health and drug development organizations certainly possess at least partially altruistic motivations in addressing mitochondrial disease, a disease advocacy organization is singularly interested in facilitating the most rapid possible development of effective treatments and cures for those affected by mitochondrial disease.

So what is the MDCR, who should register and why is it important? The MDCR will collect patient-centric health data that will be utilized to develop treatments, identify new symptoms (leading to better diagnoses) and provide information to researchers that seek to study mitochondrial disease. The MDCR will also identify new patients in need of support from our community. Our goal is to identify and characterize every person affected by mitochondrial disease, no matter where they are located, living or deceased. Caregivers and family members of those affected, whether themselves affected or not, are also encouraged to register and contribute to the community. Please also consider registering even if you are already part of another database. Each registry has its



own unique utility, and MDCR has been built in a flexible way to assure appropriate sharing of data with other registry efforts.

After registration, you will first create privacy settings that establish 1) who can see the information you provide in an anonymous way (no means of identifying who you are), 2) who can analyze your anonymous data, and 3) who can reach out to you via the registry if they are interested in your participation in a relevant research study or clinical trial. Every registrant is in full control of their privacy settings and can change them

at any time. There are also videos of mito community-active guides available to help you select privacy settings with which you can be comfortable.

Once your privacy settings are established, you will be presented an initial survey that captures diagnostic and demographic information as well as opinions on the MDCR and how it should be used in the future. The MDCR is not simply a contact database, but a tool meant to continually engage the community. Your input and ideas for future survey topics are important.

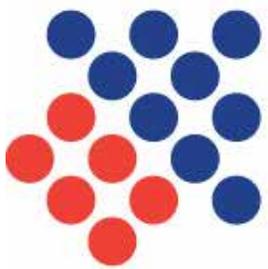
Speaking of important, we should briefly explain why your participation in this registry is VERY important. Researchers need information that can help inform them about the nature and progression of mitochondrial disease. If there is one thing our community can do in honor of those who have already succumbed to this terrible malady, or who are currently living with mitochondrial disease, or for those who will someday be diagnosed with a mitochondrial disease, it is to graciously and openly share our information. If we don't share, the chances of developing treatments and cures are greatly diminished.

The Mitochondrial Disease Community Registry: Our Disease. Your Information. Our Best Hope for Treatments and Cures! To participate in this very important patient driven community registry, please visit www.umdf.org/registry.

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Contents

UMDF News - pages 5 - 7

UMDF Member Stories - pages 8 - 11

UMDF Education - pages 12 - 21

Adult Corner - page 22

UMDF National - pages 24 - 25

UMDF Events - pages 26 -29

UMDF Connect - pages 30 - 31

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From the Chairman

The UMDF received some very exciting news this summer as we gathered for our annual symposium, Mitochondrial Medicine: 2014. We learned that the J. Willard and Alice S. Marriott Foundation awarded the UMDF a five-year grant of \$975,000. This grant was awarded to us because the Marriott Foundation wants to put their support behind our current and future efforts for treatments and cures for mitochondrial disease. This is a significant investment that will help us move forward at an even greater pace than we have been able to achieve in the past several years.

When we applied for this grant, we were able to demonstrate to them that the UMDF is making great strides in enhancing existing collaborations and promoting the sharing of information between the academic, research and clinical communities, Congress and governmental agencies, and pharmaceutical innovators. This improved communication and collaboration will further enhance the development of treatment and cures for mitochondrial disease.

We are also involved with other work of established mitochondrial disease agencies such as the Mayo Clinic bio-repository and the National Institutes of Health (NIH) funded North American Mitochondrial Disease Consortium (NAMDC). These groups are very important because they are working on the critical information process that is needed for clinical trials. We have recently learned of another piece of good news - NAMDC has been awarded an additional NIH five-year funding grant. We were informed that NAMDC's new funding was due in part to UMDF's direct involvement and financial support. In addition, the NIH has decided to allocate more mitochondrial resources within the National Center for Advancing Translational Sciences, which should further our cause.

As we revealed at the symposium, and further outline in this newsletter, the UMDF has created a patient mitochondrial registry. This is a registry for the ENTIRE mitochondrial disease community and is designed to help researchers and drug developers. Additionally, we have helped fund and assist the launch of a new Mitochondrial Sequencing Data



Resource Tool (MSeqDR). This tool in combination with our registry will be a great source of data about our community. With the input from over 100 international mitochondrial disease experts, MSeqDR will create a world-wide genomic data resource that will help diagnose primary mitochondrial diseases and increase the potential for new treatments targeted to precise

disorders.

My term as Chairman of the UMDF Board of Directors has now come to an end. I have been honored to serve as Chairman since 2008 and have enjoyed every day of it. Patrick Kelley has begun his first term as the UMDF Board Chairman, and we wish him much success! But, I am not going away! With my term as chair fulfilled, I will remain on the UMDF board and will serve as Vice Chairman.

In reflecting on my years as Chairman, I had the opportunity to re-read the first message I placed in the Spring 2008 issue of this newsletter. In that message, I expressed my excitement about the many organizational milestones we reached and the evolution of the UMDF. What a difference six years has made! The UMDF is now poised to help facilitate better treatment developments in a shorter time-frame. Our research grant program is rebalancing to provide funding for potential clinical trials. We have developed a patient populated registry that will provide patients who are ready for clinical trials and a potential cure. While those are all exciting and promising developments, I will end this message the same way I ended my first. We have a long way to go, but I sense an intense excitement and enthusiasm because the future is bright for our cause.

Remember - the UMDF's goal is to achieve real treatments and cures that can help real patients. We ARE getting so much closer.

Energy to All,

W. Dan Wright, UMDF Chairman

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8085 Saltsburg Road, Suite 201 | Pittsburgh, PA 15239
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UMDF's Path to A Cure



The United Mitochondrial Disease Foundation (UMDF) is excited to announce the creation of the “UMDF’s Path to A Cure”. The UMDF’s Path to A Cure is a virtual online brick path designed to pave the way for funding real cures for mitochondrial disease. Affected individuals, family members, friends, and coworkers can participate simply by purchasing a brick and placing it on UMDF’s Path to A Cure. All purchased bricks stay on the path forever. You can visit the plan at www.umdopathtoacure.org.

UMDF’S Path to A Cure offers three different types of virtual bricks for purchase:

- The “Hope” brick allows the purchaser to add up to 75 text characters for a \$50 donation.
- The “Energy” brick allows the purchaser to upload a picture (JPEG format no larger than 1 megabyte in

size) and add up to 125 characters of text for a \$100 donation.

- The “Life” brick allows the purchaser to upload a picture (JPEG format no larger than 1 megabyte in size) and a video (uploaded from You Tube or Vimeo and can be any length) and text up to 250 characters for a \$250 donation.

The idea for UMDF’s Path To a Cure came from Marni Falk, MD, Chairman of the UMDF’s Scientific and Medical Advisory Board. Dr. Falk thought it would be a great tool to raise awareness both in and outside of the mitochondrial disease community while raising money to support faster treatments and cures. After months of development and testing, UMDF’s Path To A Cure was launched on July 1, 2014. All costs to create the website were through generous donations made to the UMDF for this specific project.

“UMDF’s Path to a Cure really is a great tool to honor the life or memory of an affected individual,” said Charles A. Mohan, Jr., CEO/Executive Director. “This allows us to expand our message beyond the mito community, because you are able to buy a brick as a birthday gift, for holidays, weddings, or any occasion, really.”

After the brick is purchased, brick owners are able to share a link showing the brick purchase on social media or through email and encourage others to add to their personal path or start a path of their own.

Start your path now or donate a brick to another’s path simply by visiting www.umdopathtoacure.org. For frequently asked questions and instructions, visit www.umd.org/aboutumdopathtoacure.

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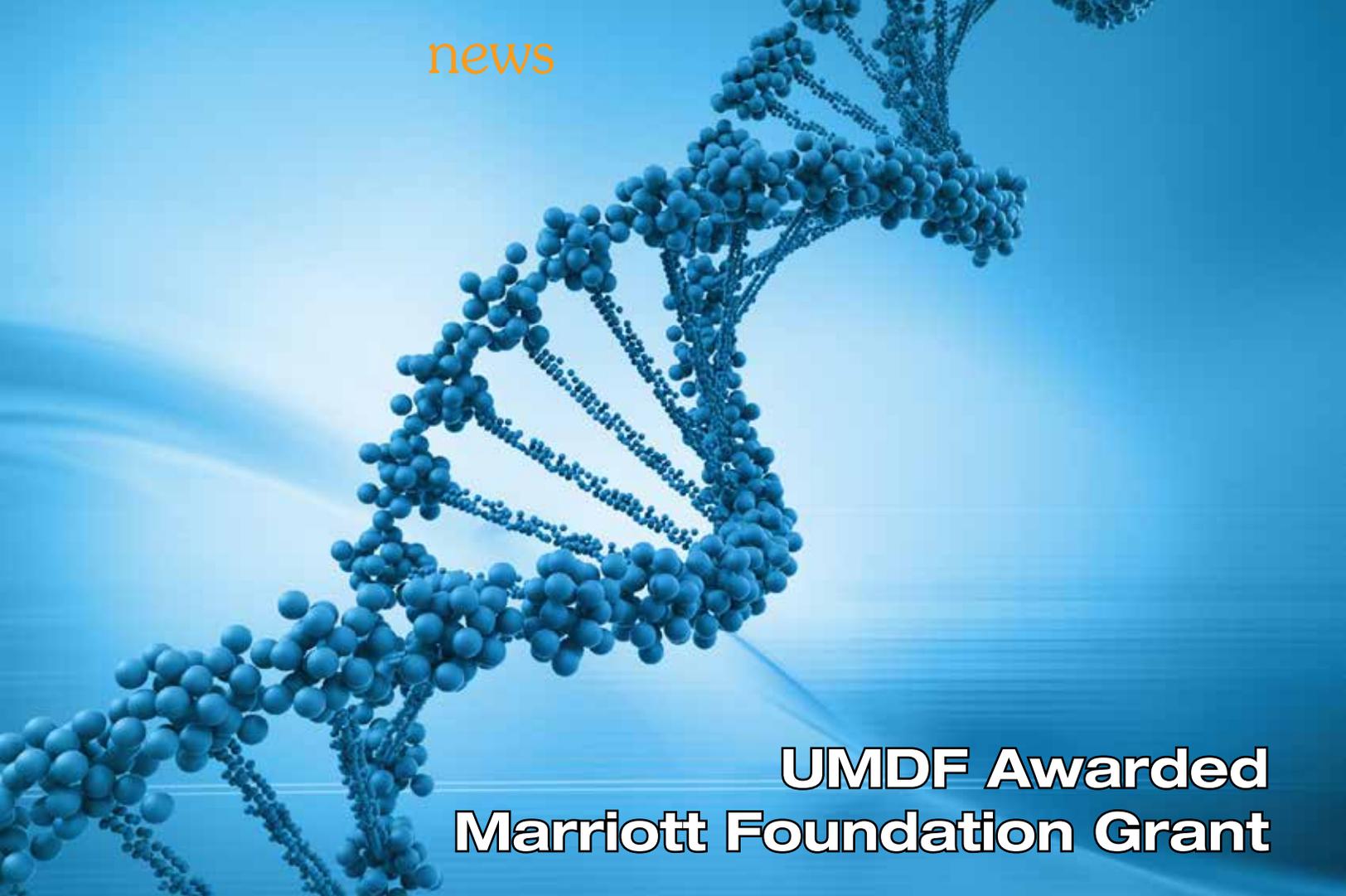


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UMDF Awarded Marriott Foundation Grant

The United Mitochondrial Disease Foundation is honored to report to our members that we have been awarded a nearly \$1 million grant from the J. Willard and Alice S. Marriott Foundation. With the proceeds from the grant, the UMDF will be able to support our Patient-Populated Registry and Biobank Project and support the UMDF Mitochondrial Sequencing Data Resource Tool Project. These elements blend together to create a vision of the UMDF playing a critical central role in coordinating and facilitating the development of treatments and cures for the mitochondrial disease affected community.



“A robust patient registry has been described as the most important asset that a rare disease community can possess in the quest for development of treatments and cures,” said Charles A. Mohan, Jr., CEO and Executive Director of the UMDF. “This UMDF-led initiative would place the UMDF as a patient advocacy group in a stewardship role, casting a wider net over the affected community and helping to ensure maximum patient engagement in upcoming research studies and clinical trials.”

