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UNITED MITOCHONDRIAL DISEASE FOUNDATION®

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UMDF Congressional Briefing Features Discussion on EPI-743 and FDA Approval Process

UMDF CEO Chuck Mohan addresses the attendees of the Congressional Mitochondrial Disease Caucus in Washington, DC.

With representation from a dozen congressional offices, the Congressional Mitochondrial Disease Caucus held a Capitol Hill briefing entitled *"Mitochondrial Drug Development and a Primer on the FDA Drug Approval Process."* The meeting, convened at the request of Rep. Anna Eshoo (D-CA-19) and Rep. Tim Murphy (R-PA-18), enabled the UMDF to educate congress on the exciting developments with EPI-743 and on the Food and Drug Administration (FDA) approval process for similar drugs.

Speaking at the March 13, 2014, briefing was Guy Miller, MD, PhD, CEO of Edison Pharmaceuticals of Mountain View, CA. Dr. Miller briefed those in attendance about the process involved in developing EPI-743, current clinical trials and the pathway his company traveled through the FDA. Also speaking at the briefing was Michael Werner, J.D. Werner, who was recently named "Top 50 Global Stem Cell Influencers," educated attendees about the process of developing and bringing new drugs to market. This information is critical for healthcare staffers to understand as more drugs are being approved, playing a vital role in healthcare and rare diseases.

The UMDF, through Rep. Eshoo and Rep. Murphy, schedules briefings four times a year in order to continue educating members of congress about mitochondrial disease and issues that affect the mitochondrial disease community.

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Contents

From the Chairman

Each spring hundreds of cardboard packages and envelopes are delivered to the UMDF offices in Pittsburgh. It is an exciting time, because we hope that the start for a cure and possible treatments to mitochondrial disease lies somewhere in one of those manila envelopes or boxes. Contained in these packages are the detailed research proposals that are coming from scientists and researchers who have been invited to submit for research funding. Each proposal will then be rigorously reviewed by a person on the UMDF Grant Review Committee. This year, the committee will review about 50 proposals.

In April, the UMDF Grant Review Committee will gather in Pittsburgh to review and discuss all of these proposals and the reviewers' comments. Based on that discussion, all of the proposals will be ranked. UMDF will then award grants for the research projects that receive the top scores and are designed to bring us closer to a cure. The total number of grants, of course, will have to be restricted by the amount of funding available; this is why every donation and each and every walk are so vitally important. The grant process is a very intense activity and designed to be able to provide the most research with the resources available.

But there needs to be even more – Specifically, UMDF is increasing the amount of activity we spend in therapeutic development. In order to make progress and have access to new drugs, we need YOUR help. Without a robust patient registry, participation in biobanks and other tools we have available, clinical trials and drug development cannot occur at the pace we need. I urge you to read the article in this newsletter from our Chief Science and Alliance Officer, Dr. Phil Yeske. Dr. Yeske outlines some of the positive steps we are taking that we know will move us towards treatments and cures.

As new research and better information about mitochondrial disease becomes available, we all need to become a better prepared advocate for our affected loved ones or a better informed patient. Education is the key, and included in this issue is a registration form for Mitochondrial Medicine 2014: Pittsburgh. We will gather for this symposium in the first week of June. The scientific meetings start on June 4th and education for affected individuals and families begins on June 6th. If you haven't been to a symposium, I would urge you to attend; if you have been in the past – attend another one. Our annual meeting is the place where you are able to learn, interact with mitochondrial disease specialists, and network with people who share your experiences with mitochondrial disease. Last year, at our 2013 Symposium, we heard from scientific and medical experts who reported on the progress that has been made in the last decade. They outlined 20 potential therapies developed in that timeframe. I hope you will register by mail or online and attend! I look forward to seeing you in Pittsburgh!

Energy to all,

W. Dan Wright, UMDF Chairman

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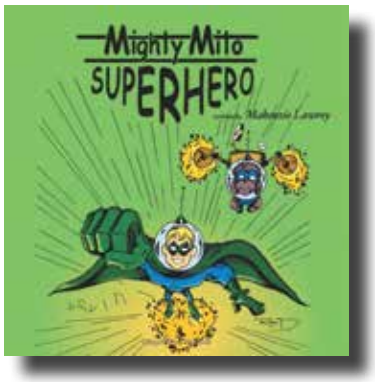
members

Uplifting stories about our members

Young Author Supports Brother

Nine-year-old Makenzie Lawrey is a big sister on a mission to save her little brother, Gavin, and others that suffer from Mitochondrial Disease. She's a little girl with big dreams. She wants the whole world to know what Mito is and believes that one day there will be a cure. She wants people to know the kinds of incredible superheroes kids with Mito Disease are. She wrote and published a book "Mighty Mito Superhero" in honor of her brother's life long journey with an ultimate goal to raise $1 million for UMDF. Join Makenzie, Gavin and their family to spread the word and the awareness. To read more about Makenzie's mission, visit her website http://www.hope4mito.com/



Awareness Gone Viral: Corynna's Impact

by Liz Weiss, UMDF Staff

Corynna Strawser didn't let Mitochondrial Disease define her. She chose to live her life to the fullest. This very wise 16-year-old made sure to get a daily dose of laughter even on the most difficult days. As her illness progressed, the more positive her attitude became. Like many with Mito, Corynna saw a number of doctors, endured countless medical tests, and had numerous unanswered questions before receiving a diagnosis. After understanding that Mito was the reason for her health issues, she decided to spread awareness and educate others about it.

The director of Akron Palliative Care, where Corynna received care, asked her if she would like to participate in a television show through Western Reserve PBS called: *A Closer Look: Mitochondrial Disease.* Although her health was poor during the filming, it was how she wanted to spend her energy. It is tentatively scheduled to air in April 2014 and will also be used for training purposes at Northeast Ohio Medical University.

Corynna and her parents, Kristi and Sean, created a Facebook page in August 2013 called *Courage for Corynna* to update her followers about her journey. Kristi said that she and Sean changed the name to *Courage from Corynna* after she passed away in December of 2013 because others needed the courage more than Corynna did. The page currently has over 7,300 followers. On the Facebook page, a picture was posted of the presentation of a $5,400 check, which was received solely from selling T-shirts. The shirts were even sent to supporters in Alaska and Canada.

Thanks to the efforts of Corynna and her family, everyone in the close-knit community of Greenville now knows something about mitochondrial disease.

The fundraiser inspired fellow Greenville, OH, community members to organize the declaration of "Mitochondrial Disease Awareness Day," on November 25th. Kristi stated, "The journalist from the local paper prepared the declaration and the county commissioners came to the home that day to make it official." This event motivated the Mito community in Evansville, IN, to also declare November 25th as "Mitochondrial Disease Awareness Day." Everyone in the close-knit community of Greenville now knows something about Mito. Corynna led her community to come together and having Mito simultaneously brought the community to her. Over 200 community members came together to sing Christmas carols outside Corynna's home.

When the Strawser family realized how successful the t-shirt fundraiser was, they decided to start a research fund through the UMDF. Kristi explained that starting a research fund allowed Corynna to see the impact she made and to see how her legacy would live on. Corynna picked out pictures for the brochure/webpage herself. Her research fund currently has over $20,000 in donations. To check out her research fund, please visit www.umdf.org/corynnastrawser.

Driven to Succeed


Stevie at the 2013 Houston EFL

Many of our attendees at the 2014 Energy for Life: Houston Walkathon would probably agree that this year's final walk finisher, Stevie Reid, finishes first when it comes to determination and inspiration! Stevie first started training and walking five years ago. Due to an unusually icy winter this year in Houston, Stevie had less training time. Nevertheless, Team Stevie returned for their second walk, led by Stevie's bright smile and great attitude.

Every year, Stevie has walked the Houston walk route using his gait trainer. His stepfather, David, told us that he went into this year's walk with lower energy levels. Still, Stevie completed 1.09 miles of the walk route! As he was nearing the finish line, every walker and volunteer began to cheer on Stevie and his team. We could tell he was tired and that he had to fight for those last ten yards, but his determination never flagged.

As his stepfather said, "Stevie's main focus and greatest desire is to inspire others to be all they can, to overcome their circumstances and live life to the fullest with a positive and joyful attitude. While exhaustion can be a problem for those with mitochondrial disease, he sees the chance to inspire others worth the discomfort of his fatigue. He looks at me and says, 'You only live once, make it count!'"

When Stevie crossed the finish line, his face lit up once again. Walkers around him were inspired to tears, and volunteers from the University of Houston's Pre Pharmacy Association gave up their "gold" finisher medals to Stevie. But at the 2014 Energy for Life: Houston Walkathon, Stevie won so much more than a neck full of medals--he won all of our hearts!


Stevie posing with the University of Houston's Pre Pharmacy Association at the 2014 EFL


Stevie with his medals

Justin's Story

Years ago, mitochondrial disease was thought to only affect children. As science and diagnostics progressed, we learned that many adults are affected and are living with mitochondrial disease. For many years, many affected adults knew something was wrong, but lived undiagnosed. Some are able to manage their care by themselves, but many others are being cared for by parents, some of them who have medical issues of their own. It can be a daily struggle for both the patient and the parent.

Justin is currently 41 years of age. Five years ago, at the age of 36, he was diagnosed with a deletion on the nuclear gene NDUFA1 located on chromosome Xq24. The NDUFA1 subunit, essential for complex I assembly and function, has been shown to interact with the mtDNA encoded subunits during the complex I assembly process. For Justin, who is now approaching middle age, it means seizures, aphasia, ataxia, and atrophy of the brain. Justin is one of the growing number of adults whose care is being overseen by their aging parent or parents.

According to his mother, Rita, Justin was born a healthy, happy 6 lb. 10oz baby boy on November 14, 1972. By the age of 3, he had been to many specialists. Rita was assured by doctors this was a stage and that Justin would grow out of it. "At the age of 3-1/2, he started to withdraw into himself. He became afraid of what was going on, and it was happening fast," she said. Justin started talking at a very young age. His clear and correct speaking pattern reverted to infantile babbling. Justin's pediatrician suggested to Rita and her husband that they take their son to see a neurologist.

