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UNITED MITOCHONDRIAL DISEASE FOUNDATION®

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Caitlyn's Crusaders cheer on Caitlyn Yoder as she walks across the Start Line for the very first time with her mother, Jennifer, at the 2016 Energy for Life Walk in Charlotte, NC.

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From the Chairman

by Brent Fields, UMDF Chairman

As my first "Chairman's Message", I am delighted to have the opportunity to share some of my thoughts with you in my new role with the United Mitochondrial Disease Foundation. In the fall of 2016, I was nominated to the role of Chairman of the Board of Trustees. I was honored to accept the position and humbled by the confidence placed in me by my fellow board members.

Please allow me to thank our past chairman, Patrick Kelley. Over the past few years, he laid the ground work to position the UMDF as a leader in the search for treatments and cures. It was under Patrick's leadership that the Mitochondrial Disease Community Registry (MDCR) was launched, our new strategic plan was created, and we began the "Roadmap to a Cure", which is something that you will be hearing more about as we progress through the year. Patrick remains a UMDF Board Member and valuable champion for the mission. UMDF has benefited from many passionate, talented and committed trustees over that past twenty years, and I am honored to carry that torch forward in gratitude for their service. Dan Wright, who recruited me to the board several years ago, is another great example of the tremendous past leadership that brings us to this transformational place in the life of the organization.

Like many of you, my involvement with the UMDF began with the question, "What is mitochondrial disease?" My wife, Suzette, and I were looking for answers to the many questions we had about our youngest, Chloe, who battles mitochondrial disease. We found tremendous resources, support and information with UMDF.
We eventually made the decision to get involved as donors, advocates and reaching out to other parents who were experiencing struggles similar to ours.

Suzette and I proudly support the UMDF efforts in our Central Texas Region. In the past five years of participation with the local Energy for Life Walk, our Team Chloe crew has rallied resources to help UMDF make a difference for everyone who is or will struggle with a mito disease, not just our daughter or those in our community.

In my time of involvement with UMDF, I've been amazed and inspired by those affected individuals, family members, trustees, donors, staff, scientists, clinicians, and advocates who have done so much to support the mission. Meeting so many of you at the symposiums, reading your stories in this newsletter and hearing about your journey from staff members is a constant reminder of what a diverse and powerful community we are. While our mito experiences and UMDF contributions may look different, we are united in our commitment to do work that leads to the best possible care, support, treatments, and eventually a cure.

As the Chief Executive Officer of Big Brothers Big Sisters in Central Texas, I understand firsthand the challenges and successes of a non-profit like the UMDF. I hope to use my experience as a former hospital administrator and Vice President of the American Heart Association to contribute leadership that will support our continued progress. Chloe is the reason we came to know about and need UMDF. I will continue my service, inspired by you, am honored to serve with you, and am committed to work diligently on your behalf!

Yours toward a cure,

Brent Fields

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In Memoriam

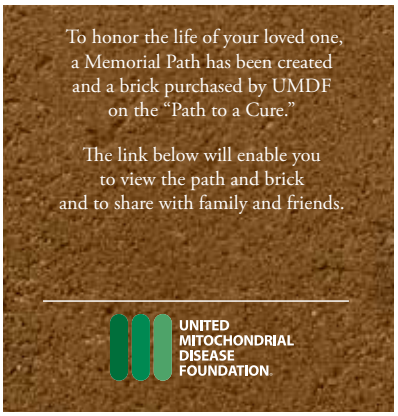
The UMDF is saddened to learn the following have lost their battle with mitochondrial disease. Below are the names of those who, according to our records, became Mito Angels between January 1 and February 28, 2017.

Anthony Varga – Niles, MI
Jennifer Davis – Austin, TX
Genesis Fierst – New Castle, PA
Caroline Llarena Neumann – Pembroke Pines, FL

UMDF has created a brick in memory of each on UMDF's Path to a Cure. To visit each path, go to *www.umdfpathtoacure.org*

UMDF Path to a Cure

The UMDF is relaunching our Path to a Cure as a part of our Grief Support Program. If your loved one has passed from a mitochondrial disease, you and your family are added to our Grief Support Program. This program currently includes a sympathy card, a tear soup book one month after the passing date, a birthday card on your loved one's birthday, and an angelversary card on the year mark of their passing date. UMDF's updated program will now include a 'brick' inside the initial sympathy card with a unique website link to your loved one's path that you can share with family and friends. The first brick will be started by UMDF. Questions or comments about the Grief Support Program? Contact Tara Maziarz at *TaraM@umdf.org*.