With a resource on staff in the form of the Science & Alliance Officer to take the lead, UMDF has identified through extensive benchmarking of other rare disease patient advocacy groups the strategic importance of initiating our own patient-populated

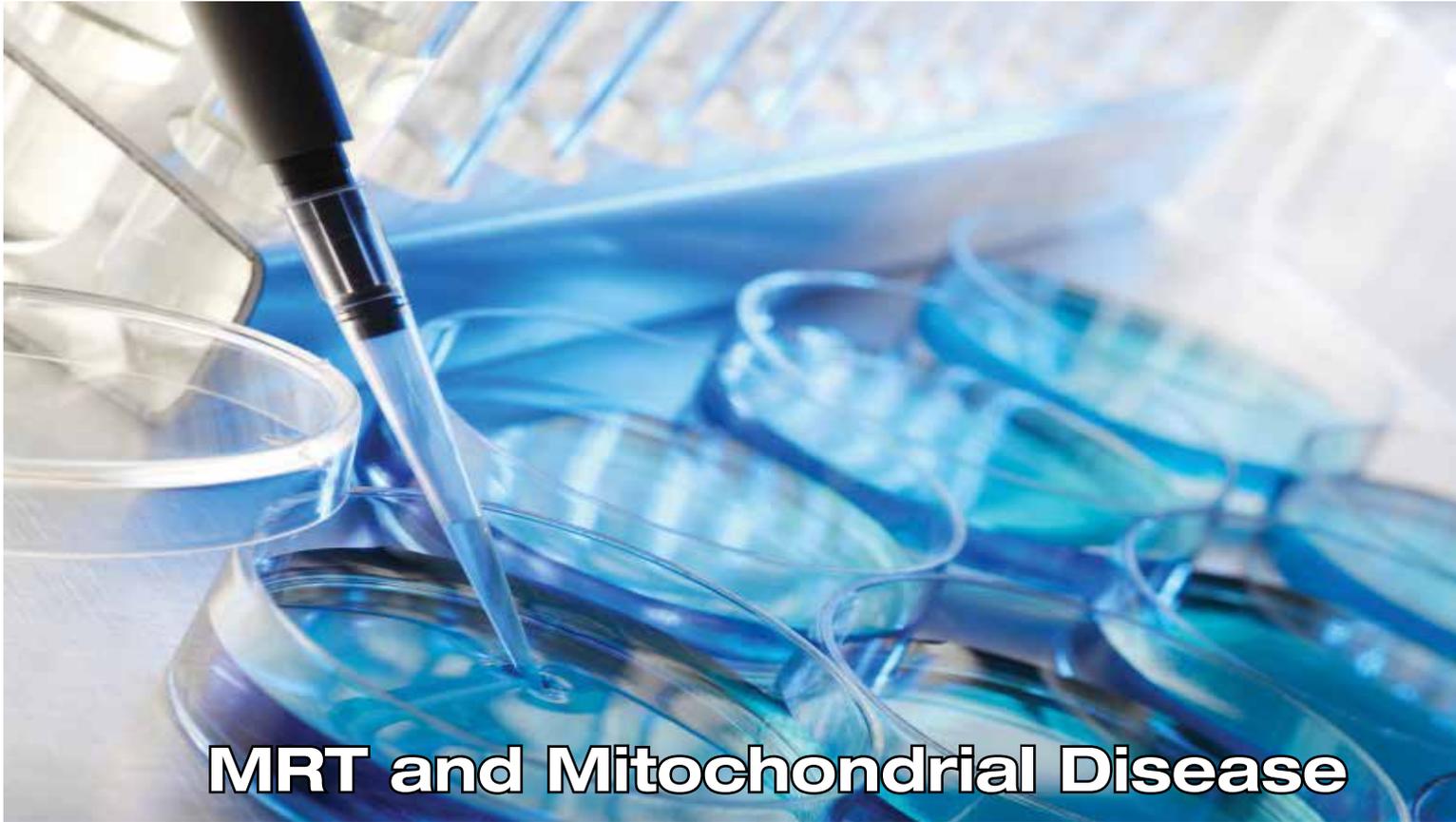
registry to complement the scientifically-populated NAMDC registry.

An equally important element in the overall therapeutic development strategy of the UMDF is the MSeqDR Project, which was initiated at the June 2012 Annual Meeting of the UMDF. This project, which engages and unites more than 100 international mitochondrial disease experts, creates an international genomic data resource for the mitochondrial disease community. It is the interplay of genomic and medical data (such as captured by NAMDC and the UMDF Patient-Populated Registry described

above) that affords the maximum opportunity for researchers to develop treatments and cures.

“The MSeqDR Project increases our ability to diagnose primary mitochondrial diseases and provides greater opportunity to design research projects to understand the mechanisms underlying specific mitochondrial disorders,” Mohan added. “Importantly, this database brings together genomic information that is already being collected in clinics and labs around the world, thus increasing the potential for rapid development of new treatments targeted to precise disorders.”

The grant, which amounts to \$975,000, is payable over five years.



MRT and Mitochondrial Disease

by Phil Yeske, PhD, UMDF Science & Alliance Officer

Recently, the New York Times Magazine published an article entitled “The Brave New World of Three Parent I.V.F.” The article featured in vitro fertilization procedures where a woman’s eggs are extracted, and injected with her husband’s sperm and cytoplasm from another woman’s eggs with the result of a pregnancy following the implantation of the fertilized eggs.

At the same time, the Food and Drug Administration is being asked to allow clinical trials for a new technique that replaces the mitochondria of a mother with the mitochondria of a donor. Researchers are hoping that this ‘mitochondrial replacement therapy’ (MRT) will prevent a variety of diseases caused by mutations in the mitochondrial DNA. Far from creating a “designer baby,” these are breakthroughs that can majorly and joyfully change the quality of life and longevity for a child or adult, much like stem cell and bone marrow transplants have done for those suffering with blood cancers. MRT technology will be beneficial to those who might carry disease, but even more exciting, understanding mitochondrial dysfunction and diseases has the potential to transform medicine and have a much broader impact on human health.

Mitochondria produce the energy needed by the body to sustain life. They are the ‘power plants’ in our cells. Mitochondrial diseases result when there is a defect in energy production. When these power plants fail, organ systems may begin to

decline and the life of the child or adult affected is compromised or ended. Research shows that one in every 4,000 people are born with mitochondrial disease. There are currently no approved medications or cures for mitochondrial disease. The United Mitochondrial Disease Foundation (UMDF) maintains that every individual has the right to safe and effective health care as well as access to all current therapeutic innovations for the alleviation and prevention of mitochondrial diseases. We strongly support further scientific investigation of oocyte MRT as well as constructive debate towards the clinical approval of this therapy in women with mtDNA-related diseases. If demonstrated to be safe and efficacious, this technique should be made available as an option to families who carry mtDNA defects.

Since we all have mitochondria, the UMDF strongly encourages our readers to educate themselves on the role they play in human health and disease. Research increasingly implicates mitochondrial dysfunction as a component in Alzheimer’s disease, Parkinson’s disease, diabetes, hypertension, heart disease, osteoporosis, cancer and the aging process. Understanding mitochondrial disease, even stopping it before it occurs, not only helps those affected by this progressive and often fatal illness, but it can open a new world of medical knowledge. Ultimately, this work can pave the way for developing better diagnostics and treatments for a range of other diseases – alleviating suffering and changing devastating outcomes for the better.

From My Mito Perspective

by Liz Kennerly

I have had mixed experiences when it comes to health professionals. Some doctors are very “black and white” in how they approach a problem. While there are rules and guidelines in medicine, I need doctors who will work with me, because I seem to be atypical in every way. For example, when my body is under serious stress, I have myoclonus in my left arm and right leg—not symmetrical as is usual. After working with me for years, my team has learned to roll with the punches. Recently I asked, “This might seem like a random question, but...?” One of my doctors responded, “Liz, with you I’ve learned that no question is random.” I think I finally have him trained!

There have been times when doctors have gone in circles trying to figure out why my body is such a rebel. I am curious too, but I’m not eager to spend several months on it. In the end, does it really matter when we are successfully treating a specific problem related to Mito? There have also been times when I’ve worked for years on an issue and made no progress. I have had severe spasticity for 13 years, and countless neurologists have analyzed my gait, but none have been successful alleviating this in the long term. After being asked to “walk down the hall and back again” for the billionth time, and doctors recommending meds that would turn me into a zombie, I consulted Orthopedic Surgery. Sometimes the best medical decisions have been those I’ve made myself!

My doctors sometimes make me crazy—but in the end, they are the best, because they truly care about me. Some suggested the possibility of changing my team after I moved home, but it took me five years to build this one. I went to college in Boston and lived there for two years after. I was forced to move in with my parents in Pennsylvania since I couldn’t find a job. While this



would be easier logistically and decrease travel expenses, it could be detrimental in the long run. I would have to teach an entire team my quirks and learn their style of practice. Thankfully, I found a PCP here who is familiar with Mito and likes complex cases. There is a lot of traveling involved to see my specialists in Massachusetts, but at 27, I have no intention of permanently living with my parents.

One of the reasons I like my team so much is that they let me exercise a lot of control. My epileptologist is a great example. He prescribed a medication that, while helpful, proved to be very dangerous for me at its standard dose—it affected my gait so much that I was afraid to be home alone. After a few phone calls, he gave me his blessing to choose my dose, and I haven’t needed him since.

I have also had trouble even getting to see some doctors. Since Mito is most commonly diagnosed during childhood, many specialists equipped to treat it are in pediatrics. A few years ago, I called a Pediatric GI office and was told I was too old to be seen. I called again and only after explaining my diagnosis, was I allowed an appointment. Out of all of my specialists, four of them see both pediatric and adult patients.

My best experiences have been with doctors who are patient, willing to listen, and consider my ideas and opinions. Doctors have a difficult job—they are the professionals and have spent years training; but as patients with a complex disease, we know our own unique body the best because we live it. Especially when dealing with a difficult case such as mine, medical professionals need to be flexible and think in color instead of “black and white”. And as patients, we need to be persistent, yet respectful and inoffensive, until we have built the right team that works for us.

Treating Mitochondrial Disease Through Yoga

by Nalanie Chellaram

I have a beautiful grand-daughter called Talia. She has been diagnosed with a Mitochondrial disease, but to date we have not had a definitive diagnosis as to which type it is.

Talia was born 2kg in weight. From the beginning, my daughter, Shani, suspected something was wrong. None of the pediatricians could give us any information or diagnosis and as we soon found out, very few doctors even knew Mito existed!!

We realized we were in for a long ride. Shani started surfing the internet for any kind of information that would explain Talia's inability to thrive and grow. She couldn't swallow, had reflux, hypertonia and all manner of problems. She could not even sit; forget about walking or talking! We could all feel her beautiful soul, but was she aware of us? Was she suffering? Was she in pain?

Shani decided that whilst all her early morning and late night researching was going on (that is how she found Chuck and the UMDF), that the family should focus on making Talia happy and comfortable. At the age of four months, Talia started her world in physiotherapy.

We watched her improving slowly but surely. It was heart-wrenching as every time Talia would put on a few pounds and finally show some muscle tone, she only had to catch a cold and everything she had put on was lost. What a struggle it was to feed her! This roller coaster ride started to be a constant in our lives...

Being a yoga teacher and trainer, my daughter asked if I would do some yoga exercises with Talia. Initially I was thrilled, but at the same time I was a little apprehensive. I had taught and worked with so many children and adults for over 20 years.

But Talia was so delicate! Putting aside my personal emotions, focusing on the work ahead, following my instincts, we started our first class. What a joy it was! Talia could move much more than

I expected. She loved it and was responding well. Within a few months, together with all the other therapies, Talia started to look stronger. Her spine straightened and she started to become more aware.

The real meaning of yoga is to 'Yoke on to your Higher Self'. In other cultures and languages this 'Self' can be translated to mean God, Universal Consciousness, Unconditional Love etc. So my work was to connect with Talia's soul energy, or what we call prana or chi in Eastern Spirituality. We needed to build up her life force and energy, and awaken blocked nadis (nerves, of which there are over 72,000 in the body).

How does yoga do that?