"We lived outside Toledo, OH, at the time, and there was only one pediatric neurologist around. I called to make an appointment, and they told me it would take a month or so to get an appointment. I told them we didn't have that much time because Justin seemed to worsen daily," Rita said.

Test results revealed that Justin was having seizures. By this time, he stopped speaking and was only making sounds. None of the psychiatrists, neurologists, and many other doctors they saw could give Rita and her husband any answers.

It would not be until 1993 that they heard an unfamiliar phrase – mitochondrial disease. Skin, nerve and muscle biopsies were performed. The results came back unspecified.

In 2008, 15 years after the biopsies, Rita received a call from the University of California at Irvine MITOMED Center for Mitochondrial Medicine. After checking DNA from everyone in the family, the mutation was confirmed!

Justin has a younger sister, Jaime, three years younger than Justin. "She was tested and it was determined that she is a 'carrier'," Rita said. In this particular mitochondrial disease, it is carried by the female in one of her "X" chromosomes. If passed along to a male, they become affected. If the mutation is passed along to a female, they become a carrier. Testing also revealed other affected family members. "I had a brother who was affected by this disease even though there were no good records back then. I also have a nephew who was tested at the same time as Justin and confirmed with the same mitochondrial disease," Rita explained. "Parents should keep pushing for everything that is needed for their children."

Rita lives in the Atlanta, GA, area now with her husband, who was left disabled by a stroke. Due to the progression of his illness into adulthood, Justin now requires total care. Ever since Justin aged out of the public school system, he has lived in another family care setting with two other adults. "My biggest fear for the future is that he will outlive me," Rita said. "I am very blessed to have found a wonderful family that provides Justin's care."

Below are reader-submitted poems from Allisa Whitt of Bethel, Ohio. Allisa is the mother of Megan and Leslie Whitt. Leslie was affected by mitochondrial disease. On the left is a poem written by Leslie in 2004 on the occasion of her sister, Megan, leaving for college. On the right is a poem updated by Megan and read at Leslie's funeral in 2012. Publishing these poems was something that was on Leslie's "bucket list" of things she wanted to do before losing her battle with mitochondrial disease.

Love You Always
by Leslie Whitt (2004)

Every night my fear grows worse.
The thought of you leaving is too grim.
I need you in my life,
You are my hero.
Please don't leave.

When you go to college,
you will make new friends.
And I will fade into the distance.
An occasional call or letter,
but no more hugs and giggles.
Please don't leave.

On the day of your wedding,
Mom will be by your side.
I will be watching from afar.
Knowing that I still love you,
Please don't leave.

When you move away from here,
You will start a new part of your life.
One without me.
I don't want to lose you.
Please, don't leave.

Even if you have to leave…
I want you to know
There will always be a place for you in my heart.
I love you Megan…Always.

Love You Always
by Megan Whitt (2012)

Every night my fear grows worse.
Thoughts of your death; too grim.
I need you in my life,
You are my hero.
Please don't leave.

When you stop breathing,
I will lose my best friend.
My confidant, the other half to my whole.
My thoughts will turn to you often.
But no more hugs or giggles.
Please don't leave me

On the day of your funeral,
I will stand by your side.
Afraid and not understanding,
but knowing that I still love you.
Please, don't leave.

When you close your eyes for the last time,
a new chapter will begin.
One day without you.
I don't want to lose you.
Please, don't leave.

Even if you have to go...
I want you to know
There will be a place for you in my heart.
I love you Leslie….Always.

Wish Comes True for Coast Guardsman's Son

by Petty Officer 2nd Class Walter Shinn, U.S. Coast Guard
Originally published at Military.com

A boat is sinking with four people aboard in the middle of 30-foot seas that are crashing into each other, spraying salt into the air and creating deep swells giving way to the sense of riding on an unstable roller coaster. Scenarios like these are what Coast Guardsmen prepare for. Service members who are operational within the Coast Guard are expected to train rigorously for treacherous conditions or dangerous situations during the workday and on watch. For Coast Guardsman Lt. Adam Schmid, this is his career.

Schmid, an operations officer for Maritime Safety and Security Team Boston, has been in the service for nine years. His job responsibilities include oversight and directing operations including missions within maritime law enforcement, Anti-Terrorism/Force Protection and waterside security mission.

Schmid is also a husband and father of four. One could say he is prepared for many different scenarios. However, a challenging scenario revealed itself in October 2012 when his family discovered their 7-year-old son Thomas' biopsy results confirmed he had mitochondrial myopathy, which ultimately led to mitochondrial disease. Mitochondria are responsible for creating more than 90 percent of the energy needed by the body to sustain life and support growth. When they fail, less and less energy is generated within the cell. Cell injury and even cell death follow. If this process is repeated throughout the body, whole systems begin to fail, and the person's life is severely compromised. The disease primarily affects children, but adult onset is becoming more and more common.

Last fall Schmid submitted an application to Make-A-Wish Massachusetts and Rhode Island. "So soon after I submitted Thomas' information to "Make-A-Wish," a wish team visited our home," said Schmid. "When Thomas was asked what his wish was, he wished to take his whole family to Disney's Animal Kingdom. From there, the Wish team took action and set up a trip to Orlando the week before Christmas 2013."

Ultimately, the Schmid family visited all of the major Orlando theme parks creating memories that will last for generations. "The trip was an opportunity for Thomas to do whatever he wanted and enjoy all that the parks had to offer while his health is relatively good," said Schmid. "As a family, we were able to forget about the stresses that we have dealt with over the last year and share moments that we will talk about the rest of our lives." While the entire family enjoyed the trip to all the theme parks, it was especially significant for Thomas. "The trip was more fun than art class, and art class is wicked, wicked fun," said Thomas. From beginning to end Make-A-Wish Massachusetts and Rhode Island enabled the Schmid family to step away from Thomas' physical therapy treatments he goes through four times a week.

"We were blown away by the red carpet "Make-a-Wish" rolled out for us, from a limo ride to the airport to VIP passes to all the major Orlando theme parks." said Schmid. "The genuine care for Thomas and the rest of the family by those who made this amazing trip possible was overwhelming. We can't thank Make-a-Wish, Give children the World and the Orlando theme parks enough."

The Schmid family continues to pursue treatment for Thomas' ailment and is partnered with the United Mitochondrial Disease Foundation (UMDF) to bring awareness to the disease.

Medical Child Abuse

It can be a parent's worst nightmare. An accusation can be made in a number of ways that parents and caregivers are not acting in the best medical interest of your child. The formal name is Medical Child Abuse. What you may not know is the UMDF has been instrumental in helping parents diffuse many of these situations before they became public. We have worked with hospitals, educators, and lawyers in providing important information about the unique and often misunderstood and complex medical needs required for the patient's care.

Prior to the media reports of the current medical child abuse case, as part of its family symposium in June, the UMDF planned to devote an entire session to the issue at Mitochondrial Medicine 2014 in Pittsburgh, June 4-7. Realizing this issue could not wait, we provided patients and families with information on our website and through a webinar. You can see this information at www.umdf.org/mcapatientinfo.

Many affected individuals wonder why mitochondrial disease and other patients with complex metabolic issues seem to be at a higher risk for being suspected of medical child abuse. According to Mark Korson, MD, the answer, in part, may be a communications problem.

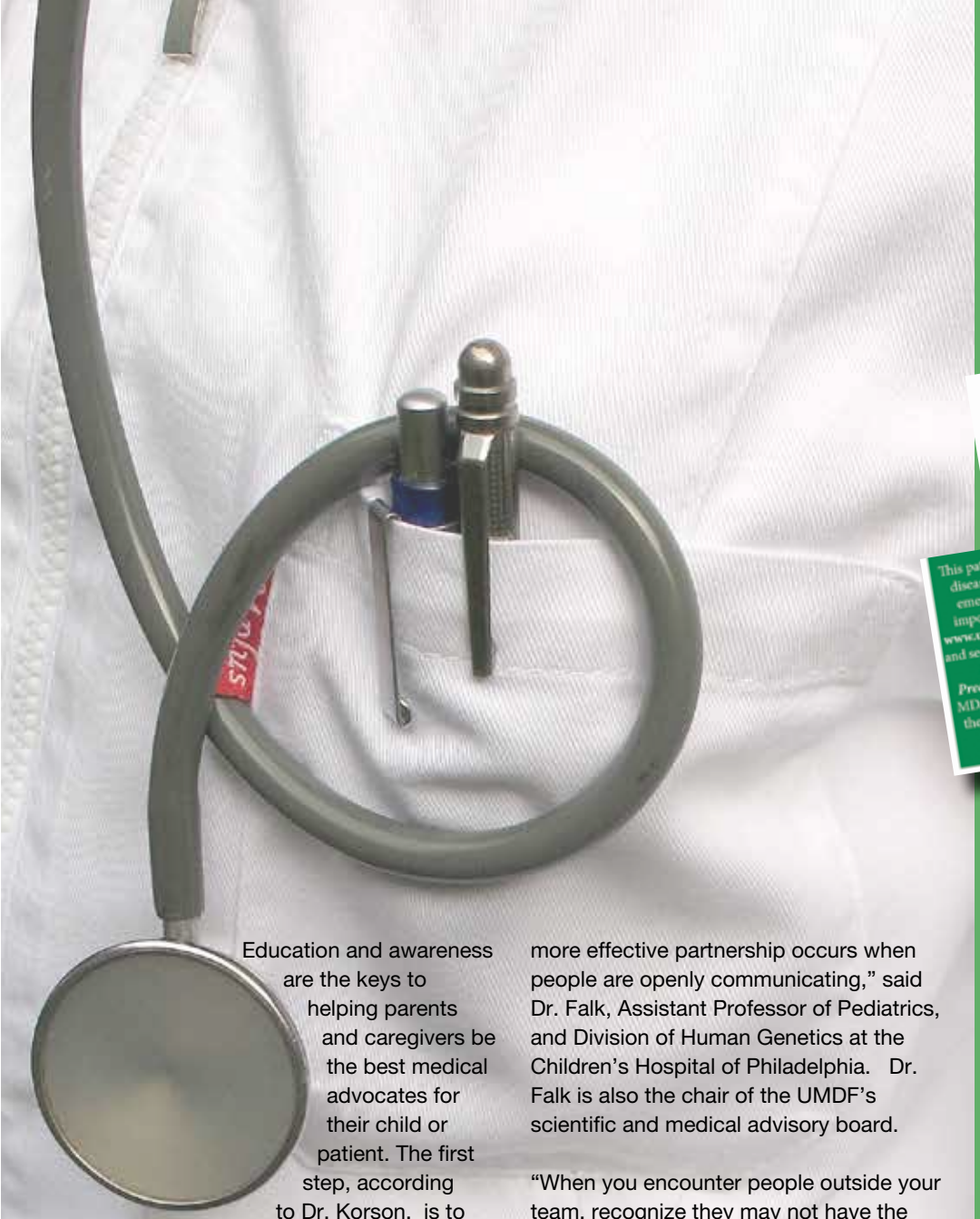
Dr. Korson, Chief of the Metabolic Service at the Floating Hospital for Children at Tufts Medical Center in Boston and associate professor of pediatrics at Tufts University School of Medicine, says that because multiple systems are involved in mitochondrial diseases and other complex disorders and because these diseases vary and are difficult to diagnose, patients must deal with multiple doctors. Dr. Korson, who was interviewed on the subject for a UMDF webinar, believes that sometimes the communication between the multiple doctors and numerous hospitals seeing a complex case is not optimal. In many cases, that is when suspicions may be raised.