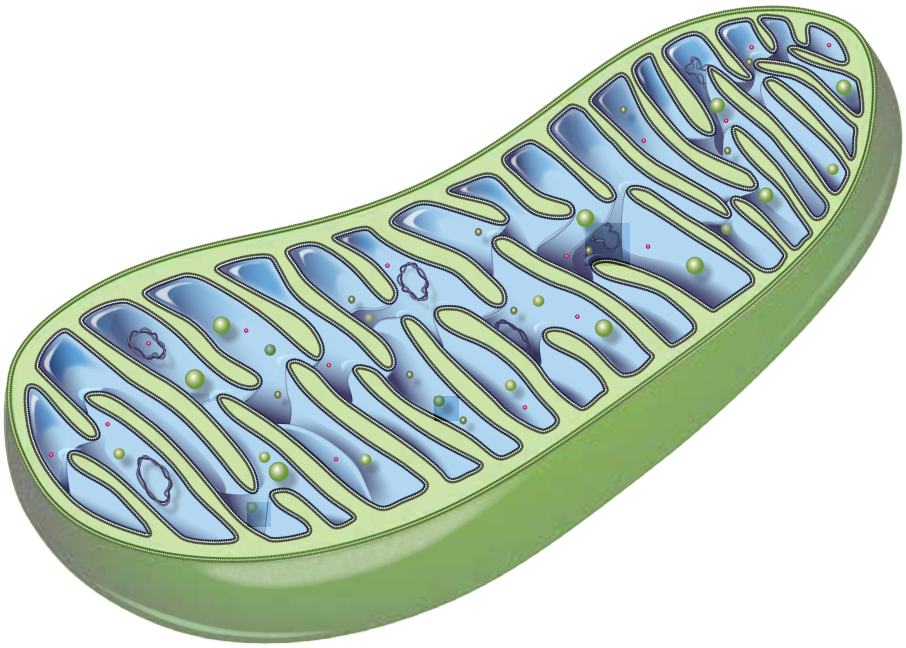


Day on the Hill

Will you join us on June 29, 2017 for our fourth "Day on the Hill?" This is your chance to meet representatives from Congress and the United States Senate and advocate for issues important to the mitochondrial disease community. Among the talking points will be issues centered on the repeal and replacement of the Affordable Care Act and issues that we believe can be enhances in the Cures legislation. Please register now by visiting *www.umdf.org/symposium/day-on-the-hill*. Please complete the registration to help us schedule transportation and an appointment for you on Capitol Hill. Because information included in the registration is required by the House and Senate, we will need to discard registrations that are not completed. Also, we cannot guarantee that you will have a meeting scheduled with your Congressman or Senator. It has been our past experience that speaking to a member of his/her staff is just as important. We hope to see you on Capitol Hill in June!

Stealth BioTherapeutics

is committed to the development of therapies for mitochondrial disease and proudly supports the advocacy efforts of the UMDF



To learn more about our work, please visit StealthBT.com or follow us on social media:

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Affordable Care Act Update

We have been hearing a lot in the news about the Trump Administration's plan to repeal and replace the Affordable Care Act (ACA), also known as Obamacare. The administration says it plans to work with Congress to make sure a series of reforms are ready for implementation. President Trump would like to see a plan that he says will "follow free market principles and that will restore economic freedom and certainty to everyone in this country. By following free market principles and working together to create sound public policy that will broaden healthcare access, make healthcare more affordable and improve the quality of the care available to all Americans."

Of course any changes in health care laws could have an impact on the mitochondrial disease patient community members and their families. Because of that, UMDF has concerns, too.

Many who suffer with mitochondrial disease have historically experienced difficulties obtaining affordable health care coverage. The requirements of the ACA regarding robust coverage of those with pre-existing conditions under family, individual and small group plans has been particularly important for many patients and families.

While UMDF's preference would be to leave the ACA's current coverage scheme in place and work on sensible modifications to address premiums, coverage and other issues, we recognize that Congress and the President are likely to make more dramatic changes. You can rest assured we will be advocating for this issue on your behalf on Capitol Hill. However, during this process, let your voice be heard.

Visit the UMDF Advocacy Action Center and send a letter to your Congressman and U.S. Senators asking them to consider some important issues affecting mitochondrial disease patients and families as they work to repeal and replace the ACA.

UMDF recommends Congress consider the following as they work to "repeal and replace":

- Policies sold in the individual and small business market must provide guaranteed, non-discriminatory access at regular rates to individuals with pre-existing conditions.
- Insurance policies should provide adequate and comprehensive coverage, without lifetime or annual limits.
- The option for parents to keep their children on the family policy up to age 26 should be preserved.
- Incentives for State Medicaid plans to cover adults with comprehensive benefits should be preserved.