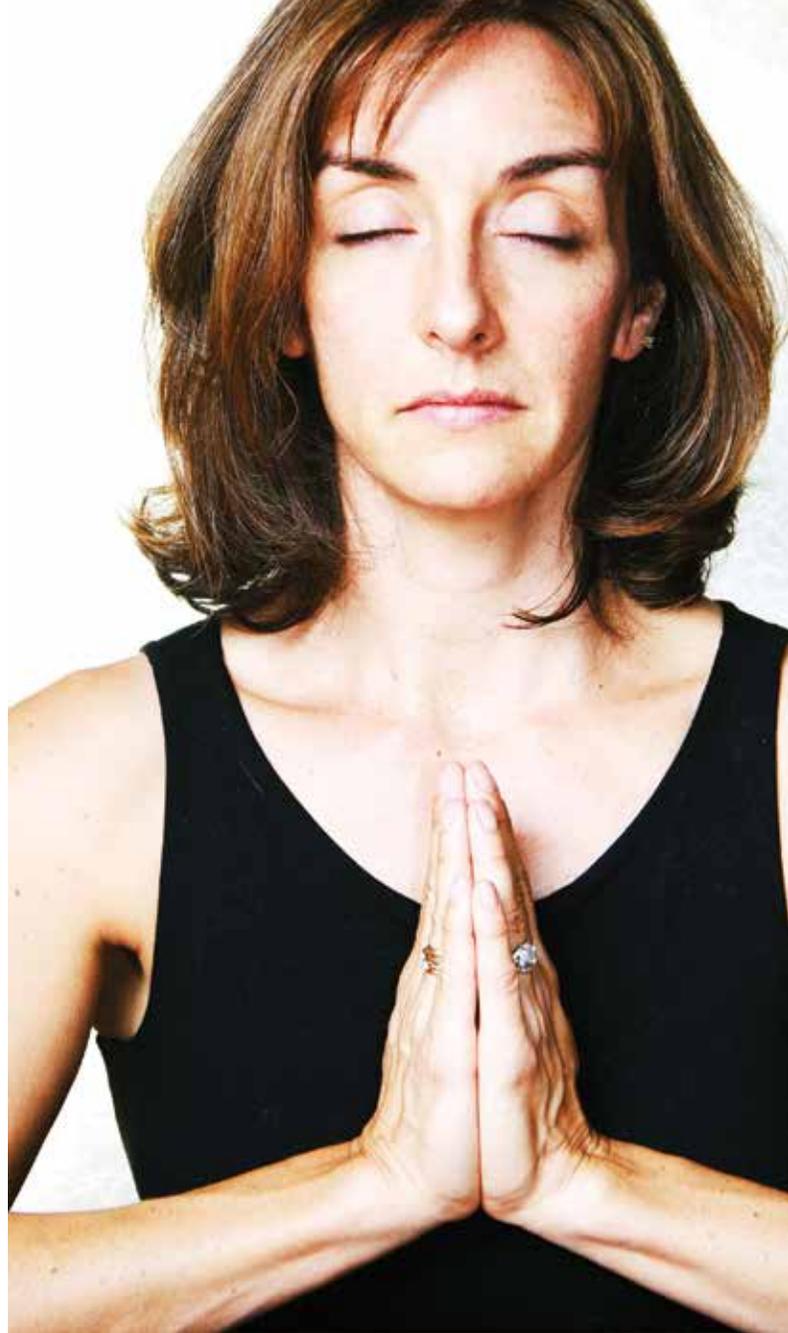
We incorporate four key elements into our routine and these are: sound therapy, physical postures, breathing and deep relaxation.

Chanting

We start our routine with chanting repetitive mantras. Sound, we already know, has an incredible influence on all of us. Play a song that you love and how does it make you feel? The word mantra actually means that which protects and liberates. It also helps a person to focus.

Postures

After the chanting, we apply some physical yoga (Hatha) postures. We go over a series of postures with me guiding her and gently guiding her into different postures. These postures work on different energy centers in our bodies. They also help to stretch and strengthen muscles. By stretching certain areas we are working with nerve centers that may be blocked. In the beginning, every time Talia got into a pose, she would laugh and ask for a hug! She was responding! That really thrilled both of us and encouraged us all to continue.



Breathing

We also work on her breathing. The way we breathe has a great impact on our daily well-being. We used yoga to teach her how to inhale and exhale forcefully and how to breathe through her nose. There is still so much that can be done to help Talia with her Mito through breath control. We are just at the beginning. Not only can breathing properly help us to build up energy and reduce fatigue, but it can help one to focus and can also be used to alleviate pain and control one's body temperature.

Relaxation

Finally we finish the class with deep relaxation. Or at least we attempt to! It is difficult for children to stay still. But more and more, she stays relaxed for longer periods. They say a 20 minute relaxation is the equivalent of seven hours sleep. For Mito kids who need to keep on recharging their batteries, deep relaxation could really help them to get through tough days and help them to build up energy.

Talia is 7 years of age now. She can walk, run, albeit lop-sided, but it is endearing all the same! She can imitate all postures without me constantly manipulating her moves! She loves the chanting, it unwinds her totally. She is a happy child! Yoga works! Yoga has given us a special bond.

More than any therapy, I have learnt that 'Love Heals'. The love that has been showered unconditionally by her mom, dad, siblings, family, teachers, therapists and doctors has given her the 'life force' that she needs in order to live a beautiful life.

My daughter and I are now working towards creating a program called "Yoga for Mito" so we can share this wonderful therapy with as many as possible.

About the Author: Nalanie Chellaram is a modern spiritual teacher, a true Soul Doctor dedicated to selfless service. She was born in 1954 into the distinguished Indian family the Harilela's. She grew up in the presence of a close family friend, who, unbeknown at the time, was to have a deep and lasting influence in her life. This frequent visitor and friend was Swami Satchidananda, her beloved Guru.

Nalanie travels the world to spread a message of wisdom, compassion and love, based on the philosophy of yoga. Nalanie is the founder of the Integral yoga Centre Gibraltar and George Harilela Hall in Sotogrande, she gives Satsangs both there and Internationally.

In March 2008, Nalanie received an Honour in Her Majesty Queen Elizabeth the Second New Years Honour's List. She was recognised for her 'Services to Humanity'. In October she also received a "Humanitarian Award" from the Satchidananda Ashram in Virginia, USA.

In 2009 Nalanie set up an association called "Service in Satchidananda", a non-profit international collective of charities established in honour of Sri Swami Satchidananda and based on his core teaching of selfless service.



Symposium Family Recap

Raise your hand if this is your first UMDF Symposium,” UMDF CEO/Executive Director Charles A. Mohan asked patients and families gathered for the opening meeting of Mitochondrial Medicine 2014: Pittsburgh. As attendees across the crowded ballroom started to put their hands into the air, it was apparent that the majority of those present were attending this annual meeting for the first time. They came from as nearby as the Pittsburgh suburbs to as far away as western Canada. All of them were eager to learn more about their disease, network with each other, and meet with the physicians who were there to help.

“This symposium is unique because you have the opportunity to become empowered by sharing your experiences and having the opportunity to meet some of the top clinical mitochondrial specialists from around the world,” Mohan told those gathered.

For those who are new to the meeting, the UMDF offers patients and families sessions dealing with mitochondrial disease. Patients were also presented important information about the new mitochondrial disease patient registry, an update on clinical trials, and information on transitioning from child to adult care.

UMDF also presented two speakers to

address the issue concerning many of our parents with affected children. Medical Child Abuse has been in the news over the past few months. UMDF has worked with numerous families facing this issue and we thought it best to present information designed to help parents now. One of the topics presented was “Patient Care.... in a World that is Still Learning about Mitochondrial Disease”. This critical information was presented by Dr. Mark Korson of Tufts Medical Center in Boston, MA. UMDF recorded Dr. Korson’s presentation and will make it available to you on the UMDF Website.

Also presenting was Darice Good, J.D. Good’s presentation was titled “Six Degrees of Prevention: “Easing Parents’ Fears of Child Protective Services”. Good outlined the role of child protective services, how they investigate alleged cases, and how parents can ease their own fears and work to prevent such allegations. Her presentation will also be on the UMDF website in the coming months.

Next year, Mitochondrial Medicine 2015 will be held in the Washington D.C. area. The two day family meeting will take place on June 19-20 at the Hyatt in Herndon, VA. On June 18, 2015, the UMDF will conduct its third “Day on the Hill” for patients and families to educate elected officials about our specific needs.



Hoppel Presented UMDF Vanguard Award



Each year, the UMDF presents the Vanguard Award to an individual recognized as being in the forefront of medical or scientific contributions in the field of mitochondrial medicine or research for many years. In 2013, the first ever UMDF Vanguard Award was presented to Salvatore DiMauro, MD, of Columbia University Medical Center.

The 2014 recipient of the UMDF Vanguard Award is **Charles Hoppel, MD**. Dr. Hoppel demonstrates a lifetime commitment of progress towards a cure, whether through research or medical treatment. He diligently served the UMDF on both the Board of Trustees and the Scientific and Medical Advisory

Board. He also provided inspiration to others in an effort to bring new scientific or medical experts into the field of mitochondrial medicine and research.

Dr. Hoppel is Chief of the Division of Clinical Pharmacology at Case Western Reserve University and Associate Director of Research at the Louis Stokes Veteran Affairs Medical Center in Cleveland. His lab does extensive research in mitochondrial disease. Dr. Hoppel has a grandson affected by mitochondrial disease.

It is with extreme honor that the UMDF awards Dr. Charles Hoppel our Vanguard Award for 2014.



Symposium Awards

At its annual symposium, Mitochondrial Medicine 2014: Pittsburgh, the United Mitochondrial Disease Foundation (UMDF) honored several volunteers for their efforts and dedication in supporting the organization and the patients and families it represents.



Devin Shuman, 21, of Seattle, WA, was selected as the 2014 **LEAP Award** winner. LEAP is short for Living, Encouraging, Achieving and Persisting with mitochondrial disease. The award is presented to an individual who is 14 or older living positively with mitochondrial disease. She was chosen for the award from a field of several dozen nominees nationwide.

In order to be selected for the UMDF LEAP Award, an individual must demonstrate a positive attitude, have hope for a brighter future for the mitochondrial disease community, and possess enthusiasm that inspires others.

“Devin’s experience with the UMDF inspired in her a lifelong passion for working with youth with genetic conditions,” said Charles A. Mohan, Jr., CEO and Executive Director of the UMDF. “She is helping others discover the positives that come with a disability community and its identity.”

Shuman first attended a UMDF Symposium in 2011 as a youth ambassador. Since that time, she started a Facebook group for teens and young adults that is a place where they can share support and information. She graduated from Smith College in May and will pursue a degree in genetic counseling.



Karen Ricci, of Akron, OH, is the 2014 UMDF **Energy Award** winner. The award honors an individual who embodies the UMDF mission of promoting research and education for the diagnosis, treatment, and cure of mitochondrial disorders and providing support to affected individuals and families.

Ricci and her husband are the parents of three children. Their youngest, Joey, has mitochondrial disease.

“Karen’s greatest joy is meeting new families to share information, fundraising ideas, talking about their children’s similarities or success stories of things to come,” said Mohan.

Ricci is an active member of the Northeast Ohio UMDF Akron/Cleveland chapter for the past 8 years and serves as a walk committee member and as a co-chair for the EFL walk.

“Karen is a tremendous asset to our families with her wise advice and help. She embodies the energy behind all of our affected individuals and families,” Mohan added.



Jade Thompson, 16, of Crooksville, OH, was named the 2014 UMDF *Heartstrings Award* winner. The award is presented to a child or teen that has donated or raised funds for the UMDF, enabling the UMDF to continue its mission.

In order to be selected for the UMDF Heartstrings Award, an individual must meet the criteria of age, time invested, talents demonstrated, effectiveness, generosity, and how the individual has “tugged at the heartstrings” during their efforts to raise funds for the UMDF.

“Jade has taken on numerous fundraising projects like bake sales, t-shirt sales, volleyball and softball fundraisers,” Mohan said.

“Jade also is very helpful to her mother, Jody, with an annual event entitled ‘Bet on Baylee’ which has raised thousands of dollars for the UMDF.”

Jade conducts all of these fundraisers in honor of her younger sister, Baylee, who battles mitochondrial disease.



Victoria Helms, of St. Louis, MO, was honored with the 2014 UMDF *Stanley A. Davis Leadership Award*.