"You have parents who often know more than doctors about what's wrong with their child, relating information from doctor to doctor. Parents think they're doing a good deed but, in fact, it can put them in a bad light," Dr. Korson added.

A PARENT'S PERSPECTIVE ON HOW THE UMDF'S MCA INFORMATION HELPED HER:

"We have been fighting for appropriate school accommodations for our son since last October. They ignored every single piece of outside documentation we have ever provided them. Someone at the school filed a report, falsely accusing us of Medical Child Abuse. Child Protective Services considered this to be a mid-level threat and must be investigated within 24 hours. I called our UMDF regional coordinator and also contacted the UMDF headquarters. I was sent links that are super-useful to anyone who hasn't spent a lot of time on the UMDF website already. (I use the UMDF website a lot for directing patients to learn more, so I know it pretty well, and I love the videos and fact sheets that are offered there.) The turnaround time was quick. I printed off the UMDF information and gave it to the school representative; she expressed that their medical professional would be interested in learning more about mito and that she had learned about mito from reading about it for my son. She admitted she knew very little about it before getting our case.

We also have a great team of doctors that were very upset about the report; so the school district contacted them and they provided the information to support the interventions my son was getting. The school district had 30 days to clear us; on day 29 we got the call that we were cleared and that the report was unfounded."



Education and awareness are the keys to helping parents and caregivers be the best medical advocates for their child or patient. The first step, according to Dr. Korson, is to provide education. "It's worthwhile to guide any investigators or concerned clinicians to the UMDF website to get more information. There's nothing more important early on than to get all providers together to share what they see, to share – not only to share their experiences, but to hear what other people have seen, what other people's concerns are and that can go a long way to allaying fears and concerns and suspicions," said Dr. Korson. "Sometimes there's no need for further investigation because the questions get answered."

Marni Falk, MD, who also participated on the webinar, believes parents and caregivers need to work together with medical providers in the care of a child with complex medical issues. "I think the more effective partnership occurs when people are openly communicating," said Dr. Falk, Assistant Professor of Pediatrics, and Division of Human Genetics at the Children's Hospital of Philadelphia. Dr. Falk is also the chair of the UMDF's scientific and medical advisory board.

"When you encounter people outside your team, recognize they may not have the expertise, but they may be able to help you with a specific type of problem, or if in an acute setting, involve your regular team," Dr. Falk said. "I think respect goes a long way in both directions and to really stay focused on optimizing the care of this sick individual."

In June, Dr. Korson will lead a discussion on the topic of medical child abuse. The discussion will be presented to family members at our international symposium in Pittsburgh. One of the issues to be discussed is how patients and caregivers can reduce the risk of unnecessary allegations. It is the third time in as many years that UMDF has presented this information at symposium.

IMPORTANT INFORMATION FOR PATIENTS AND CAREGIVERS:

Do you have the UMDF EMERGENCY ROOM PROTOCOL CARD?

This card easily fits into a wallet or purse and can be given to medical personnel. The card states "This patient has a mitochondrial disease and requires specific emergency room protocols". The card provides a smart device link to the UMDF Emergency Room Letter that can be viewed online and provides for a smart device link to Mito 101.

Would you like the CARD FOR CLINICIANS?

This card was developed by the UMDF and provided to patients to give to their medical professionals. It provides an immediate link to the UMDF website and to Mito101, our resource for physicians and patients.

These are two very important pieces of information that you should have. If you would like to receive them, please call 888-317-UMDF (8633) or e-mail us at info@umdf.org.
You can also immediately download copies at www.umdf.org/mcaparentinfo.

Patient Registry and Genomic Data

by Phil Yeske, PhD, UMDF Science & Alliance Officer

Facilitating the development of treatments and cures for the mitochondrial disease affected community remains a key component of the UMDF's vision. How best for our foundation to do that, given limited resources, needs to be revisited on a regular basis as the scientific and business landscape changes. Today, I would like to share some thoughts on the strategic direction of the UMDF in this regard.

Historically, the UMDF has invested significant funds into basic research on mitochondrial dysfunction. Over the past decade, dozens of grants totaling millions of dollars have been awarded by the UMDF to academic researchers around the world. The focus of the grant program has always been, and remains, how to get the biggest bang for our buck when considering what science to support. Toward this end, considerable time and effort is put into a peer review process (scientists critiquing other scientists' ideas) that is both transparent and thorough. The current 2013-2014 grant cycle is well underway, and we've already whittled down over 200 high quality initial Letters of Intent to around 50 full proposals for consideration by reviewers made up of a diverse group of leaders on mitochondrial science. Their job is to objectively score the proposals in advance of a face-to-face meeting of the roughly 10-person Grant Review Committee in April during which a final set of applications will be recommended for funding. My colleague, Jean Bassett, does most of the hard work in coordinating this process, and her efforts are much appreciated by all involved. We continue to believe that supporting basic research is a critical step in facilitating the development of therapies for mitochondrial disease. However, that alone is insufficient.

A true therapeutic development initiative requires stakeholders in academia, medical and government institutions as well as drug development companies working in concert. Our benchmarking of other rare disease communities produced a clear, consistent message: the most efficient route to therapeutic development occurs when a patient advocacy group is positioned as the central steward of the process. That same benchmarking also reinforced the importance of a well-characterized patient community via a robust patient registry, well-stocked biobanks and a clinical research network. The good news for our community is that there are established building blocks in place, including NAMDC and the Mayo Clinic Biobank. There is room for improvement in what exists and certainly a need for additional pieces of the puzzle. This stewardship role is what the UMDF aspires to, and there are two key near-term projects that we intend to pursue toward this goal.

The first project is the creation of a patient-populated patient registry to complement the scientifically-populated NAMDC registry. There are currently about 500 patients enrolled in the NAMDC registry, and it continues to grow at a pace of about 15 new registrants per month. We know there is a much larger patient community with vital information to share and our intention is to be the guardian of that precious data. By knowing who you are, how your condition manifests itself, and how you deal with life on a daily basis, we will build a database that similar foundations have termed the most important asset a rare disease community can possess in the quest for development of treatments and cures.

The second project involves the integration of genomic data with medical information. The combination of this data is what will lead to faster and better diagnoses, as well as yielding biological targets for development of treatments. The mito community, again, is fortunate to already have a prototype initiative underway called Mitochondrial Sequencing Data Resource Tool (MSeqDR), in which genomic data (both mitochondrial and nuclear) from whole exome sequencing is shared and aggregated in an anonymous way, then made available to the research community. This project is already being financially supported in-part by the UMDF and, by this summer, should go "live" and begin accepting massive amounts of new data.

Contributing to the creation and maintenance of these projects is only a small part of the envisioned UMDF stewardship role. Ensuring that all the pieces of the puzzle work together with an unwavering focus on the patient community is the real job. We have, and will continue to, carefully but forcefully utilize the trust you have placed in the UMDF to advance our shared mission of bringing HOPE to the mitochondrial disease affected community in the form of treatments and cures.

I look forward to sharing further details in future articles, but even better would be the chance to see and speak with you in person this coming June at either Mitochondrial Medicine 2014 in Pittsburgh or Euromit 2014 in Finland!

Edison Pharma's partnership offers hope for Mito community

The nation's leading mitochondrial disease patient advocacy organizations have joined together to congratulate Edison Pharmaceuticals in their announcement last week of a nearly $4.3 billion strategic partnership with Dainippon Sumitomo Pharma Co., Ltd. of Japan. The Mitochondrial Disease Action Committee (MitoAction) and the United Mitochondrial Disease Foundation (UMDF) believe that the partnership between the two pharmaceutical companies is monumental for the mitochondrial disease patient community.

The partnership enables Edison to have the resources to bring drugs specifically created for pediatric mitochondrial disease patients, like EPI-743, to the marketplace. The partnership also enables both companies to research, develop, and build a pipeline containing 10 new drugs targeting various aspects of cellular energy metabolism. Those efforts will allow the two companies the ability to target adult neurological diseases that also directly impact the mitochondrial disease adult community.

"Not only does the partnership between Edison and Dainippon Sumitomo give us real hope for a treatment for mitochondrial disease in the foreseeable future, this alliance instantiates the field of 'mitochondrial medicine,'" said Cristy Balcells, RN MSN, and Executive Director of MitoAction. "Our patients, patient community and separately Edison have been pioneering this concept. Today, our dream has become a reality. We believe the field of mitochondrial medicines will grow and establish itself as a standard medical and household term," Balcells added.