We will advocate for these policies on Capitol Hill. But, we need you to do the same. Visit the UMDF Advocacy Action Center at *www.umdf.org/advocate*. It is there you can see our position and follow a link that allows you to visit the UMDF Advocacy Action Center to send a letter to Congress urging them to consider these important points as they work on a new health care proposal.

UMDF Flashlight Program

Think back to the first time you attended a UMDF Symposium. There is a lot of information presented and in some cases, patients and parents go back to their hotel room feeling a little overwhelmed. There are meetings, questions and answers to be had. UMDF wants to make it easier for first time attendees to participate in the UMDF Symposium. That is why we are instituting the UMDF Flashlight Program. The program will pair a new symposium attendee with an ambassador who is a veteran attendee. They will help guide the new attendee to the right information and help shine the light on some of the concerns and questions that may pop up about symposium. If you are a veteran symposium attendee who would like to serve as a UMDF Flashlight, contact us at 888-317-UMDF or connect@umdf.org.

REATA LAUNCHES A MITOCHONDRIAL MYOPATHY CLINICAL STUDY

Reata is the leader in developing antioxidant inflammation modulators (AIMs), which are potent activators of the biological transcription factor Nrf2.

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Nrf2 controls the body's production of hundreds of antioxidative and cytoprotective molecules. Activation of Nrf2 has been shown in non-clinical studies to improve mitochondrial function and energy production, which are impaired in Mitochondrial Myopathy. To learn more about Reata Pharmaceuticals, visit **reatapharma.com.**

Clinical Study on RTA 408 Capsules

Reata Pharmaceuticals is actively enrolling the MOTOR study, a placebo- controlled, multicenter clinical study of RTA 408 capsules in Mitochondrial Myopathy. For more information, go to **clinicaltrials.gov** and search "RTA 408 MOTOR".

MOTOR
a study in mitochondrial myopathy

Spotlight: Team Zoe

Zoe, our 8-year-old daughter, has Mitochondrial Disease. It is difficult for her to vocalize, swallow and move. She does not crawl, walk or talk. But her spirit is strong, and she is a fighter! Zoe loves to make choices and interact with her peers. She loves books, swimming and hanging out with friends and family, especially her 11-year-old brother, Noah. She has a smile for everyone and wants to be seen, included and loved….like we all do.

Zoe was diagnosed in March of 2012. Even though we are a family practice doctor and a registered nurse by trade, we had never heard the words "mitochondrial disease." It is a devastating diagnosis.

Although it has been a difficult road, Zoe amazes us every day with her strong spirit and her will to live life to the fullest of her ability. She is now in second grade, which is a wonderful milestone! Zoe's determination and persistence encourages all who know her! Without using words, she makes us want to work harder in our constant battle to raise awareness and funds to support mitochondrial disease research! Zoe is doing great at the moment and loving her life! We are grateful for every day because life is precious and Mitochondrial Disease is unpredictable. We must find a cure & we will not give up!

In 2012, we formed "Team Zoe" for our local Energy for Like Walkathon. We wanted support most of all, but also understood the importance of raising awareness of mitochondrial disease. Every year we send out an informational card with Zoe's picture on the front and we send e-mails to our family, friends, community and school to raise awareness and encourage people to walk and or donate in support of Zoe and mitochondrial disease research.

This year we also held "Concert for a Cure –Backyard Party" at a friend's home, which raised over $5,000. It was such a success that we plan on continuing this new tradition!

September 2016 was our fifth year participating in the walk and was our biggest year yet! Over the years, Team Zoe has raised almost $50,000.00 and typically have over 100 participants. We look forward to walking with "Team Zoe" 2017!

Thank you,

Cindy & Joel Kavan

I received this story while I was struggling to find an interesting topic for this issue of the newsletter. We have been doing a lot of traveling visiting many members across the country and discovered that many of our members have courageous and interesting stories. I invited a few of them to write and send me an article about how they live with their mitochondrial disease. This first story is from a special friend and very courageous mitochondrial warrior, **Rachael Pipp.**

Dr. Wayne Dyer said, ***"When you change the way you look at things the things you look at change!"*** Rachael is living proof.

Chuck Mohan

Behind My Beautiful Blind Eyes...

by Rachael Pipp

My eyes aren't typical seeing eyes anymore. I truly miss my vision. It's been over two years now, but behind these beautiful blind eyes I can still remember and imagine what the world looks like. I have memorized all the colors of the rainbow and colors of my tie dye shirts that I made for my UMDF EFL Walk Teams; I can still picture all those beautiful things in my mind.