The award, presented annually, honors a UMDF volunteer leader exemplifies dedication to the UMDF. The award was created seven years ago to honor the work and dedication to the UMDF shown by the late UMDF Board Chairman Stanley A. Davis.



Victoria Helms

Helms is a Licensed Practical Nurse currently working for Washington University in the Otolaryngology clinic at St. Louis Children’s Hospital. She is the mother of two children, Cody, 12 and Branden, 11. Branden is affected by mitochondrial disease.

“Victoria’s children drive her to help find a cure for mitochondrial disease,” said Mohan is presenting the award. “Victoria has been the Support Liaison for the St. Louis Chapter for 4 years and enjoys having the opportunities to meet new families, to share in their journey, and to help along the way when possible,” Mohan added.

In addition to all of her work in support of families, Victoria co-chaired the 4th annual Energy for Life Walk in St. Louis in 2014.

Research Grant Awards

The United Mitochondrial Disease Foundation awarded nearly a half million dollars in research grants at its annual symposium, Mitochondrial Medicine 2014: Pittsburgh. The research grant awards were presented on June 6, 2014. The UMDF has now funded more than \$13,500,000 in research grants aimed at treatments and potential cures. The UMDF remains the largest non-governmental funder of research for mitochondrial disease.



Sean & Kristi Strawser and Dr. Carlos Moraes

Carlos Moraes, PhD, Department of Neurology, University of Miami Miller School of Medicine was awarded \$120,000 for his project, “Developing specific mitochondrial nucleases to eliminate mutant mtDNA.” Dr. Moraes has developed a process for removing disease-causing mitochondrial DNA mutations from affected mitochondria. Extension of this research seems likely to lead to the development of gene therapies for human mitochondrial disease.

The grant was presented by **Sean and Kristi Strawser** of Greenville, OH. **Corynna Strawser** lost her valiant battle with mitochondrial disease at the age of 16, but her vibrant spirit lives on though the **Corynna Strawser Family Research Fund**. Corynna never missed an opportunity to educate and inspire everyone she met. Kristi and Sean are engaged in various fundraising and awareness activities, including co-chairing the inaugural EFL walk in Cincinnati in the spring of 2015. They joined with the **Ayden & Faith Hingsbergen Research Fund** and the **Breyton Senn Research Fund** to support Dr. Moraes this year.



Will & Nicole Dalton and Dr. Francisca Diaz

Francisca Diaz, PhD, Department of Neurology, University of Miami Miller School of Medicine, was the recipient of an \$80,000 UMDF Research Grant. Dr. Diaz’s project is entitled “Modulation of GSK3 activity to maintain neuronal survival in complex IV deficient mouse.” Dr. Diaz is using a much studied mouse model in which a mitochondrial respiratory enzyme has been deactivated in nerve cells. She will study the effectiveness of modulating glucose metabolism as a treatment for these mice, with the potential for extending this therapy to human mitochondrial disease patients.

The grant was presented by **Will & Nicole Dalton** of Franklin, MA on behalf of the **Emma Frances Dalton Family Research Fund**. Their daughter, Emma Frances, lost her battle with mitochondrial disease before her first birthday in 2011. The Daltons and their family and friends have raised nearly \$16,000, including a marathon run managed through the UMDF’s Active-ate your Mitochondria site. Emma’s fund is also currently supporting Dr. Natalie Niemi’s UMDF project at the University of Wisconsin.

Editor’s Note: Special thanks to Steven G. Bassett, PhD, Associate Professor of Biology, Seton Hill University, for developing descriptions for these projects for both this newsletter and at www.umdf.org/fundedprojects



Erin Seifert, PhD, Department of Pathology, Thomas Jefferson University, is the recipient of a \$120,000 UMDF Research Grant. Dr. Seifert's project is entitled "Pathogenesis of myopathies caused by mitochondrial phosphate carrier mutations." Dr. Seifert is studying mutations that severely affect the delivery of phosphate for ATP synthesis in the mitochondria of skeletal muscle and the heart. This foundational research will provide new insights into important mechanisms responsible for mitochondrial disease.

Cheryl Porter & Sebastien Cotte of the Atlanta, Georgia area are currently co-chairs of the Atlanta Energy For Life walk. Over the last three years the Atlanta walk has generated an amazing \$200,000 of support. David Porter's team, "Porter's Pounders" and Jagger Cotte's Team, "Moves Like Jagger" have generated an astounding \$40,000 of donations combined over that time.



Scot Leary, PhD, Department of Biochemistry, University of Saskatchewan, received a \$100,000 UMDF Research Grant. Dr. Leary's project is entitled "Targeted delivery of copper to mitochondria: investigating its therapeutic potential for the effective treatment of patients with mutations in SCO1 and SCO2." Dr. Leary is investigating therapies for copper delivery to mitochondria in patients with impaired ability to synthesize a vital mitochondrial respiratory enzyme that requires copper as a building block. This research could lead to the development of early intervention therapies for mitochondrial disease.



Scot Leary, PhD

Michael James Bell, MD, University of Pittsburgh, was the recipient of a \$25,000 grant award. Dr. Bell's project is entitled "Improving CNS delivery of brain antioxidants after acute metabolic decompensation in mitochondrial disease." Dr. Bell will investigate a combination of two FDA-approved drugs for their effectiveness in treating children and young adults with Leigh's Syndrome. This work has the potential to improve brain function in patients with a mitochondrial disease for which there are currently no proven treatments.



Michael James Bell, MD

Marc and Shelly Hingsbergen, of Cincinnati, OH, presented the grant with their son **Ayden**, 9, and daughter **Faith**, 7 on behalf of the **Ayden and Faith Hingsbergen Family Research Fund**. Their UMDF Family Research Fund has raised over \$43,000 from a wide variety of activities over the years, including Coins for a Cure, Mito Mania and the Southern Ohio Bikers fundraisers. This grant also supported by the **Nicholas Torpey Family Research Fund**.

Hubert Smeets, PhD, Department of Genetics and Cell Biology, Maastricht University, The Netherlands, received a grant award of \$25,000. Dr. Smeets' project is entitled "Development of an autologous myogenic stem cell therapy for carriers of a heteroplasmic mtDNA mutation, a proof of principle study." Dr. Smeets has developed a process using transplantation of a patient's own muscle stem cells that have been freed of mitochondrial DNA mutations. The resulting formation of normal muscle fibers promises to set the stage for significant new therapies for mitochondrial disease.



Hubert Smeets, PhD

The UMDF Research Grants are funded through the work and dedication of UMDF volunteers and donors who support Energy for Life Walkathons and other special events, Holiday Appeals, UMDF's Path to A Cure, and UMDF Family Research Funds. If you would like to learn more about helping further our research with a gift, please contact the UMDF Development Department at 888.317.UMDF (8633).

Mitochondrial Research Thrives in the Steel City

by Steven G. Bassett, PhD

Mitochondrial Medicine: 2014 convened in Pittsburgh, PA in June at the Sheraton Station Square Hotel. Researchers traveled to Pennsylvania from eighteen other states and from fourteen other countries (including Argentina, Lebanon, Portugal, and Russia) to present cutting-edge research on mitochondrial function and disease. A significant number of researchers and clinicians from the Pittsburgh area were also involved. Seventeen Pittsburgh-based researchers were active participants in both the platform presentations and abstracts. This indicates the presence today of a robust community of scientists conducting mitochondrial research in the same geographical area where the UMDF was founded by Chuck Mohan just 19 years ago. The web site for the University of Pittsburgh's Molecular and Cellular Cancer Biology Program states that its researchers conduct "studies of cellular metabolism, apoptosis, and mitochondrial physiology in normal and cancer cells." The Neurogenetics and Metabolism Clinic at Children's Hospital in Pittsburgh "is the region's leading clinic for the diagnosis and treatment of mitochondrial, metabolic, and other neurogenetic disorders in infants, toddlers, adolescents, and young adults." Drs. Jerry Vockley and Amy Goldstein, who co-chaired the Symposium's scientific sessions, are both associated with the Clinic. They invited a cross-section of members of the international mitochondrial research community to share their most recent work. Following are highlights of some of these presentations.

Roles of Mitochondrial Sirtuins

In the opening session, Dr. Vockley stated that the characteristics of a specific mitochondrial disease cannot be explained solely in terms of a genetic mutation but must also take into account post-translational modification. Once the genetic code has been expressed

to manufacture the functional proteins of the mitochondria (a process known as "translation"), one or more enzymes regulate these proteins' functions by modifying the proteins' chemical structures. For example, sirtuins regulate the removal of acetyl groups or other chemicals from mitochondrial proteins. Dr. Matthew Hirschey of Duke University followed up on this idea by discussing the roles of sirtuins in mitochondrial disease. Hyperacetylation of mitochondrial proteins (i.e., addition of too many acetyl groups) can lead to mitochondrial dysfunction, which sirtuins can prevent through their activity as deacetylases. Acetylation of proteins is an essential regulatory component of virtually all known metabolic pathways. A mutated form of a sirtuin known as SIRT3 leads to mitochondrial protein dysfunction. According to Dr. David Lombard of the University of Michigan another sirtuin, SIRT5, has a negative effect on mitochondrial function by removing succinyl groups from at least two important mitochondrial enzymes. Insights provided by these and other researchers into the effects of post-translational modification on mitochondrial function would seem to have great potential for the development of important new drug therapies.

Mitochondria Dysfunction and Neurodegenerative Diseases

Dr. Stephen Archer of Queen's University in Canada described the roles that mitochondrial fission (splitting) and fusion (joining together) play in a number of diseases. Increased fission is associated with diseases such as Huntington's disease and Parkinsonism, neurological disorders that severely impair movement. Mitochondria in the nerve cells of patients with these diseases manifest a fragmented appearance, rather than occurring in fused networks. The research of Dr. Archer and others suggest the possibility of treating

specific diseases by causing fragmented mitochondria to re-fuse. A trio of scientists from the University of Pittsburgh presented insights derived from their research on this topic. Investigations by Dr. Robert Friedlander have demonstrated a specific link between the mutation that causes Huntington's disease and mitochondrial dysfunction. Developing treatments that restore normal protein transport into a patient's mitochondria could help patients with the disease. Dr. Sarah Berman shared extensive evidence for a role played by mitochondrial fission/fusion in Parkinson's disease. Her research is aimed at uncovering the relationship between mitochondrial dynamics and early neurodegeneration during the course of Parkinson's. A third Pitt researcher, Dr. Bennett Van Houten, provided an extensive list of other diseases related to oxidative stress and mitochondrial dysfunction. These include multiple kinds of cancer, heart disease, and at least three other neurodegenerative disorders. One goal of his lab group is to develop a reliable means of preventing oxidative stress from damaging mitochondrial DNA, since this can lead to destruction of mitochondria and the cells that house them.

Animal Models

A session on the utility of animal models for the study of mitochondrial function and disease featured two UMDF research-grant recipients: Dr. Michael Palladino of the University of Pittsburgh and Dr. Gerald Shadel of Yale University. Dr. Palladino has been able to insert genetic information into fruit flies afflicted with a form of mitochondrial disease. This results in a "genetic rescue" of the animals, a finding that could point the way to similar therapies in human patients. Dr. Shadel employs both mouse and yeast research models to study stress signaling pathways in mitochondria and to discover how they are linked to diseases and aging. He



has demonstrated that a combination of inherited, environmental, and age-related factors contribute to impaired energy metabolism in complex ways. Dr. Alicia Pickrell from the National Institute of General Medical Sciences also presented her research on a mouse model for Parkinson's disease. The problem with this disease is that a specific population of neurons loses the ability to make dopamine, a neurotransmitter chemical that is important in brain control of muscle contraction. Her results suggested that the enzyme known as Parkin protects these neurons by regulating mitochondria that possess deleterious mutations. Others seeking to provide animal models that researchers can use to ask questions about mitochondrial disease include Dr. Michael Perez of the University of South Alabama, who has developed mouse cell line models of a number of disease-causing mitochondrial DNA mutations and Dr. Michael Karlsson of Sweden's Lund University, who has developed a pig model that can be used to study pharmacological interventions for mitochondrial Complex I disorders.