"This is a huge victory for mitochondrial medicine," said Charles A. Mohan, Jr., CEO/Executive Director of the UMDF. "When a partnership of this magnitude is announced, it brings attention to the broader impact that finding treatments and cures for mitochondrial disease has on all of us," Mohan said.

Mitochondrial dysfunction is linked to many neurological diseases such as Parkinson's, Alzheimer's, ALS, and other diseases like diabetes and some cancers.

"Our expanded partnership with Dainippon Sumitomo Pharma codifies a common value system and vision for discovering and developing new drugs for patients suffering from diseases that share a common mitochondrial mechanism," said Guy Miller, MD, PhD, Chairman, CEO, Edison Pharmaceuticals, Inc. "By combining efforts with DSP, we are able to scale our initial discoveries and therapeutic programs, and to begin to establish our commercial enterprise. This will start with EPI-743 currently being developed for children with a variety of rare mitochondrial diseases."

As patient advocacy groups, UMDF and MitoAction have worked in a collaborative effort over the past five years to provide education and information about Edison's EPI-743 and other novel therapeutics in development. Both organizations are grateful to the Edison team, specifically Guy Miller, for his dedication, inspiration, and transparency in providing information to, and receiving input from the mitochondrial disease patient community as he continues his mission toward providing the first approved mitochondrial disease drug.

UMDF
education
Bringing the experts to your community

Patient Care Concerns About the Affordable Care Act

From time to time, patients and families e-mail the UMDF office with questions about the Affordable Care Act and its implications for those who have mitochondrial disease.

This is especially true when a patient or family member is suddenly unemployed from a job that provided health insurance benefits. They find themselves venturing onto the internet to find coverage options on what is known as the 'insurance exchange'. Some are contacting private insurance companies for policy information.

One of the common concerns we hear from patients and families involves the "in state" restrictions placed on beneficiaries of policies offered on both on the exchange and private insurances. For example, some exchange and private insurance policies may only allow a patient medical treatment in their home state. Many mitochondrial disease patients receive their care at facilities that are not located in their home state. That is a significant issue for many of our patients and families.

Miranda Franco, Senior Public Affairs Advisor of Washington DC-based Holland+Knight, says that in order for plans to be sold on the exchange market, insurers must follow rules like marketing their plans fairly and must offer at least one plan in the "silver" and "gold" categories.

The United Mitochondrial Disease Foundation provides patients and families with insurance resources on our website. Please visit the page at www.umdf.org/insurance

"Some plans offered on the exchanges may not include a beneficiary's current provider or hospital. For instance, only Medical Mutual of Ohio offered in the Ohio exchange includes the Cleveland Clinic in their network. It is probably fair to say that some insurance companies may narrow their network to stay within premiums," Franco said. However, she says situations vary considerably by state. "For instance, Johns Hopkins in Maryland is mandated under state law to accept all insurance companies," Franco added.

Since the Affordable Care act is under implemention, it might be difficult to change any of its parameters in 2014; but that doesn't rule out the UMDF's ability to help refocus the issue so that we can broaden health insurance network participation. In order to do that, we need you to let us know the problems that you may encounter in finding adequate medical coverage for mitochondrial disease in your state. We can use this information to advocate for increased network adequacy requirements for 2015 and ensure that health plans are delivering the benefits and reasonable access to mitochondrial medical experts as promised under the terms of the contract.

Tell us how this impacts you at news@umdf.org. We will share your letters with members of Congress with the goal of providing the access our patients and families need.

Advocating For Your Child: Navigating Through Your Health Insurance

by Dino M. Scanio, Pediatric Orthotist

The face of healthcare has changed dramatically in the last five years due to legislative influence and a spiraling economy. This has forced health insurance companies to reexamine their policies and how they provide coverage. Under examination, your health insurance company may need to make changes to save money. Their potential savings will come from issuing denials and/or limiting coverage. This is especially true when it involves coverage for children. What do you need to know in an effort to ensure proper coverage for your child?

Health Insurance Plan Documents
Your insurance plan documents are the documents of truth. As a parent of a special needs child, it is important to read through the insurance plan documents. Make sure that a service is a provided one before you seek care. BUT just because it is stated in your policy as a provided service DOES NOT mean it will be approved. The trend with health insurance companies is to deny services and hope that you will not proceed with an appeal. If you give up, it will save the insurance company money.

Prescription
It is important that your paperwork is in order prior to obtaining medical services/ treatment or DME (Durable Medical Equipment). Make sure the prescription is properly filled. This will reduce the "red flags" that health insurance companies rely on as an easy way to issue a denial. The prescription MUST have:

- Patient Name
- Date
- Doctor's Orders (Physical therapy or Ankle Foot Orthosis are examples)
- Doctor's Signature
- Diagnosis

Medical Necessity
To support your effort as a parent advocate, it may behoove you to have your child's physician provide a statement of "medical necessity". This is a letter that explains the diagnosis/condition, why the prescription was written, and the expected positive outcomes, as well as any negative effects the patient might incur if the prescription is not filled. The letter must be signed by the physician and must include their credentials. Medical necessity letters are vitally important when seeking nursing care.

Digital Photographs and/or Video
Pictures and video speak louder than words so it may be advantageous to take a few digital photographs/videos of your child. For example: if your child is unable to walk appropriately without leg braces then make a video of your child attempting to walk without braces, and make sure you include the process of putting on the braces and how they improve your child's gait. Another example is the need for nursing care to manage a wound. Make sure to take digital pictures of the wound site.

Preauthorization/Determination of Coverage
When coverage is questionable, you should be able to ask for a preauthorization for the requested services/prescription. Some insurance companies will say this is not necessary, but it is in your best interest to request one. This will be your ammunition for the appeals process if services are denied. All preauthorization statements will include a disclaimer to the effect of "this is not a guarantee of payment/coverage," because insurance companies are looking to save money, ***not*** spend it.

Now you have been given the information to make you a better advocate for your child and some insight on navigating through murky waters of insurance trends. So what do you do if services are denied? Consult your insurance provider and request their guidelines for the appeals process. Insurance companies hope that the policy holder will not appeal or give up during the process because it is such a mundane task. ***Giving up will save them money - at the expense of your child's health!*** If your appeal is denied, you may have the option to request a doctor to doctor appeal, an external review or mediation. This information will be outlined in your plan documents but if further assistance is needed, you can request help from the State or Federal Insurance Commissioner. You can also file a complaint with the Commissioner which will require them to assign an investigator to your case.

We as policy holders cannot and should not allow insurance companies to dictate care. It is humanly wrong for them to say when, how and why or why not health services are provided. Doctors are experts that have the right to make decisions based on the interests of their patient. They are held to a high standard by a medical code of ethics. In my opinion, if a doctor orders it, then it should be covered by the insurance company. Be your child's advocate and fight against the tyranny of the health insurance companies.

Ask the Mito Doc℠

Living with mitochondrial disease presents many twists and turns, and a maze of questions. UMDF is pleased to offer answers to some of those questions as taken from Ask the Mito Doc℠ at www.umdf.org. Please note that information contained in Ask the Mito Doc℠ 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 4 year old son has not officially been diagnosed with mitochondrial disease. He had a liver biopsy in June that showed some abnormal mitochondria. We are currently waiting for the rest of the results from that biopsy. When he was 3 months old he got a high fever from his immunization shots. After 4 days of feeling very ill, he turned jaundice and doctors at the ER told me to hydrate him. This would occur every time he seemed to have the flu or would get his shots. Finally, this past February I took him to the Children's Hospital and they said his liver enzyme levels were dangerously high. He recovered on his own as usual. Just this time was aided with IV. His kidney function is perfect, his MRI was outstanding, his glucose levels are perfect, his opthomolgy appointment was excellent. He has no muscle pain. He is growing above average and his learning is remarkable. On a normal blood test all levels are great. Only when he becomes ill that everything changes. He won't eat, drink, sleeps the day away, turns jaundice then feels better. Have you had many mito patients with this one symptom of acute liver failure?

A: I have seen children with mitochondrial disease that just manifest with liver disease or liver failure. They typically present at a younger age and their course is complicated with high liver enzymes and jaundice. As the liver is important for carbohydrate metabolism, they may develop low blood sugars as well. Most of these children do not grow well and in some cases their brain can be affected. Children with liver disease may have a condition called mitochondrial DNA depletion where the amount of mitochondrial DNA declines over time. It would be important to measure the amount of mitochondrial DNA in the liver tissue of your child. The clinical course in your child seems to be milder than what is typically seen in children with mitochondrial disease and liver problems. In the meantime and until a diagnosis is found, it would be important to stay away from medications such as valproic acid that is sometimes used in children who develop seizures since it could worsen his liver problems. ***- Fernando Scaglia MD, F.A.C.M.G.***

Fernando Scaglia, MD

Q: Is "Brain Fog" a common complaint for adults with mitochondrial disease? What are the possible causes? Can medication (such as pain meds) commonly taken by patients with mitochondrial disease cause or contribute to "brain fog?" What, if anything, can be done to help alleviate it?

A: Complaints about memory problems and difficulty concentrating are quite common in mito. Waxing and waning nature of brain functioning is also a common feature, when at certain times a person feels sharp and thoughts and language flow well while at other times, language is slow, retrieval of words can be challenging, focus can be difficult and mental fatigue sets in. These cycles can happen within the same day (sometimes within the same hour) or over several days. Insufficient ATP production is believed to cause this poor brain functioning. Your brain simply runs out of gas which affects higher cerebral functioning including memory, language, concentration, etc. Cerebral hemispheres in mito patients are typically sacrificed when the body runs low on energy and the organs vital to life are given a priority, such as brainstem, midbrain, thalamus and basal ganglia as well as heart, liver, kidney, lungs, etc. Pain medications (depends on which, but typically narcotics) can certainly worsen cerebral functioning even in healthy people and mito patients are more prone to their sedative effects. That's why I use them as a last resort and choose meds like gabapentin, pregabalin and amitryptiline that help chronic pain in mito. Adherence to a regular caloric intake with starch at night to avoid catabolism, regular exercise without overexertion and vitamin cocktail crafted individually (compounding pharmacy is very helpful to my patients) based on plasma and leukocyte levels would be the best choice. High-dose CoQ10 (ubiquinol), leucovorin (folinic acid) and NAC (N-acetylcysteine) can help. Fish oil can also be used (at least 1000 mg of DHA and EPA). ***- Dmitriy M. Niyazov, MD***

Dmitriy M. Niyazov, MD

Q: My children and I carry the mitochondrial gene for Lebers Hereditary Optic Neuropathy, LHON. It is fine unless the disease is triggered and then the person loses his/her vision. I know certain drugs are damaging to the mitochondria. My son has ADHD and has trouble in school; we have tried lots of homeopathic intervention. I have been advised to put him on a medicine to help him focus. I have heard that the drugs like Ritalin are not safe. One doctor recommended that we try Strattera and said that should be safe and not damage the mitochondria. I am trying to find any answers. Do you know if this class of drug is safe? I really don't want to trigger blindness in my child.

Sumit Parikh, MD

A: I am sorry to hear that your family carries an LHON mutation. It is important to note that not all patients with LHON become symptomatic, and the genetic test results including the mitochondrial DNA haplotype can help your medical team determine the risk of developing symptoms. It is also important to note that Idebenone may help vision recovery if symptoms occur. If you or a family member are at-risk for possible symptoms, we mostly worry about triggers such as alcohol overuse and tobacco/nicotine as potentially bringing on vision loss.

On some level, many drugs can interfere with mitochondrial function; however this does not mean that they are not safe to use in patients with mitochondrial disease. We do not have a definitive list of medications that one must avoid. There is no clear evidence that any prescription medications bring on symptoms in LHON. If your family member needs a trial of stimulant medication, there is no mitochondrial contraindication to using the medicine (in LHON or other mitochondrial diseases). I recommend discussing any medications used with your mitochondrial physician. ***- Sumit Parikh, MD***

Q: I am a 69 year old woman. My doctor suspected a mito disorder and when I had genetic (cheek swab) testing it showed I have a Complex 1 disorder. I also have most of the symptoms of Autonomic Disorder, (can't get heart rate up, totally acontractile bladder, inability to orgasm, balance issues, depression, lack of energy, parkinsonian movements, tingling hands and feet, wake up with hands clenched and others, some quite severe). I have been tested for Parkinson's and I do not have it. In addition to that, even though I do not take any supplements that have B6 in them, my B6 is possibly higher than the highest value the test can determine. I'm told there is nothing that can cure any of these things, but I wonder what the relationship between them might be, given that the symptoms of all three disorders overlap. Based upon reading past posts I am considering going on a keto diet. What is your opinion of trying that?

Russell Saneto, DO, PhD

A: I am sorry that you are having so many problems. Let me first give you my opinion about trying the ketogenic diet. I am the person who does all the dietary therapy at the Children's Hospital that I am a part of, so I can give you some inside information. As you know the diet is 90% fat. This fat content is very hard to tolerate, as the dietary selections are limited and the high fat is hard to give flavor/taste to and is very bland. The diet is very concise in the way it has to be done; the ketogenic diet ratio (carbohydrate + carbohydrate producing protein/fat + fat producing protein = 1:4) must be kept for all meals. We are talking about meals that have almost all fat. This is extraordinarily difficult to maintain, even for children. Here is a typical breakfast for one of my patients: 1 boiled egg, 20 grams of cream, 1 piece of bacon, 22 grams of butter. We have had children eat their pet's food, beg for cookies at the neighbors claiming child abuse, rummage through trash cans, etc., trying to find carbohydrates. The diet also should be watched over by a dietitian that works with the ketogenic diet. The diet is terribly constipating and lacks in calcium that you need to supplement. So, as you can tell, it is very difficult to do. Furthermore, I am not sure if this would help your conditions.

I am not sure how I would counsel you on the buccal swab testing as the only validation that has been published on the use of buccal swabs to detect complex I dysfunction only looked at 26 patients with muscle proven complex I deficiency and only 20 were correlated (20/26). This is much too small of a number to give much weight to sensitivity and specificity of a test. This does not mean that you don't have complex I problems, but it suggests that to be confident of the diagnosis, you would probably need added evidence, either biochemically or genetically. Certainly, we do see patients with mitochondrial disease who have your symptoms, but there are other diseases that can give you similar symptoms that are not primary mitochondrial in origin. So, you need to be careful on how you approach the treatment of your symptoms.
- Russell Saneto, DO, PhD

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.umdf.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.

Research Report Review

by Jean Bassett, UMDF Staff

Problems of Adults with a Mitochondrial Disease – The Patients' Perspective: Focus on Loss

Authors: Greet Noorda, Theo van Achterberg, Truus van der Hooft, Jan Smeitink, Lisette Schoonhoven, and Baziel van Engelen; SSIEM and Springer-Verlag Berlin Heidelberg 2012

On February 24, 2011, the above research report was published online in the journal JIMD Reports. The aim of the study was to identify problems that adult patients with mitochondrial disease might face, resulting in better care and quality of life for affected adults. To achieve this goal, 16 adults with a mitochondrial disease participated in a single private interview in their homes.

As a result of these 16 interviews, the research team identified that "loss" was the most significant issue experienced by the participants. The dominant loss was of energy, which then precipitated other losses in all areas of life. The results were divided into seven categories by the researchers, including five types of losses: energy, independence and autonomy, social participation, personal identity, and dreams and future. Lack of healthcare, as well as coping and adjustment, rounded out the categories.

Loss of Energy: All participants experienced this loss. They reported various lengths and degrees of one or more symptoms such as weakness, fatigue, exhaustion, memory problems, muscle weakness and pain, and difficulties with movement or speech. For some, each day's symptoms were similar. For others, the symptoms were more unpredictable. But for all, energy loss was progressive over time.

Loss of Independence and Autonomy: For most of the participants, loss of energy then precipitated a lessening of daily activities and independent living. Mobility became an issue, which impacted activities such as walking, driving, housekeeping, childcare, writing, shopping, and eating. Increasing reliance on devices such as a cane, scooter, or wheelchair was typical. Difficulties with hearing, vision, concentration, or speech increased their dependence on others. Some participants could no longer live on their own, while others moved to more disability-accessible accommodations.

Loss of Social Participation: Loss of energy and independence resulted in restricted social interactions in the participants' personal and public lives. This impacted employment, income, leisure activities, and relationships with family and friends. Some reported that the lack of understanding and support from others was one of their most difficult problems, resulting in loneliness and isolation. In response, some withdrew from social interaction, while others reported meaningful new friendships with other affected adults.

Loss of Dreams and Future: Most participants were concerned about their future and the loss of goals and dreams they once held, especially in terms of relationships. They found themselves asking the more "spiritual" and "why"- type questions about themselves, and about life, suffering, and mortality in general. They struggled with concerns about heredity, testing, and what to tell family members. Two participants mentioned hope in medical therapies.

Lack of Health Care: Lack of information about their specific case, and the medical community's under-recognition of patients' intangible psychological, social, and spiritual needs contributed to the participants' difficulty in coping with their losses. Poor coordination of follow-up care, and differences in knowledge and approach by various medical personnel, added to their stress.

Loss of Personal Identity: For many of the participants, changes in their health and lifestyles resulted in lowered self-esteem and questions about self-perception. They were no longer sure about their own roles or those of family members and friends involved in their lives. There was concern over the negative opinions of others, i.e., sick, lazy, whining, etc.

Coping and Adjustment: In many cases, the road to a mitochondrial disease diagnosis was long and convoluted. Once the participants finally received the correct diagnosis, the coping and adjustment process began, often starting with many difficult feelings and questions. Next came the palliative phase, in which participants focused on a trial-and-error balance between energy and activities, often accompanied by stress.

As losses increased, the participants experienced a range of negative emotions including grief, anger, shame, and despair. Some reported positive attitudes as well, when they focused on the good in life, happy memories, support from others, humor, mottos, inspiring literature, and other types of encouragement. Nevertheless, it remained difficult to adjust to their increasing losses, much less accept them.

In conclusion, the research team determined that adults with mitochondrial disease face multiple tangible and intangible losses, and utilize a range of strategies to cope with them. They believe that by sharing their findings, medical/allied health professionals and patients' significant others will have a better understanding of the issues encountered by affected adults. This can lead to better care, support, and management of patient needs and a better quality of life.

Mitochondrial Medicine: 2014

Scientific Program
June 4 - 7, 2014

2014 Platform Sessions Include:

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

Course Chair: Jerry Vockley, MD, PhD
Course Co-Chair: Amy Goldstein, MD
CME Chair: Bruce H. Cohen, MD

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

For updates, details and CME information, please visit www.umdf.org/symposium.
Please use code SCI1408 for online registration.

Patients/Family Program
June 6 - 7, 2014

Special LHON Program: June 5, 2014

Topics for 2014:

- Mitochondrial Disease: What is It and What to Expect?
- Current Therapies in the Treatment of Mitochondrial Diseases
- Screening and Evaluation in Patients with Mitochondrial Disease
- Ask the Mito Doc Panel Discussions for Adult and Pediatric Patients
- The Challenges and Risks in Navigating Patient Care….in a World that is Still Learning about Mitochondrial Disease
- Palliative Care
- Exercise and Nutrition
- Adult Specific Topics
- Young Adult and Teen Sessions

For updates, scholarship and hotel information, visit www.umdf.org/symposium.
Please use code FAM1408 for online registration.

FAMILY PROGRAM REGISTRATION FORM
Mitochondrial Medicine 2014: Pittsburgh
Sheraton Station Square - Pittsburgh, PA
June 5 - 7, 2014

REGISTER NOW TO GUARANTEE YOUR ATTENDANCE!

Four Ways to Register:
1. Complete the registration form below and mail it back to the UMDF.
2. Complete the registration form below and fax it to UMDF at 412-793-6477.
3. Register online at www.umdf.org/symposium/registration. Use the registration code FAM1408.
4. Scan the QR code located to the left and register online. Use the registration code FAM1408.