Tools For Mitochondrial Research

This session included a masterful presentation by Dr. Chuck Hoppel, recipient of UMDF's 2014 Vanguard Award, on how his clinical lab at Case Western Reserve University provides detailed assessments of patient tissue samples. The lab's approach is to evaluate metabolism at a number of levels, providing an integrated analysis of mitochondrial activity that aids in diagnosis and treatment of patients. Dr. Carla Koehler of UCLA highlighted her research on a mechanism responsible for transporting the correct RNA messages required by mitochondria to synthesize their enzyme components. Understanding how this mechanism operates could also provide a means of deactivating

incorrect RNA messages resulting from mutations in mitochondrial DNA and might lead to development of new treatments. Employing an analytical device known as a LabSphere, Dr. Robert Balaban of the National Heart, Lung, and Blood Institute is able to monitor the concurrent activities of all mitochondrial enzymes in special tissue preparations. Use of this device provides a dynamic approach to studying coordinated function of mitochondria as they carry out the sequential steps of energy metabolism. This is just a small sample of the investigative machinery and protocols available to researchers from a variety of disciplines, all of who are seeking answers to important questions about mitochondrial disease.

New Horizons in Drug Therapy

Dr. Peter Wipf of the University of Pittsburgh presented research on an investigatory drug that appears to have therapeutic effects on a number of conditions by protecting enzymes of the inner mitochondrial membrane. The drug neutralizes electrons escaping from the electron transport chain before they can damage respiratory enzymes and other mitochondrial components. In another mitochondrial disease model employed by Dr. Vishal Gohil of Texas A & M University, copper supplementation restored the activity of cytochrome c oxidase in defective mitochondria. A number of drugs similar to coenzyme Q10 were prepared and tested by Dr. Sidney Hecht of Arizona State University. These analogues suppressed the formation of reactive oxygen species in cultured cells, preventing the extensive damage that they can inflict on mitochondria. Dr. Marni Falk of the Children's Hospital of Philadelphia has demonstrated the beneficial effects of an Epicatechin drug. She was able to significantly increase the lifespan of animals that had a severe mitochondrial Complex I deficiency. Dr.

Carlos Moraes of the University of Miami is a 2014 UMDF research grant recipient and is taking a different approach to treatment of mitochondrial disease. He has developed a means of removing mutated mitochondrial DNA from patient-derived cells. In one experiment, treatment with engineered mitochondrial nucleases led to permanent reductions in these disease-causing mutations. UMDF's funding of this promising research is emblematic of its long-term commitment to pursuing multiple avenues that could lead to a cure.

Additional research on promising drug therapies is being conducted in Japan (Dr. Yatsutoshi Koga of Kurume University: sodium pyruvate corrects chronic lactic acidosis), Switzerland (Dr. Thomas Meier of Santhera Pharmaceuticals: idebenone promotes recovery of vision in LHON patients), Sweden (Dr. Johannes Ehinger of Lund University and NeuroVive Pharmaceuticals: a new class of cell-permeable prodrugs for treatment of mitochondrial Complex I disorders), China (lab of Y. Luo of Sun Yat-Sen University: peptide SS1 is a potential new treatment for diabetic retinopathy through its effects on mitochondrial function), and Detroit (Dr. Hani Sabba of Henry Ford Hospital: Bendavia (Stealth Peptides) is a potential treatment for Barth syndrome).

No one could have predicted in 1995 that in less than two decades so many different research approaches would be answering vital questions about mitochondrial disease and that a number of therapeutic options would already be available, with many more in the pipeline. The Pittsburgh mitochondrial community is proud to be part of the global search for a cure.

Ask the Mito DocSM

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 DocSM at www.umd.org. Please note that information contained in Ask the Mito DocSM 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.

Q: My child has been diagnosed with DGUOK deficiency. Is that a primary mitochondrial disease? Can you tell me something about it?

A: Deoxyguanosine kinase (DGUOK) is a gene found in the regular nuclear gene pool. So, it is not a mitochondrial DNA encoded gene but a nuclear encoded gene found on chromosome 2. The type of disease seen in infants and young children is almost always autosomal recessive. When two mutations happen, one on each allele are present, the patient can have disease. Two mutations must be present and usually this happens when one mutation comes from mother and the other from the biological father. The gene product of the DGUOK gene is an enzyme that is responsible for recycling the building blocks of DNA. This is needed as the mitochondria cannot make its own nucleotides (building blocks) of DNA for the mitochondrial DNA. The building blocks are specific for two nucleotides GMP and AMP (this part is not that important). But without the proper function of this gene, the mitochondria cannot make enough DNA. So there is mitochondrial DNA depletion in tissues.



Russell Saneto, DO, PhD

Most infants will express multiple system dysfunction, but the vast majority will have liver involvement and some neurological involvement. The most pronounced part of this disorder is liver involvement with neurological findings of eye movement abnormalities called nystagmus and opsoclonus. Now depending on the type of mutation, a patient can have severe disease with liver failure, severe hypotonia, developmental stagnation or regression and poor long term prognosis. Other type of mutations (missense) can cause milder disease with much less neurological involvement and liver dysfunction that can be treated and supported. Longer survival in this latter population is common into early teenage ranges and perhaps longer. These patients may also present later in life, but when the disease presents is not as important as whether there is neurological involvement. Early demise of the patient, often in 1 – 2 years from onset in those patients with neurological involvement is most common.

The adults who carry one of the mutations may develop symptoms later in life, but not always. Likely, this depends on mutation type and location. - *Russell Saneto, DO, PhD*

Q: I came across some info about MELAS stroke-like episodes being treated with taurine. I found articles online saying taurine deficiency symptoms are almost identical to MELAS symptoms, just by Googling taurine deficiency and MELAS. Another article said two patients taking high dose taurine had no stroke-like episodes for 9 years. That is the best news I have read recently, so I had to do some digging. I found a 2013 clinical trial in Japan recruiting MELAS patients for a taurine study. Do you (or anyone at UMDF) know anything about this? I emailed the researcher to ask for the dosage and he wrote me back with it. Also, do you know anyone with MELAS who has tried stem cell therapy? I am going to see a stem cell doctor at UCSD cancer center very soon.

A: Taurine is not yet an accepted therapy - it may function as an anti-oxidant but we know far more about COQ10+alpha lipid acid + vitamin E + creatine in actual patients in an RCT showing a lowering of oxidative stress than the scant literature on taurine. Stem cells are not a therapy for mitochondrial disease except maybe in Pearson syndrome and MNGIE but these are VERY specific indications, and side effects of a stem cell transplant are major.



Mark Tarnopolsky, MD

- *Mark Tarnopolsky, MD*

Q: My daughter is 6 years old. For about 3 years now she has been having “attacks” of stomach pain and vomiting. Recently she also started having motility issues and soiling herself. We have had a GI work up done and it shows she has no GI problems. My father has MELAS and the doctor mentioned to us our daughter could have a mitochondrial disease as well. She had a recent Carnitine level of 12. Is it possible for my daughter to have this since it was my father who is the one with MELAS? And can she present with mostly GI symptoms? Is the carnitine level alone enough to go on? She had a normal lactate and normal pyruvate level.

A: It's important to know how your father's MELAS was diagnosed. Was it definitively confirmed by a known mtDNA mutation? Or it was MELAS-like syndrome actually caused by a nuclear gene? Since your daughter would get it from you, do you have any symptoms? The chances are extremely low that mtDNA mutation can be passed from the father but it's not zero since there are some reports of inherited paternal mtDNA (probably from a minimal retention of sperm mtDNA or "paternal leakage" when it penetrates the egg while in the vast majority of cases, only 23 nuclear chromosomes from sperm are retained and the rest is extruded). But the simplest thing to do would be to test your father for a genetic cause of his MELAS, then test you and then your daughter. However, your daughter may very well have a different etiology of her GI problems whether mito or non-mito. She could have another metabolic disorder that could give low carnitine but it's important to look at plasma actylcarnitines, urine organic acids, carnitine and actylglycines. Neurologic causes need to be ruled out and consultation with neurologist could be of benefit. - *Dmitriy Niyazov, MD*



Dmitriy M. Niyazov, MD

Q: My 4 year old daughter has presented with persistent hematuria and the reason is unknown. I was wondering if there could be a mito relation to this and what that would be?

A: In general, persistent hematuria is an unusual finding with respect to mitochondrial disease. On the other hand, kidney involvement, including renal tubular acidosis, is relatively common. It would be important to know the type of hematuria, i.e. microscopic or macroscopic, and to have a thorough investigation by nephrology and urology in this case. There have been reported cases of mitochondrial disorders being associated with a rare type of kidney disease called focal segmental glomerulosclerosis. However, a mitochondrial connection with hematuria is rarely made. - *Greg Enns, MD, ChB*



Greg Enns, MD, ChB

Q: I was looking on your website and found prednisone listed under medications that may be helpful for mitochondrial disease. There were no details given regarding this and I was wondering if you had any further info. I suffer from MELAS (main symptoms are gastroparesis, migraines, muscle pains and fatigue) as well as arthritis and have taken prednisolone (which I think is the same as prednisone) for my joints. When I take it I tend to notice a huge improvement in all my symptoms. My MELAS symptoms have been deteriorating over the last couple of years now and I'm fairly desperate for anything to improve the situation. I've tried the prednisolone for the last couple of weeks and am feeling better on it, but have been warned it can be detrimental to people with mito disease. Any advice/info you can give me would be greatly appreciated.

A: The question of using steroids in mitochondrial patients is complex. An article in Neurologist 2012 May 18(3):159-70 describes a patient with the 3243 MELAS mutation who presented with recurring episodes of aphasia, hemianopsia and other symptoms. MRI imaging during these episodes noted that neuronal loss during acute episodes occurred in regions of the brain with increased cerebral blood flow. The patient was treated with high dose intravenous corticosteroids, resulting in marked and sustained clinical improvement. It was hypothesized that the use of the steroids reduced the inflammatory mediators and blood brain barrier dysfunction that leads to hyperperfusion in MELAS and the secondary cascade of events that results in neuronal death. This clearly demonstrates that under these circumstances steroids can be useful in MELAS patients. However, to the contrary, there have been reports of deterioration in patients with Kearns Sayre Syndrome on steroid therapy and I have seen rhabdomyolysis [rapid breakdown of muscle fibers] occur in several of my own patients with fatty acid oxidation defects treated with steroids. Long term benefit of treatment remains unclear. - *Fran D. Kendall, MD*



Fran D. Kendall, MD

You can quickly and easily find "Ask the Mito Doc" Q/As on topics of your choice by going to the UMDF home page at www.umdff.org. Go to "Find Support" and click on "Ask the Mito Doc." Click on the search "Ask the Mito Doc" link; this will pull up a search box. Type in a keyword or phrase you are interested in and click "Go." The search engine will pull up every Q/A that mentions your word or phrase. If you are not satisfied with the results, try variations or synonyms of your word/phrase.

An Appeal to the Mitochondrial Community....

Hello, my name is Gail Wehling, and over thirty-five years ago I was diagnosed with Chronic Progressive External Ophthalmoplegia (CPEO) a form of Kearns Sayre Syndrome (KSS).

I first became involved with UMDF over ten years ago and currently have the honor of serving on the UMDF Chicago Chapter and as the co-chair of the UMDF Adult Advisory Council Team (AACT) whose purpose is to represent, serve, and support the adult mitochondrial community.

In June 2011, the UMDF held its 13th annual International Symposium: Mitochondrial Medicine Chicago. Hundreds of physicians, scientists and researchers along with affected individuals and families gathered together to attend family, clinical, and scientific sessions.

For the first time at the UMDF Symposium, Friday afternoon brought both the scientific community and patients/families together for one very important session - clinical trials. The session and topics ranged from working with the FDA in developing new therapies, to a family's perspective on clinical trials. It was a dynamic afternoon, and a call to all of us - patients, families, physicians, scientists, and researchers - to become involved in clinical trials or studies.

Shortly after this exciting afternoon, I began to inform and educate myself about current mitochondrial-related clinical trials and studies. After in-depth study and timing, in 2013 I was accepted into Dr. Ronald Haller's Exercise Study being conducted at Texas Health Presbyterian Hospital of Dallas.

I am excited and feel privileged to be a part of this very important

research. I am committed 100% to the study and to following its protocols and guidelines. How I may benefit from the study is secondary to me than what may be gained from the study as a whole. It has been a great experience with Dr. Haller and his outstanding team - a true partnership with sharing of information and open lines of communication at any time. I look forward to the second phase of the study and where it may lead me. Realistically, it is unlikely cures will be discovered during my lifetime. However, my great hope until breakthroughs happen are that these first vital clinical trials and studies can help all who are affected by this disease, especially mito-kids so they do not have to follow in the same footsteps as many of us adults.