Scholarships
For those experiencing financial hardship, limited funding is available to offset part of the cost of attending the symposium. To inquire, contact the UMDF toll free at 1-888-317-UMDF or visit www.umdf.org/symposium for more information. The scholarship application deadline is April 15, 2014.

Program Accessibility
Handicapped-accessible rooms are available upon request, and there is wheelchair access in all public areas. If you have any special needs that require additional assistance, please contact the UMDF staff at 1-888-317-UMDF. Requests should be received at least two weeks prior to the conference.

Scooters & Assisted Listening Devices
UMDF has arranged for assisted listening devices and scooters for those in need of this type of assistance. In order to ensure that we can meet your needs, please be sure to request an assisted listening device and/or scooter on your registration form. The assisted listening device is a headset that receives sound waves from the speaker's microphone that goes directly to the user's ears.

Cancellation Policy
Because attendees or family members may face unforeseen illness, the UMDF will refund the registration fee, less $25 cancellation fee, for cancellations received by May 30, 2014. All refunds will be made after the symposium is over.

Permission to Use Photos
By registering for this conference, you have given the UMDF permission to use pictures from this event for educational and promotional purposes for this and future conferences. If you do not want your picture to be used, please submit a letter by July 1, 2014, to UMDF, Attn: Cliff Gorski, 8085 Saltsburg Road, Suite 201, Pittsburgh, PA 15239.

Registration Code: FAM1408

REGISTRATION FEES

- ☐ $225 Individual Registration
- ☐ $400 Family Registration *(2 adults/same household)*
- ☐ $50 LHON Program **Only**
- ☐ $65 Additional Friday Night Banquet tickets *(per ticket)*

Registration fees includes hard copy of syllabus, daily continental breakfast, lunch, refreshment breaks, Thursday LHON session and the Friday Night Banquet.

TEEN REGISTRATION *(Sessions are free but registration is required!)*

- ☐ FREE Teen Registration *(Lunches and banquet not included)*
- ☐ $5.00 Friday Lunch *(per teen)*
- ☐ $10.00 Friday Night Banquet *(Special pricing for teens)*
- ☐ $5.00 Saturday Lunch *(per teen)*

Special Assistance ☐ Scooter ☐ Assisted-Listening Device ☐ Other _____

Special Dietary Requirements ☐ Vegetarian ☐ Gluten-Free ☐ Other _____
(Contact hotel directly prior to arrival to confirm)

One form per registrant. Please copy this form for additional attendees and TEENS.

Total Full Registration Fee: **$** _____

To pre-submit questions for "Ask the Mito Doc", visit www.surveymonkey.com/s/2014AskDoc

** A $25 late fee will be added to any full registrations received after May 26, 2014.*

Please make all checks payable to: **The United Mitochondrial Disease Foundation**

Please charge this registration to the following: Visa MasterCard Discover American Express

Card Number _____ Expiration Date _____

Name as listed on card (please print) _____

Signature _____ (invalid without signature)

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Email: _____

Phone: _____ Fax _____

Mail to UMDF, 8085 Saltsburg Road, Suite 201, Pittsburgh, PA 15239 or fax to 412-793-6477

Cheryl Lawson (left) accepts the 2013 LEAP Award from Sharon Shaw-Reeder on behalf of Cynthia Stewart at the 2013 UMDF Symposium

symposium AWARDS

Every year, the UMDF honors the accomplishments of our constituency. A number of awards are invited for nominations from the public. The UMDF invites you to nominate candidates for the three awards listed below. The deadline for nominations is April 15, 2014.

The awards will be presented at the UMDF's annual symposium on June 6, 2014. The winners will be honored with a plaque and will be featured in the UMDF Newsletter.

Energy Award

The purpose of the UMDF Energy Award is to recognize an individual who embodies the spirit of the UMDF and its Mission of "promoting research and education for the diagnosis, treatment, and cure of mitochondrial disorders and providing support to affected individuals." There is no age restriction for the nominee. You may nominate an individual for the UMDF's Energy Award by filling out the form online with a 100 word explanation as to how this individual has exemplified the UMDF Mission.

You may also mail or e-mail supportive documents of your nomination to info@umdf.org (please put Energy Award Nominee and their name in the subject line) -- listing projects, activities, or other information that highlights their efforts in supporting the UMDF mission.

Submit your nominations online at https://www.surveymonkey.com/s/EnergyAward2014

Heartstrings Award

The Heartstrings Award recognizes a child or teen that has donated or raised funds for the UMDF, enabling the UMDF to continue its mission. The individual recognized must be under 18 years of age at the time of the donation or fundraising activity.

As part of the criteria for the award, the nominee must implement a fund raising project, demonstrate how the project was communicated to the community for awareness, and show the time invested in the project. The amount raised in comparison to the age of the individual will be considered. For nominees who donate funds, the judges will consider the generous spirit shown, communication, and amount donated in relation to the age of the individual.

You may nominate an individual for the Heartstrings Award by filling out the form online with a 100 word explanation as to how this individual has "tugged at your heartstrings" through fund raising for or donation to the UMDF. Identify important features of the nominee's activity, such as the time invested, creativity, communication skills, determination, effectiveness, and generosity.

Submit your nominations online at https://www.surveymonkey.com/s/2014Heartstrings

LEAP Award

The LEAP Award recognizes an individual who is living positively with mitochondrial disease (Living, Encouraging, Achieving, & Persisting).

In order to be nominated, the nominee must be at least 14 years of age or older. The nominee must have a confirmed or suspected mitochondrial disease and has demonstrated how they overcome the daily challenges to achieve their goals in career, family or volunteer service. The nominee must demonstrate a positive attitude, hope for a brighter future, and have enthusiasm that inspires others.

You may nominate an individual for the LEAP Award by filling out the form online with a 100 word explanation as to how the nominee demonstrates a positive attitude, hope for a bright future, and inspires others. If you wish, please provide copies of articles about the nominee, lists of projects, activities, or clubs to which the nominee belongs.

Submit your nominations online at https://www.surveymonkey.com/s/LEAPAward2014

UMDF national

News from the national office.

Bill Goade, Managing Principal and former CEO of Cresa, visited the UMDF office in December to present a check for $20,000 from a company fundraiser. Bill has a friend whose son is afflicted.

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UMDF Mission

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

ORDR's Groft Retires

A public servant for more than four decades and a tireless advocate for rare diseases research, Stephen C. Groft, Pharm. D., gave thousands of patients and their families a renewed hope and voice. On Feb. 8, 2014, Groft retired from his current position as director of NCATS' Office of Rare Diseases Research (ORDR). He leaves a 30-year legacy of advancing rare diseases research and improving the lives of patients with these conditions. Groft began his career in 1968 as a small-town pharmacist in Pennsylvania, helping patients understand their conditions and medications and building connections that would influence his lifelong focus on rare diseases. His policy-related efforts began in 1982 at the FDA Office of Orphan Products, a division dedicated to advancing the evaluation and development of therapeutics for the diagnosis and treatment of rare diseases.

Throughout the 1980s, Groft served in a variety of leadership roles to help advance the national research and drug development agendas for rare diseases, beginning with his support of the Orphan Drug Act (ODA) in 1983. Prior to the act, pharmaceutical companies often neglected therapeutics for rare diseases because the cost of research and development frequently outweighed the return on investment for products that only a small number of people needed. The ODA provided financial and regulatory incentives to pharmaceutical companies to make production of so-called "orphan drugs" more cost-effective. Since the law's enactment, the FDA has approved more than 450 orphan products, and the rate of approval has increased: In 1983, two orphan products were approved; in 2012, 26 were approved.

"It's not often that you meet a person who successfully combines service with sincerity and integrity. But when you do, you have met a person who will change the world. Steve Groft is one of those people, and he has positively changed the world of rare diseases," said Charles A. Mohan, Jr., CEO and executive director, United Mitochondrial Disease Foundation.

During his more than two decades as ORDR director, Groft worked with legislators, regulators, researchers, pharmaceutical representatives, patients, families and patient advocacy groups to build a community that could address the needs of rare disease patients more effectively. Pamela M. McInnes, D.D.S., NCATS deputy director, will serve as acting director of ORDR during the search for a new director.

UMDF

events

The energy providing education, support and research.

Fundraisers Benefitting the UMDF

September 6, 2013 Shawn Stewart, who has LHON, held his 2nd annual golf tournament called, "Birdies for the Blind," which raised $3,000. Between the 1st and 2nd annual "Birdies for the Blind," a total of $7,000 was raised and 72 players participated!! Thank you, Shawn!

September 21, 2013 A total of $814 was raised in memory of Wyatt DeStephano during awareness week through a combination of efforts, much of which came from Wyatt's sisters: Maggie and Jilly. They made bracelets and green ribbons that they sold at their schools every morning during awareness week. Maggie is in the 6th grade at the Octorara Area Intermediate School and Jilly is in the 2nd grade at the Primary Learning Center. Thanks again to all who helped out!

September 2013 Baja Bistro in Seattle, WA hosted a coins for a cure fundraiser throughout the month of September. Nearly $200.00 was raised for the UMDF in honor of Joe Gabert.

September 2013 Thank you to the North Georgia Mito Families who raised $52.08 by selling "Keep Calm and Raise Awareness for Mitochondrial Disease" t-shirts during awareness week. Your "Boosterthon" was a success!

October 3, 2013 Pacific Gas and Electric (PG&E) held a bake sale which raised $684.71. This was made possible by The Campaign for the Community (CFTC). A special thanks to Linda Wilkinson for spending her time and energy to help out!

October 18, 2013 The first annual Joe Burket Charity Golf Outing was held in Prospect, KY in honor of Joe Burket. Over 60 golfers came out to support the UMDF! Thank you to Chad Miller for hosting the event!

October 19, 2013 A sand art booth was set up at the Publick House Harvest Festival in Sturbridge, MA this fall, and donations were collected for the UMDF. People were able create beautiful sand bottle necklaces! Thank you to the Kalick Family for organizing this event!

October 19, 2013 A total of $880 was raised at the Bake and Craft Sale at the Indiana Mall in Pennsylvania, which was organized by Cathy Chiplis and Diane Brady in support of the EFL: Tampa Bay team - "Eliana's Cause"! Thank you to everyone who supported this event including friends, family and companies that made in-kind donations: Sheetz, Tate's Supermarket, CC Production, and Mohawk Lanes!