Phil Wyrick, Exercise Physiologist; Gail Wehling; Marta Newby, Research Nurse at the Texas Health Presbyterian Hospital of Dallas Neuromuscular Center Institute for Exercise & Environmental Medicine, March 2014

Currently, there are over 300 mitochondrial-related clinical trials and studies ongoing worldwide. Over these past ten years, I am often asked, "How can I help?" Well, I cannot think of a more important and pivotal time than now. And, to paraphrase Dr. Peter Stacpoole during the clinical trials session, "No patients, no trials, no treatments, no cures," says it all.

Simply put, it begins with us and it is now up to us, the patient and family communities, who have battled this disease for so long to take the first step. Otherwise, we cannot reach our ultimate goal - cures for mitochondrial diseases.

So, please take the first step and go to the UMDF website at www.umdf.org/clinicaltrials to read about current clinical trials and studies seeking participants, and to learn more about the newly created mitochondrial disease patient registry discussed in this newsletter at www.umdf.org/registry. We are in this fight together. Now, please become a part of the path toward its cure!

Thank you.

Adult Advisory Council Team (AACT)

Jennifer Schwartzott, AACT Chair, NY
Gail Wehling, AACT Co-chair, Illinois
Bob Brief, New York
Linda Cooper, California
Whit Davis, Pennsylvania
Kirah Fasano, Pennsylvania
Rev. David Hamm, Maryland

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Debra Makowski, Arizona
David McNees, Ohio
Sharon Shaw Reeder, California
Gregory Yellen, Maryland

Medical Advisors:

Bruce H. Cohen, MD

Amy Goldstein, MD

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 and evaluate, provide advice and guidance, and make recommendations to UMDF on adult-related issues.



Stealth BioTherapeutics

Stealth BioTherapeutics, formerly known as Stealth Peptides, is a leading-edge biotechnology company developing first-in-class mitochondrial-targeted compounds. The company's lead candidates are Bendavia™ and Ocuvia™, which target disease via mitoprotection—the ability to preserve and restore energy production in mitochondria while decreasing oxidative stress.

Mitochondria are the cell's source of energy; in the normal process of generating energy, all mitochondria produce a low level of reactive oxygen species (ROS)—a by-product of cellular respiration and a type of oxidative stress. When mitochondria do not function properly, the result is reduced energy supply and increased oxidative stress. The excess ROS that unhealthy

mitochondria produce can damage cells, including DNA, proteins, lipids and the mitochondria themselves. In many diseases, dysfunctional mitochondria are the main source of oxidative stress, which may lead to inflammation and other abnormal states that can cause serious harm to the body's organs.

Bendavia™ and Ocuvia™ are the first and only therapies under development designed to specifically treat the underlying defect in the mitochondria to reduce oxidative stress and increase energy supply to affected cells and organs. Unlike any other medicines, Bendavia™ and Ocuvia™ reach the inner membrane of mitochondria, where they enhance mitochondria's natural function—the production of energy via the electron transport chain. Within the inner membrane of the mitochondria, Bendavia™ and Ocuvia™ interact with a lipid called cardiolipin, which is found

only in mitochondria and is critical for maintaining healthy cellular function and energy supply. In multiple disease models, the interaction of Bendavia™ and Ocuvia™ with cardiolipin promotes normal energy production and reduces the creation of excess ROS by mitochondria. By restoring normal mitochondrial function, Bendavia™ and Ocuvia™ may help to protect organs from oxidative damage caused by excess ROS production.

This promising therapeutic platform is supported by more than 100 independent, peer-reviewed publications and presentations, and has the potential to address a wide variety of diseases with unmet treatment needs, including heart failure, diabetic macular edema, skeletal muscle disorders as well as orphan mitochondrial diseases.



Stealth

Stealth BioTherapeutics strongly believes in the potential of Bendavia and Ocuvia to deliver new treatment solutions to patients. Our platform is being developed for mitochondrial diseases where there are no FDA-approved treatments. The company is working with advocacy groups, including UMDF, and is constantly seeking insights from those impacted by these conditions, to ensure that the patient's voice guides the development of these new treatments.

At Stealth BioTherapeutics, our work is personal, and our team is committed to developing new treatments for millions of patients in need.

UMDF national

News from the national office.

Welcome Crissy!

The UMDF is happy to announce that Crissy Eastin is now the UMDF's Central Region Coordinator. Eastin replaces Cassie Franklin, who has been promoted to Gift Officer with the UMDF.

"My only objective is to help and educate others about mitochondrial disease and its impact on families in the central region," said Eastin, a New Braunfels, Texas, resident.

Eastin has a background in fundraising around Texas with Big Brothers & Big Sisters and other youth and family oriented groups. The mother of two says her favorite quote is by Anne Frank – "No one has ever become poor by giving."

Please join us in welcoming Crissy to the UMDF!



Congratulations Nicole!

The UMDF is also proud to announce Nicole Shanter has been promoted to Northeast Regional Coordinator. Nicole will oversee activity in Pennsylvania, Virginia, West Virginia, Delaware, Maryland, New York, New Jersey and the District of Columbia. Nicole brings much valuable experience in working with our membership, supporting the Energy for Life Walks in many

capacities, and grant writing for a variety of UMDF projects, including our national symposium. The UMDF is confident that Nicole's initiative, experience and dedication to the mission will translate into much success growing all support, education and fundraising activities in the Northeast Region.

UNITED MITOCHONDRIAL DISEASE FOUNDATION STAFF

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Regional Coordinator - Northeast

Anne Simonsen

Regional Coordinator - Great Lakes



**UNITED
MITOCHONDRIAL
DISEASE
FOUNDATION®**

HOPE. ENERGY. LIFE.

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.

Update on the UMDF Board of Directors

The United Mitochondrial Disease Foundation's Board of Directors welcomes two new members to the board. Starting his first, three-year term on July 1, 2014, is John Kieffer of Austin, TX.

Kieffer is a sales and business executive with the 3M Company and has held numerous leadership roles in the US, Asia, and Europe throughout his 30-year career at 3M. Recently, Kieffer has been leading 3M's customer-facing "Business Transformation" program, which is a global project to move all of 3M's customers and employees to a unified set of customer engagement and order management business processes/systems.

John and his wife Andrea have two grown children, Matt and Melissa, both in their 20's. Melissa has been recently diagnosed with a mitochondrial disease (MELAS). John, Andrea, and Melissa have been active in the Minnesota Chapter of the UMDF. They recently relocated from St. Paul, Minnesota to Austin, Texas.

Sharon Shaw Reeder will rejoin the board of trustees as Member Liaison. Sharon Reeder was diagnosed with mitochondrial disease in 1999 and currently resides in Aliso Viejo, California, with her husband and son. She has a background in meeting planning, special events, business strategy, and marketing. Sharon has assisted the UMDF in many ways over the years, including helping the UMDF to obtain the recent Jack Black public service announcements that can be viewed on the UMDF's website.

Shaw Reeder previously served on the UMDF's Board of Trustees for over a decade. She served as Vice Chairman on the Board of Trustees for the four years, and she first began serving on the Board as chapter representative. In addition, Sharon Reeder has previously held the position of president of the UMDF's Southern California Chapter. She served as Chairman of the UMDF Adult Advisory Council Team (AACT) and was currently appointed onto the FDA's first Mitochondrial Patient Advisory Committee. Sharon keeps her mental focus positive by helping UMDF in their mission to educate, raise money for research. She lives by motto, that "being of service and part of the solution, is the best medicine to navigate the ravishes of Mitochondrial Disease

Shaw Reeder replaces Norma Gibson of Ukiah, California. Gibson ended her two-year term as Chapter Liaison on June 30, 2014. Gibson joined the UMDF in 1997, along with her daughter, Heidi Marie Daniel. Heidi, who lost her battle with mitochondrial disease in 2001, was an inspiration to everyone she met and she worked very hard to educate the public and the medical community about mitochondrial disease. Throughout Heidi's life, Gibson supported her dedication to making a difference and now Gibson continues her passion every single day. Gibson has volunteered her time and energy in various activities such as the symposium and the Energy for Life Walk in San Francisco. She continues to be a vital part of the UMDF family.



Members of the UMDF Board of Directors: John Kieffer (above), Sharon Shaw Reeder (below) and Norma Gibson (bottom).



Fundraisers Benefitting the UMDF

March 11, 2014 - Best Deal! Auto Sales hosts a contest every month where \$1000 is donated to the charity that receives the most online votes. Thank you Mindy Stump for accepting this check for the UMDF in honor of Sarah Landrigan!

March 15, 2014 - A group of runners participated in the Shamrock 8k and half marathon in Virginia Beach, VA, with the goal of raising money in honor of Claire Williams. Thank you Mia Ronson for organizing this event that raised nearly \$800.00.

March 15, 2014 – The Wilkinson Family organized the 9th annual UMDF Dinner and Silent Auction in memory of Brittany Wilkinson. The annual event has benefitted the Brittany Wilkinson Research Fund for nearly a decade. Thanks to Linda and family for your hard work and dedication to continuing Brittany’s passion.

March 22, 2014 - The fifth annual Jackson-Culley MitoWhat 5k was held in Millington, TN, at USA Stadium. A big congrats to Angie Nunn and Cindy Kraft for organizing this race which has raised over \$100,000 for the UMDF over the past five years.

March 22, 2014 – The first EFL Walk in Tampa Bay was a smashing success! The walk brought over 600 walkers and raised over \$62,000! A very special thank you to all of our committee members, volunteers and team leaders on a great first year!!

March 29, 2014 – EFL: Dallas – The inaugural Energy for Life Walk was held at

Downtown Dallas Garland Square. Over 550 participants gathered together to help raise almost \$64,000! Congratulations on an amazing first year, Dallas!

March 29, 2014 – EFL: Atlanta – The 3rd annual walk was held this year at the Georgia World Congress Center due to rain. Over 400 participants came together to “dance the rain away” at the indoor EFL Party. Over \$48,000 was raised! Great job, Atlanta!

March 29, 2014 – EFL: St. Louis – The 4th Annual Energy for Life Walk was held again this year at Tower Grove Park. A chilly morning brought together over 24 teams with over 620 participants, a few 4 legged friends and 1 Mighty Mito Turtle. Together, they fought for mitochondrial disease by raising \$47,000! Wonderful job, St. Louis!!

March 29, 2014 – Roughly 100 people participated in the first ever Road Rally benefiting the Nicholas James Torpey Research Fund at Roger’s Roost in Sterling Heights, MI! The teams were sent out to complete as many of the 30 assigned tasks within an hour period! The event raised over \$1,500 for the UMDF and was able to spread invaluable awareness! Thank you to the Torpey Family for your hard work!

March 29, 2014 - A basketball benefit was held at New Life Christina Church in Warsaw, IN.

March 30, 2014 - Sydney Mason of North Bellmore, NY, held a skate-a-thon for her Bat Mitzvah and donated all proceeds to the UMDF in honor of Hayley Lieb. Thank

you for organizing this fundraiser and raising \$4,700!

April 2014– The Jupiter First Church Preschool held a ‘Coins for a Cure’ drive in honor of Charlie Dunn in Jupiter, FL. Over \$275 was raised by the students ranging from 2 ½ to 5 years old! A very special thank you to our future philanthropists and to their parents! Way to go!

April 2014 – The Minnechaug Regional High School Honor Society held their annual Basket Raffle in support of the UMDF! The students have held the Basket Raffle for nine years and were able to raise nearly \$5,000 this year! A special thank you to the students and advisors for your support!

April 2014 – Jessica Ross held a fundraising event at her school in Iowa in honor of Westley Clapp and herself! The event raised over \$265 for the UMDF!! Thank you Jessica, you’re amazing!!

April 5, 2014 – The 11th Bet on Baylee event was held in Crooksville, OH, in honor of Baylee Thompson! Friends and family of Baylee came together to enjoy a fun filled day of Texas Hold’em, silent & live auction, dancing, music and more! A special thanks to Jody Thompson for all of your work and dedication to the UMDF!

April 5, 2014 – EFL: Nashville – The Energy for Life Walkathon in Nashville gathered at Centennial Park for their 4th year! Over 200 walkers came out in support and raised over \$31,000! Well done, Nashville!!

events

April 12, 2014 – EFL: San Francisco – The 3rd Annual Energy for Life Walk in San Francisco was held at Golden Gate Park on yet another beautiful day! Over 300 walkers came out in support of the walk, including a few virtual walkers and teams from Southern California! Together they were able to raise over \$63,000! Congratulations California on another successful year!