October 23, 2013 Colleen Powell made ribbons and handed out fliers and pamphlets to increase awareness about mitochondrial disease. She collected $325 in contributions from those at Simmons Elementary School in Horsham, PA. Thanks Colleen and to all who contributed!

October 24, 2013 Joyce Rothleder, the owner of Ryder's Cup Coffee Shop in Lakewood, NY, collected donations throughout September from customers and employees in honor of Cooper Groves. Thank you to all who helped raise $50!

October 2013 The Jeans for Genes fundraiser in honor of Jolie Reyna and Ben Stewart was super successful in Louisiana. The following elementary schools raised a combined total of $2414.37: Legacy ($560), W.T. Lewis ($405.10), Stockwell Place ($762.16), and Benton ($687.11)! Thank you Tammy Reyna and to all who participated!

November 1, 2013 Thank you to those at Tavares Elementary, especially to Anne Veneziano, for raising $2,000 during Disability Awareness Week in honor of Lex Santo in Tavares, Florida!

November 3, 2013 A cut-a-thon was held at Clyde St. Amand's Hair Design Studio in West Hartford, CT. Over $4,000 was raised for the UMDF. Thank you to everyone at Clyde St. Amand's Hair Design Studio who helped make this event such a success!

November 9, 2013 The DC-Baltimore/ Northern Virginia Chapter hosted the fifth annual Fall Into a Cure Silent Auction & Cocktail Party in Alexandria, VA. The evening was full of amazing silent auction items, dancing and fun. A special thank you to event Chairs Anne Tuccillo and Sharon Goldin for your hard work on this year's event!

November 30, 2013 Bruster's of Ingomar, PA hosted its first annual Light Up Night. Participants enjoyed pictures with Santa, hot cocoa and crafts. Donations were accepted for the UMDF. Thank you Bruster's for your continued dedication to fundraising for the UMDF!

November 2013 W.W. Gordon Elementary School, located in Richmond, VA, held a hat day in which the children wore a hat to school on report card day and collected donations for the UMDF. Thanks to all of the students and staff who helped raise $482!

November 2013 Ansley Odum, a student at Brighton Academy in Overland, KS, is battling it out with mitochondrial disease. She and her friends held a bake sale and raised over $250 towards a cure.

December 3, 2013 The History Film Club at Schreiner University in Kerrville, TX, hosted its first annual Great Gatsby Event. Attendees donned 20's attire and danced the night away while watching the newly released *The Great Gatsby* film. Thank you Karin Roethler for organizing this event!

December 5, 2013 Riot Games partnered with Massive Black for Workshop LA 2013, a four day seminar where top creative professionals from the worlds of entertainment, film, games, comics, and fine art come together to share their techniques and ideas. On the opening night in Los Angeles, an art show and auction was held to benefit the UMDF. Thank you to Massive Black and Riot Games for raising over $3,000 for the UMDF!

December 31, 2013 Kimberli Freilinger hosted a fundraiser called "Mito Masquerade" to bring in the New Year at The Dance & Music Studio in Monmouth, OR. Thanks to the Mito Masquerade attendees who raised $115!

December 2013 Matt Calhoun, from Oxford, GA painted four Christmas cards. Matt's neighbor, Leslie Hopwah, helped out by printing the cards, which were sold at the UMDF Atlanta Holiday Party. In total, 22 boxes were sold, which raised $132 for the UMDF. He was inspired by a local artist at the UMDF Holiday Party two years ago. Thank you Matthew, Leslie, and their supporters!

December 2013 During the month of December, the Dairy Queen of Zanesville, OH, hosted a light bulb awareness campaign. Nearly $250 was raised for the The Dawnta and Levi Kendall Family Research Fund.

January 21, 2014 Sarah's sMILES, an organization formed in honor of Sarah Landrigan, who has mitochondrial disease, hosted a basketball fundraiser for the UMDF. Thank you for raising awareness in the Warsaw, IN, area!

January 2014 The 13th annual Gina Jugs Coins for a Cure fundraiser was held at St. Bernadette's Catholic School! The students collected coins for the month of January in memory of student, Gina Mohan. Over the past 13 years, the students have raised over $18,000 for the UMDF! Thank you to all of the teachers, students, and Sr. Carol for your support!

February 2014 The Energy for Life Walkathon in Houston kicked off our 'Spring' Walk Season with a great start! We had beautiful weather and a good time was enjoyed by all! Thanks to all our teams who showed amazing spirit this year!

February 13, 2014 UnitedHealthcare in Indianapolis, IN, hosted a dress down day and collected donations for the UMDF. Nearly $2500 was raised. Thank you for your generous contribution!

February 15, 2014 Thank you to everyone who participated in Brielle's Basket Brigade, which raised nearly $20,000 in Altoona, PA, in honor of Brielle Harmon! The money was raised through a combination of 200 donated baskets, a donated $1000 Sheetz gift card, and selling t-shirts and baked goods. A special thank you goes out to Diana Rabold.

February 21, 2014 Jessica Ross, a 4th grade student from Cynthia Mann Elementary School of Boise, ID, held a coins for a cure fundraiser to raise awareness of mitochondrial disease. From

Taylor Allen ran the 2014 Los Angeles Marathon in honor of Brayden Burge,raising $1,940 for the UMDF.

February 21st to March 7th, milk jugs were placed in every classroom and students collected spare change donations "in the fight against Mito." The class that collected the most money won a pizza party and also received the movie that was shown at the coins for a cure kickoff event! Thank you to Cynthia Mann Elementary School for your amazing support.

March 1, 2014 In lieu of presents for his first birthday, Evan Clement of Maryville, TN, asked that donations be made to the UMDF in memory of his big brother, Eric Clement, who lost his battle with mitochondrial disease in 2012. Thank you Evan for your kindness and contribution of over $150.

March 9, 2014 Taylor Allen & Kelli Getter were 'Running for Brayden' in the LA Marathon! The ladies ran in memory of Brayden Burge and collected $1,940 in donations on behalf of their dear cousin.

UMDF Upcoming Events

March 15, 2014 The 9th annual UMDF Dinner and Silent Auction in memory of Brittany Wilkinson will be held in Clovis, CA. The annual event is held to benefit the Brittany Wilkinson Research Fund with the UMDF. Brittany was the UMDF's first Youth Ambassador and had done so much to support the UMDF. Contact Linda Wilkinson at *dotoheven@aol.com* for more information!

March 22, 2014 The fifth annual Jackson - Culley MitoWhat 5k will be held in Millington, TN at USA Stadium. For more details, contact Angie Nunn at *angienunn73@gmail.com* or visit *www.mitowhat5k.blogspot.com* .

March 30, 2014 Sydney Mason is holding a skate-a-thon for her Bat Mitzvah and plans to donate all proceeds to the UMDF in honor of Hayley Lieb. For more information or to make a donation, please visit www.umdf.org/activemito/sydpsmom1 .

April 5, 2014 The 11th Bet on Baylee fundraiser will be held in Crooksville, OH! The fun filled day will include a Texas Hold'em Tournament, silent & live auction, a live band, dancing, and much more! To learn more, contact Jody Thompson at *buff2506@hotmail.com.*

April 13, 2014 Think Spring! Bruster's of Ingomar on Perry Highway in Pittsburgh, PA will host their annual Easter Egg Hunt. After the main event, join other participants for face painting, games and crafts. Donations will be collected for the UMDF. For more information, please contact *info@brustersingomar.com* .

April 24, 2014 Bethany Stamper of Creston, OH, will host a four day fundraising event at Cleat's Pub in Wadsworth, OH. This event will consist of a silent auction, poker run, amazing race, and restaurant night, all to benefit Kaidon Andrew Stamper's research fund. For more information, please contact Beth Stamper at *beth07@aol.com*

The 11th annual "Bet on Baylee" fundraiser will be held in Crooksville, OH on April 5.

April 26, 2014 Girlfriend's Journey to a Cure will be held at Crisp Regional Hospital walking track in Cordele, GA! You will have your choice of participating in a fun run/ walk and three separate 5k's! Go to our UMDF event calendar for registration form or call Tammy Clary at 229-322-8725!

May 17, 2014 The 5th annual Breylon Senn 5K Run/Walk/Stroll will be held at Tri County High School in Howard City, MI! The annual event is held on Breylon's 6th Birthday and we will be celebrating with a balloon release in Breylon's memory. Go to *www.umdf.org/breylonsenn5k* for more information!

June 7, 2014 Please join us for the 8th annual Greater Mito Open starting at 12:00 pm located at the Broadlands Golf Club in North Prairie, WI (Hwy 59 & Hwy 83). If you are interested in golfing, sponsoring, volunteering, donating or contributing in any way, please contact Dave Dobke at greatermitoopen@gmail.com.

June 14, 2014 The 2nd annual Nicholas James Torpey Memorial Golf Outing will be held in Macomb, MI. The "Butterfly Classic" will support the Nicholas James Torpey Research Fund with the UMDF. Go to the UMDF event calendar for updates!

June 27, 2014 The second annual Thomas' Golf for a Cure outing will be held in West Bridgewater, MA at River Bend Country Club. There will be a raffle, silent auction and cash prizes. For more information, please contact Jason Schmid at *jsc4424502@comcast.net* .

July 26, 2014 The first annual Carter Buffum 5k will be held at Cascades Park in Jackson, MI! The walk/run, which will benefit the Carter Buffum Research Fund with the UMDF, will be held in the evening! The night event will feature 'Lights for Energy' with glow sticks and fun lights! Registration will be open soon – go to the UMDF calendar of events for updates!

ONGOING Louann Carnahan of River Forest, IL, continues to sell her *Piano Masterpiece* CDs with all proceeds benefitting the UMDF. To purchase this magical CD, please visit *www.umdf.org/pianomasterpieces*.

Upcoming Energy For Life Walkathons

March 22 The first annual Energy for Life Walkathon in Tampa Bay will be held at Al Lopez Park! Join us by forming a walk team or donating to a walker!
www.energyforlifewalk.org/tampabay

March 29 The Atlanta Chapter will be holding their third annual Energy for Life Walkathon at Centennial Olympic Park in Atlanta, GA. Please join members of the chapter to make their walk a success!
www.energyforlifewalk.org/atlanta

March 29 The St. Louis Metro area Mito Group will be holding their fourth annual Energy for Life Walkathon at Tower Grove Park in St. Louis, MO. Join them and help raise funds to find a cure!
www.energyforlifewalk.org/stlouis

March 29 The North Texas Group will be holding their inaugural Energy for Life Walkathon in Downtown Garland Square in Garland, TX. Please consider starting or joining a team today!
www.energyforlifewalk.org/dallasfortworth

April 5 Join us for the fourth annual Energy for Life Walkathon: Nashville at Centennial Park! Sign up today!
www.energyforlifewalk.org/nashville

April 12 Golden Gate Park will be the place to be for the third annual Energy for Life Walkathon: San Francisco! Start your team or show your support of a team!
www.energyforlifewalk.org/sanfrancisco

April 26 The 3rd annual Energy for Life Walkathon: Pittsburgh will be held along the North Shore at The Great Lawn – Heinz Field. We are walking early this year – so be sure to get your team registered today!
www.energyforlifewalk.org/pittsburgh

April 26 The Binghamton Group will be holding their third annual Energy for Life Walkathon at Otsiningo Park in Binghamton, NY. Please join the group for a fun filled day of awareness activities and networking.
www.energyforlifewalk.org/binghamton

May 10 The Evansville Walk Committee is excited to announce the third annual Energy for Life Walkathon at Burdette Park in Evansville, IN. We would love for you to join us again this year! Register online today!
www.energyforlifewalk.org/evansville

September 13 The Indiana Walk Committee is excited for some changes for 2014! Due to popular request, we have moved to the fall! We have also found a new location this year, Hummel Park in Plainsfield, IN, is excited to have the Energy for Life Walk: Indianapolis become its new home! Stay tuned to our website for details!
www.energyforlifewalk.org/indianapolis

Fall Walks

Save the date for the Fall 2014 walks! These websites will be live soon!

August 16
Minnesota

September 6
Detroit

September 13
Indianapolis

September 20
Western New York

September 20
New Orleans

September 20
Omaha

September 20
Columbus, GA

September 21
Chicago

September 27
Delaware Valley

September 27
Milwaukee

October 4
Akron

October 18
Charlotte

October 19
Central Texas

TBA
Kansas City
Birmingham

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.

Test Your Mito IQ!

1. **What does "mitochondria" mean and what is their purpose?** The word mitochondria derives from the Greek mitos meaning "thread" and chondrion meaning "granule." They are thought of as being primarily energy factories but have a role in aging, cell death, cell signaling, activating the immune system, and combating reactive oxygen species (chemically-reactive molecules containing oxygen).

2. **Are mitochondria in every cell of the body and how many are in each cell?** There are hundreds to thousands of mitochondria in every cell in the human body except for red blood cells.

3. **Do mitochondria look and function the same in different organ systems?** Mitochondria have a different appearance and slightly different primary function depending on their location in the body.

4. **Are mitochondrial diseases considered rare or orphan diseases?** While primary mitochondrial disorders are thought of as being rare, they are present in at least 1 in 5000 individuals - which make them less rare than previously believed. A recent study showed that 1 in 200 healthy infants carry a potential disease-causing mitochondrial DNA mutation though this does not mean that 1:200 infants will develop mitochondrial disease. The majority of these infants do not have a mutation affecting most of their mitochondria, thus they will not develop disease.

5. **Can someone spontaneously develop mitochondrial disease, and if so, how?** When we speak of primary mitochondrial disorders, these are typically genetic and either inherited or new changes in the patient's DNA and something present from birth even though symptoms may present at any age in life. We know that certain environmental toxins and poisons as well as childhood and chronic diseases can injure the mitochondria and create select mitochondrial symptoms - though these are not typically considered primary mitochondrial disease but rather "secondary mitochondrial dysfunction."

6. **What is adult onset mitochondrial disease?** Mitochondrial disease presenting in adulthood. Sometimes the symptoms begin all-at-once; sometimes "milder" symptoms have been present for quite some time (diabetes, hearing loss) and not tied together until a more "red flag" mitochondrial disease symptom occurs (stroke for example).

7. **Which is the most common form of mitochondrial disease?** In truth, we do not completely know. Leigh syndrome is one of the most common type of pediatric onset mitochondrial disorders. MELAS is one of the most common adult-onset ones. However, we need better research on the prevalence and incidence of these conditions.

8. **What is heteroplasmy and can it help determine severity of the disease?** Each cell has hundreds to thousands of mitochondria and each mitochondria has hundreds to thousands of mitochondrial DNA (mtDNA) copies. All the mtDNA blueprint copies are not identical - some are "healthy" and some are "unhealthy." Heteroplasmy refers to this mixture of healthy and unhealthy copies of mtDNA impacting how well each mitochondria work. A certain threshold of unhealthy mitochondria need to be present before leading that organ to not work properly.

9. **What can harm or hurt mitochondria?** Too many things can hurt mitochondria -> Poor diet, lack of exercise, too much alcohol and illicit drugs are the most common culprits. Chemotherapy and radiation are others. Many medications can down-regulate mitochondrial function in a petri dish - but we do not yet know if they necessarily hurt mitochondria enough to actually lead to symptoms; there are exceptions -> some - like valproic acid - can bring out inactive mitochondrial disease and select anesthetic agents may exacerbate symptoms. Inflammation and infections can impact mitochondrial function but these are not always avoidable things.

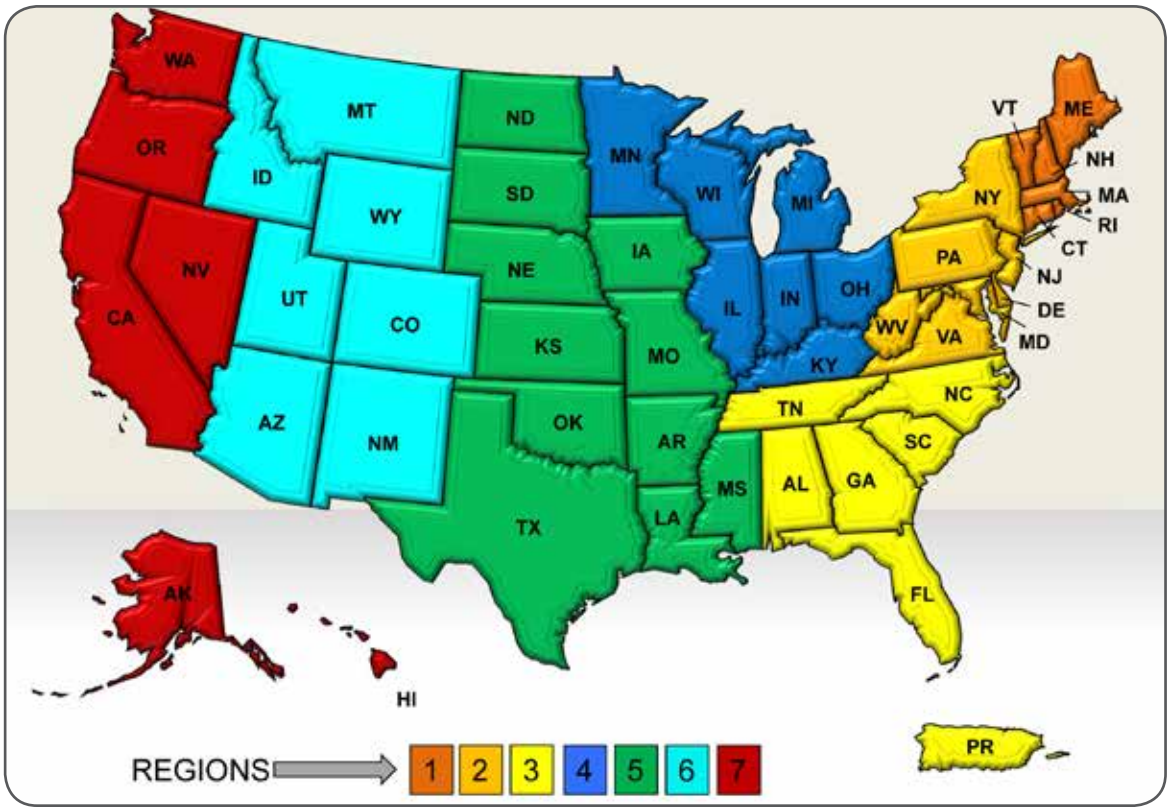
10. **What can help mitochondria function better?** Exercise, a healthy diet, and avoiding mitochondrial toxins are some things that improve mitochondrial function. There are components in red wine (resveratrol) and chocolate (epicathecin) that help. Other anti-oxidants may but we do not have enough scientific evidence yet that they work for every patient.

11. **Currently, how many mitochondrial-related clinical trials and/or studies are ongoing worldwide, and which show the most promising results?** EPI-743 is the only medication currently in clinical trials for select types of mitochondrial disease. The North American Mitochondrial Disease Consortium is a group of multiple centers in the country and led by Dr. Hirano at Columbia University that is advancing clinical research in mitochondrial disease as well. According to the UMDF's Decade of Difference, there are 305 clinical trials underway across the globe.

12. **Realistically, when will breakthroughs for treatments, and ultimately, cures happen?** We hope these changes occur soon. Mitochondrial disease is such a young field of medicine - with the 1st mitochondrial disease only identified in the 1960s and most of the research truly occurring in the last 20 years. In the world of science, we have a long way to go. For comparison - we have known about diabetes since the BC times and diseases like Alzheimers for 200+ years - and we can contrast how far these fields are in regards to disease diagnosis, management and treatment (and these are diseases that receive much more research funding). But with the help of the UMDF and a large number of dedicated clinicians and researchers we are moving in the right direction.

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!



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VERMONT
- MaryBeth LeFevre

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