April 13, 2014 - Bruster's of Ingomar on Perry Highway in Pittsburgh, PA, hosted their annual Easter Egg Hunt and donated \$68 to the UMDF. A special thank you to Bruster's for continuing to fundraise and plan events to reach the UMDF's mission!

April 24, 2014 - Bethany Stamper of Creston, OH hosted a four day fundraising event at Cleat's Pub in Wadsworth, OH. The event consisted of a silent auction, poker run, amazing race, and restaurant night, all to benefit Kaidon Andrew Stamper's Research fund.

April 26, 2014 – EFL: Pittsburgh – The 3rd Annual Energy for Life Walk was held in the Steel City on the beautiful North Shore, Heinz Field Lawn. 400 participants, including 1 giant green M&M, walked to support the UMDF. Congratulations, Pittsburgh on raising \$65,000!

April 26, 2014 – EFL: Binghamton – The 3rd Annual Energy for Life Walk was held at Otsiningo Park. Even though it was chilly and slightly rainy, the community of Binghamton gathered together, 150 strong to raise just over \$6,300. Although small, they are mighty! Congratulations, Binghamton!

April 26, 2014 – Girlfriend's Journey to a Cure was held in Cordele, GA, in honor of Katerina Clarey.

May 2014 – Jill Hannagan, along with the Marlett Children, raised money during a concert for The Love of Lucy Fund. Thank you Jill and Marlett's for your support of the UMDF in Memory of Lucy.

May 4, 2014 - Cadie Wolf of Lake City, MN, sold muffins and baked goods at the 100 mile garage sale event in order to raise funds in honor of her brother, Nathan Wolf. Thank you Cadie for raising over \$600!

May 4, 2014 – Team Unstoppable Nina participated in the Boulder Spring Half Marathon in Boulder, CO, in honor of 2 year old, Nina Hall. Nina's parents, Amy & Michael, along with friends and families participated in the event and fundraised using the UMDF's *Active-ate Your Mitochondria* site. Unstoppable Nina raised an astounding \$37,000 for the UMDF – yes, Nina's crew is Unstoppable!

May 10, 2014 – EFL: Evansville – The 3rd Annual EFL Walk was held at Burdette Park. 27 teams joined together to raise almost \$43,000 for the UMDF, \$8,000 over their goal! Co-Chairs Melissa Edmondson and Lori Meyer, along with their amazing committee, worked to get over 500 participants, 11 sponsors and helped to spread awareness in Evansville with ads, news reports and interviews. Thanks to Melissa & Lori for putting in over three years on the walk, you have been amazing!

May 17, 2014 – The 5th annual Breyton Senn 5K Run/Walk/Stroll was held in Howard City, MI, in support of the Breyton Senn Research Fund. The Senn family holds the annual event in memory of Breyton. Thanks to the supporters who helped celebrate Breyton's 6th birthday.

May 31, 2014 – The Green Carpet HOPE Event was held in Bonita Springs, FL, in support of Makenzie Lawrey's Million Dollar goal! Makenzie wrote the *Mighty Mito Superhero* book in honor of her brother, Gavin. This event featured a beautiful photo gallery, dinner, silent auctions, raffles and a special book signing by Makenzie. Together with Butterflies of Hope, the event helped Makenzie get closer to her goal! Thank you to everyone who helped make the event a success!

June 7, 2014 – EFL: Iowa – The EFL walk was held again this year at Zsavooz. The event raised over \$2,500 with over 70 participants in the small town of Cedar Falls, IA. A big THANK YOU goes out to Ronda Eick, co-chair of the event!

June 7, 2014 - The eighth annual Greater Mito Open was held at Broadlands Golf Course in North Prairie, WI. Approximately \$1700 was raised for the UMDF. Thank you to all the families who helped make the outing a success!

June 14, 2014 – The second annual Nicholas James Torpey Memorial Golf Outing was held in Macomb, MI. The annual event raised over \$21,000 in support of the Nicholas James Torpey Research Fund with the UMDF. A very special thank you to Jennifer Ruhana for organizing the "Butterfly Classic!"

June 27, 2014 - The second annual Thomas' Golf for a Cure outing was held in West Bridgewater, MA, in honor of Thomas Schmid. In addition to the outing, there was a silent auction, raffle and cash prizes to the top three teams. A special thank you to Jason Schmid for organizing this wonderful event!

June 2014 – The Freshmen at Waldwick High School worked on a Global Empathy Project that consisted of researching global issues and taking action to raise awareness. The students then initiated fundraisers throughout the school and their community. The UMDF was chosen by the students to support! Thank you, Waldwick!

June 2014 – Shelly Hingsbergen of Cincinnati, OH, and friends participated in a Cold Water Challenge and raised \$100 for the Ayden and Faith Research Fund. Shelly also spreads awareness through her community and her Senator. Thank you, Shelly, for your support of the UMDF!

June 2014 – Mel Reifsteck of Hartsburg, Missouri, held a bake sale/raffle for the UMDF in honor of her nephew, Connor. Mel and many of her great friends baked muffins, iced cakes, collected donations and volunteered their time for the event. Thank you ladies for a job well done!

June 2014 – Kelsea Manly of The Villages, Florida, will be participating in the Bank of America Chicago Marathon this fall. Kelsea has chosen the UMDF as her charity to honor the memory of Dalton Benjamin. The Villages Employee Softball League held a fundraiser for the UMDF. Thank you Kelsea and Good Luck this fall!

July 26, 2014 – The Carter Buffum 5k was held at Cascades Park in Jackson, MI, in support of the Carter Buffum Research Fund with the UMDF. The event featured a 5K walk/run with glowing accessories.

UMDF Upcoming Events

March 15, 2014 Ongoing – Jackie & Simon Scarr have pledged to participate in 4 races each year in memory of their daughter, Sophia Elizabeth Scarr, lovingly known as Sparkle. The Scarrs will be participating as team Sweat for Sparkle in 4 runs this year (one for each month that Sophia was alive). To support their efforts, visit www.crowdrise.com/sweatforsparkle.

July – August 2014 – Hairdressers United at Level Spa Salon will be hosting a fundraising event at their salon in Cumming, GA. The salon sold coupons for haircuts and will be having a 50/50 raffle at their salon until August 14. Contact info@levelspasalon.com for more details.

Summer 2014 – Team Indiana Wrestlers will be fundraising to ‘Takedown Mito’ at all of their summer wrestling tournaments. Team Indiana will donate \$1 for every takedown during their summer travel tournaments.

August 2, 2014 – The annual Run4Raley event will be held at the Philo Ball Park in Philo, IL. The annual event is held in honor of Raley Kirby and features a 5k run, 1 mile walk and a kids run! For more information, please visit www.umdf.org/run4rale.

August 9, 2014 – Faith McColl will be hosting her annual UMDF Lemonade Stand in Marietta, GA, from 10am – Noon. Faith hosts this annual fundraiser in honor of her battle with mitochondrial disease. This year, Faith will be sure to hit the \$10k mark with her outstanding Lemonade! For more information, contact Faith’s mom, Ann, at ann.mccoll@gmail.com.

August 9, 2014 – Team ‘Slippery When Wet’ will be participating in the Mudathon-Cincinnati in Hamilton, OH in memory of Leslie Witt-Williams. Get muddy in support of the UMDF by going to www.mudathlon.com/events_Cincinnati and joining team ‘Slippery When Wet’. Contact David Murphy at murphydavid@live.com with any questions.

August 23, 2014 – Support the UMDF by purchasing a \$5.00 coupon to Macy’s Shop for a Cause. By purchasing a shopping pass, you support the UMDF while enjoying a day of spectacular discounts. Shopping passes are available for sale at www.umdf.org/macysshopforacause.

August 23, 2014 – Party for Mitochondrial Disease Research is the perfect party – there’s all you can eat pizza and wings and all you can drink beer, wine and pop, plus raffle baskets and silent auction items! Join us at Snyder Bar and Grill in Buffalo, NY to support the John Geraci Research Fund with the UMDF. Register today at www.umdf.org/mitoresearchfr or email Kate at k8craw4d@gmail.com with questions!

August 30, 2014 – The Relaxed Atmosphere All Star Event Car Show will take place at USA Stadium in Millington, TN. A portion of the proceeds from the car show will be donated to the 2014 charity of choice, UMDF.

September 7, 2014 – Matthew Bain of team Unstoppable Nina will be participating in the Chicago Half Marathon and fundraising using the UMDF’s Active-ate Your Mitochondria site at www.umdf.org/activemito. Please support Matthew by searching for his name and making a donation today!

September 13, 2014 – The first annual Fight Mito for Maddix Energy Walk will be held at Coal Miners Park in Perkin, IL. Join us in our fight by going to www.umdf.org/fightmitoformaddix and registering today!

September 19, 2014 – The Carlos Alberto Memorial Golf Outing will be held at Gem City Golf Club in Fairborn, OH, in memory of Carlos Alberto. For more information, please contact Cristina Rue at cristinarue@gmail.com.

September 20, 2014 – The Samuel Cutliff Research Fund will be hosting their second annual Mitochondrial Disease Awareness Walk at Anmed Health Campus in Anderson, SC. Please contact Mary Elisabeth at cutlifcrew6@gmail.com for more information!

September 20, 2014 – The 8th annual Mito Bowl will be held at Big Al’s in Meridian, ID! Register online at www.umdf.org/energybowl for a fun night of bowling with the family! We hope to see you there for fun and surprises!

September 20, 2014 – The Jaxon’s Warriors 5K and Little Warrior Fun Run will be held at Highland Church of Christ

in Robinson, IL, in support of the Jaxon Sharma Research Fund with the UMDF. Join us as we run in memory of Jaxon – go to www.umdf.org/jaxonswarriors5k to register today!

September 20, 2014 – Joanne Kovac-Roberts will be participating in the Redman Triathlon in Oklahoma City, OK in honor of Bennett Hanneman. Joanne is fundraising through the UMDF’s Active-ate your Mitochondria site at www.umdf.org/activemito. Please consider supporting Joanne today!

October 5, 2014 – A glow in the dark 5K run/walk, Sine on Corynna, will take place at Darke County Fairgrounds in Greenview, OH. This fun filled event will be held in support of the Corynna Strawser Research Fund with the UMDF. The fund was set up in memory of Corynna and her courageous battle with mitochondrial disease. For more information, contact Kristi at kstrawser77@gmail.com.

October 12, 2014 – Team Hope Energy Life will be participating in the Bank of America Chicago Marathon in support of the UMDF, who is a proved charity. If you are already registered for the Bank of America Chicago Marathon, please consider joining our fundraising team! Show your support at www.umdf.org/chicagomarathon! Go Team Hope Energy Life!!

October 25, 2014 – The Bruster’s of Ingomar will be hosting their annual Halloween Costume contest to benefit the UMDF in Ingomar, PA. Come in your best costume and enter to win! Check out the event flyer on our UMDF calendar of events!

November 1, 2014 – The 2nd annual Bowl for Mito will be held at Pleasant Hill Bowling Lanes in Wilmington, DE from 2:00-4:00pm. Email Corrie Scarberry at cs3178@msn.com for more information!

November 15, 2014 – Fall Into a Cure Wine Tasting will be held at Breaux Vineyard in Purcellville, VA. Check our event calendar at www.umdf.org for more details on this new and exciting event!



UNITED MITOCHONDRIAL DISEASE FOUNDATION

energy for life
walkathon®

Upcoming Energy For Life Walkathons

August 16 Minnesota – The 5th Annual Energy for Life Walk will be held at Normandale Lake Bandshell in Bloomington, MN for another record breaking year! Join us at the Lake on August 16th!

www.energyforlifewalk.org/minnesota

September 6 Detroit – The 3rd Annual Energy for Life Walk will be held again this year at Dodge Park in Sterling Heights. Join us as we move one step closer to a cure! Sign up your team now!

www.energyforlifewalk.org/detroit

September 13 Indianapolis – The 4th Annual Energy for Life Walk has moved to the fall and at a new location!! Join us this fall at Hummel Park in Plainfield, IN. We are gearing up for a fun filled day at a new location! Sign up to join in the fun at

www.energyforlifewalk.org/indianapolis

September 13 Kansas City – The 5th Annual EFL will be held at TBones Stadium, CommunityAmerica Ballpark in Kansas City. Join us this year for an amazing morning. Let's hit this one OUT OF THE BALLPARK!

www.energyforlifewalk.org/kansascity

September 20 Columbus, GA – The 2nd Annual Energy for Life Walk will be held at the Golden Park Baseball Field/Riverwalk again this year. Help us beat our 1st year total by signing up today to walk!

www.energyforlifewalk.org/columbusga

September 20 New Orleans – Join us for our first EFL Walkathon at LaSalle Park in Metairie, LA. We are excited to have our very first EFL in Louisiana and hope you will be able to join us for a fun-filled day!

www.energyforlifewalk.org/neworleans

September 20 Omaha – Energy for Life is coming to Nebraska! The first EFL Walkathon: Omaha will be held at Miller's Landing. It is easy to be a part of this exciting new event. Start a team, join a team or support the walk!

www.energyforlifewalk.org/omaha

September 20 Western NY – Happy 5th Anniversary to the Energy for Life Walk in Western NY! Join us for this milestone event at the Cheektowaga Town Park. Don't delay, sign up today and register your team!

www.energyforlifewalk.org/westernny

September 21 Chicago – The 5th Annual Energy for Life Walk will be held at Katherine Legge Memorial Park in Hinsdale, IL. Join us as we celebrate five years strong as an Energy for Life Walk and over ten years as a chapter! Sign up today at:

www.energyforlifewalk.org/chicago

September 27 Delaware Valley – The 5th Annual Energy for Life Walkathon: Delaware Valley will be held at Campbell's Field in Camden, NJ. Join us as we walk along the beautiful Delaware River and enjoy a day at the ball park!

www.energyforlifewalk.org/delval

September 27 Milwaukee – Join the Southern Wisconsin group for our 3rd Energy for Life Walkathon: Milwaukee at Greenfield Park (Site 3A) in Milwaukee, WI. Start a team, join your favorite team or just come out and walk with us! Get started today at:

www.energyforlifewalk.org/milwaukee

October 4 Akron – We are gearing up for the 3rd annual Energy for Life Walkathon: Akron at Lock 3 in Akron, OH! This fun filled day won't be the same without you! So get your team signed up today at:

www.energyforlifewalk.org/akron

October 18 Charlotte – It's our 5th year of fun! Come on out to Freedom Park, Charlotte, NC on October 18th and celebrate with us! Won't you walk with us to honor all those affected? Join us by registering your team. We hope to see you there!

www.energyforlifewalk.org/charlotte

October 18 Birmingham – Hello, Birmingham, welcome to your very 1st Energy for Life Walkathon! Come see what all the buzz is about at Railroad Park. Sign your team up today and help bring mitochondrial disease awareness to Birmingham!

www.energyforlifewalk.org/birmingham

October 19 Central Texas – The 3rd Annual EFL Walk will be held at Old Settler's Park in Round Rock, TX. Due to construction, the walk was moved to Sunday, October 19. Registration begins at 11:30 am, the walk starts at 12:30pm. Thank you for your patience during the park renovation.

www.energyforlifewalk.org/centraltexas

Become an Energy for Life Walkathon Volunteer!

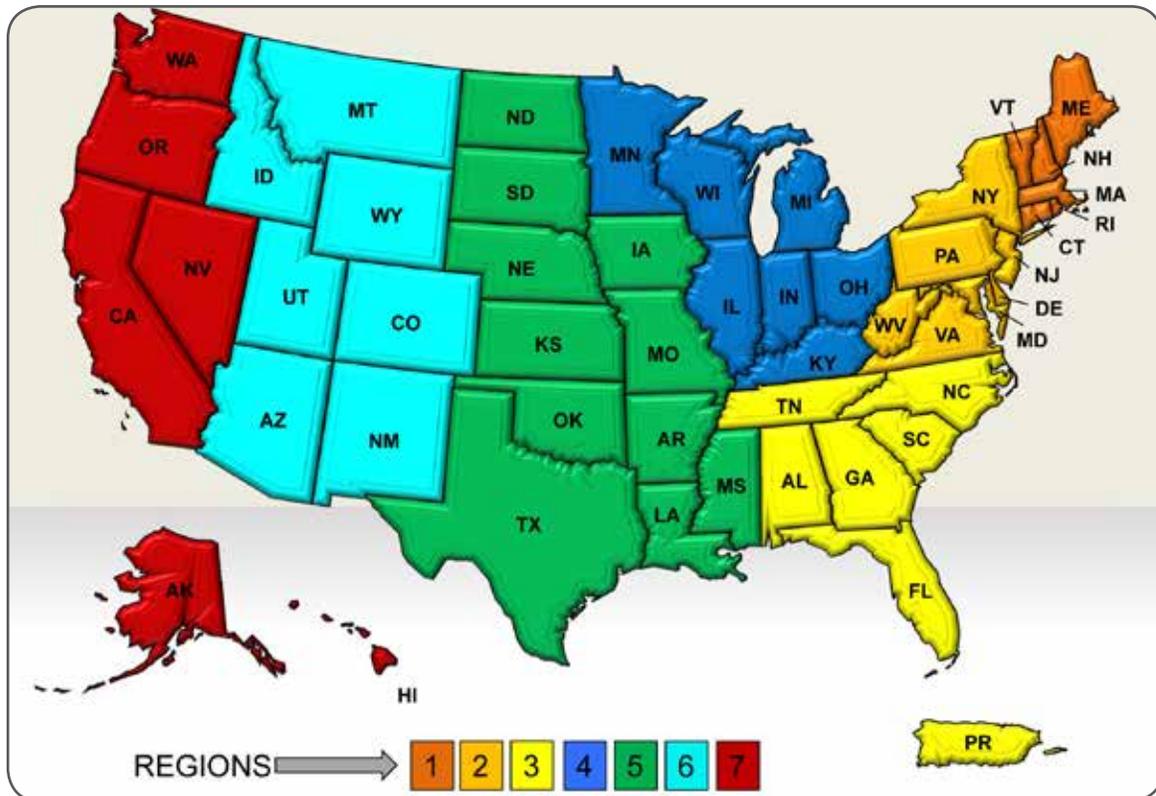
We need you! As we are gearing up for the spring walk season, we are currently looking for enthusiastic volunteers who want to be a part of one of our amazing Energy for Life Walkathon Committees. Volunteering for an Energy for Life Walkathon is a wonderful way to make a difference in the lives of the 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 or contact events@umdf.org.

UMDF connect

Resources for help & support.



In addition to the support staff at the UMDF National Office, help is available to you nationwide 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. When sending an e-mail, please include the leader's name or city/state in the subject line for us to best serve your needs. Interested in getting involved? Call 1-888-317-8633 or visit www.umdf.org/volunteeropps!



New England (1)

MAINE

- Amber Taylor, Bangor

VERMONT

- MaryBeth LeFevre

CONNECTICUT, NEW HAMPSHIRE, RHODE ISLAND

- Contact the National Office to Connect

MASSACHUSETTS

- Julie Gortze, North Attleboro, MA

Northeast (2)

DELAWARE

- Kathleen Stapleford, Magnolia
- Judy Weeks, Dover

MARYLAND

- Dawn Murphy, DC/Baltimore/
Northern Virginia Chapter

NEW JERSEY

- Laurel Smith, Delaware Valley Chapter
- Carrie Mullin, Pittsgrove, NJ

NEW YORK

- Kim Zuzzolo, NY Metro Chapter
- Linda Roesch, Buffalo, Western NY SG
- Jennifer Schwartzoff, Buffalo, Western NY SG
- Erica Beyea, Buffalo, Western NY Parents SG
- Sandy Sallaj, Buffalo, Western NY Parents SG
- Jacqueline Perrotta, Albany
- Lori Piccirilli, Binghamton
- Kimberly Dedrick, Utica

PENNSYLVANIA

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

VIRGINIA & WASHINGTON DC

- Heather Meyer, Lynchburg
- Judi Bartle, Central Virginia SG
- Sharon Hoffert, Central Virginia SG
- Sharon Goldin, DC/Baltimore/Northern Virginia Chapter
- Anne Tuccillo, DC/Baltimore/Northern Virginia Chapter

WEST VIRGINIA

Contact the National Office to Connect

Southeast (3)

Margaret Moore, Regional Coordinator

ALABAMA

- Marissa Benjamin, Leeds

FLORIDA

- Amber Ferrell, Gainesville, Central FL Mito Group
- 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

GEORGIA

- Amy Blackwell, Kennesaw
- Hannah Bossie, Athens
- Sebastien Cotte, Atlanta
- Mary Beth Morris, Atlanta
- Cheryl Porter, Atlanta
- Gail LaFramboise, West Central Georgia
- Shelly Lorenzen, Sugar Hill
- Wendy Clegg, Loyd, Columbus
- Tiffany Tuggle, Stockbridge
- Erin Willis, Valdosta

NORTH CAROLINA

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

SOUTH CAROLINA

- Christine Golden, Greenville/Spartanburg
- Hillary Miller, Charleston
- Karis Mott, Chapin

TENNESSEE

- Emily Culley, Memphis Area SG
- Karrie LaCroix, Memphis area
- Courtney Fellers, Nashville area
- Nancy Garrison, Nashville
- Brandalyn Henderson, Nashville
- Nancy and Jeffrey Rubio, Knoxville

Great Lakes & Midwest (4)

Anne Simonsen, Regional Coordinator

ILLINOIS

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

INDIANA

- Melissa Edmondson, Evansville Area
- Jamie Sterchi, Evansville Area SG
- Kaitlin Thompson, Evansville Area SG
- 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 SG
- Julie Scott, East Michigan/Detroit Area SG
- Genevieve Angeloff, Upper Peninsula
- Holly Worden, West Michigan/Grand Rapids

MINNESOTA

- Stacey Pieper, Minneapolis/St. Paul
- Atom Wolff, Minneapolis/St. Paul

OHIO

- Ruth Gerke, Central Area/Columbus
- Jody Thompson, Central Area/Columbus
- Darcy Zehe, Northeast Area/Akron, Cleveland
- Chris & Alisa Rawski, Northwest Area/ Toledo

WISCONSIN

- Mindy Welhouse, Central Area/Appleton, Green Bay
- Terilyn Musser, Central Area/Eau Claire

- Jaqueline Bohne, Northern WI/Rhineland, Tomahawk, Minocqua Area
- Josh Bartz, Southeast Area/Madison
- Karen Loftus, Southeast Area/Milwaukee

Central Region (5)

Cassie Franklin, Regional Coordinator

ARKANSAS

- Lacie Moore, Rogers

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- Ronda Eick, Northern Iowa
- Kim Novy, Des Moines SG

KANSAS

- Anne Tramposh, Kansas City Area

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- Nicole DeJean, Lafayette
- Rhonda Mailhos, New Orleans SG
- 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 SG

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- 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

OKLAHOMA

Contact the National Office to connect

West (6)

ARIZONA

- Marin Pelletier, Phoenix

IDAHO

- Jennifer Pfefferle, Boise, Idaho SG

NEW MEXICO

- Stephanie Cassady, Albuquerque

UTAH

- Laura McCluskey, Orem

COLORADO & WYOMING

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Pacific and Northwest (7)

CALIFORNIA

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

HAWAII

- Kimo Phan, Honolulu

OREGON

- Kimberli Freiling, Monmouth SG

WASHINGTON

- Mareesa Henderson, Spokane
- Joy Krumdiack, NW Washington

ALASKA & NEVADA

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- 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

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Read-a-thon Raises Awareness



In early January, Jordan Widis, a seventh grader from Alpharetta, Georgia, had to decide upon a charitable project to complete for his upcoming bar mitzvah. After struggling for several hours about what to do, he realized that he could join two things together that he really enjoys...reading and his neighbor and close friend, Maya, who has mitochondrial disease. He decided that he could host a "one-man" read-a-thon and ask for people to sponsor him while he read for eighteen (which means "life" in Judaism) hours during a specific time period in the month of March, above and beyond any reading he had to complete for school. He felt that asking people to sponsor him would be a good way to raise awareness for mitochondrial disease and the money could help the researchers and scientists who work with mito, to help find a cure. Jordan was able to raise over \$600 from his "read-a-thon" and completed all eighteen hours of the reading. He will continue to spread the word about mitochondrial disease and help to make a better "life" for those afflicted with the disease.



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