Chuck and Rachael at the 2015 Symposium

Like most of you I sleep, I eat, and then I repeat. Unlike most of you I don't work, not because I don't want to but because I can't. I have mitochondrial disease and so I have good days and I have bad days. I had to learn to listen to my body, which I'm much better at now than I used to be. It took a long while for me to learn that I am not like others. I cannot overdo activities or refuse to take naps. I finally realized why I would feel sick for days at a time and how normal activities caused me to crash. I have now learned to embrace my naps and how to conserve and make use of my energy, saving it when I need it the most. I am learning how to control the disease and how to be part of the CURE.

I've always been an artist and I miss creating all my art. I can still make some art by feel, but it's just not the same when I don't get to see the process or the finished piece. I want to see the beautiful bright colors and not just imagine them; you can't feel colors. I use to love to paint colorful character pictures and make purses, wallets and flower pens out of colored Duck Tape. I even made a Duck Tape Mito Tie for UMDF CEO Chuck Mohan. Sometimes I can still get a glimpse of colors and shadows and sometimes I can even see XXLarge letters out of the corners of my eye. I really miss seeing everything, but especially I miss the faces of all the people I love.

Since losing my vision I can't spend as much time on my art, so I had to think of something else to keep me busy and my recumbent bike really helps me do that. Riding takes time and energy and then I need more time and rest to re-energize. I really enjoy listening to music and books on tape while I ride and when I ride I don't think about my mito disease. I pretend I am riding outside enjoying the fresh air and seeing the trees and sky. The exercise really makes me feel energized and when I'm energized I feel healthy and when I feel healthy I am happy. I drink lots of Normalyte, Gatorade and water while riding. I keep my Mitochondria activated by trying to ride 6 miles almost every day. Riding makes me feel free!

Before I start my bike ride, I need to prepare more than most riders who do not have mitochondria disease. I have to have a good night's sleep, then I need my breakfast which is usually Cocoa Wheats with peanut butter, so yummy. Then it's time for

my medicines, especially my POTS ones so my heart doesn't race so much, then come my supplements and then a good long nap to digest it all. When I wake up and if I feel up to it, I hop on my bike for a 6 mile ride. It takes me about an hour to ride 6 miles and I can't do it every day, though I try. Some days I record my miles on the Charity Miles App because my goal is to add enough of them to help get UMDF as one of the charities listed to ride for and get listed on the Charity Miles App. Helping others helps me control my disease and helps me be part of the CURE.

After I ride, I need more food, usually a packet of Real Foods Blends that I take in my G-tube. My favorite is the beef, potatoes, peas and pineapple. Then, after all that, I take my shower and another nap! When I wake up it's almost dinner time! Even though riding wears me out, it also refreshes me and gets my mind off my disease and makes me feel free; it puts me in control.

Rachael riding her exercise bike

My comfy bed is my happy place where I dream and wish I could see. I dream and hope for a research grant about stem cells that could replace my optic nerves so I can see again in my lifetime. I have a fear that I may never see again and I continue to struggle with my sadness about this from time to time. Sometimes I wonder, "Why me, why can't I see like my other friends." I think maybe I was meant not to see, but I don't know, maybe this disease is just unfair. No, not maybe, mitochondrial disease is unfair. It robs you of different parts of your body and I don't like navigating this beautiful world through this dark tunnel.

I wonder if having mitochondrial disease and losing my vision has made me stronger. I do know it makes me work harder and when I work hard I really feel optimistic in spite of it all. I still try to live life to the fullest and make a difference for all of us with mito. I do my best to keep myself pumped up and I advocate for treatments and cures. It motivates me get out there and raise more money for UMDF. It's the best thing I can do for myself and others and it helps me control the disease and be part of the CURE and not a victim.

When I'm in the hospital I do my best to be brave. I try not to be scared and even if I am, I just tough it out as best I can because I know the doctors and nurses are there to help me. Sometimes you have to respectfully advocate for what you or your family knows you need. Sometimes the nurses and doctors don't understand your Mitochondrial Disease or certain things about yourself personally that you know will help. The doctors and nurses think I am brave and always tell me that my smile lights up the room and brightens their day. Sometimes you just have to smile. Sometimes the smile is the best medicine. The smile helps me control the disease.

I try to always say please and thank you to all my caregivers who help me at home, and especially my mom. I have the disease but my mom is also affected by it so I must always try to be grateful, cooperative and flexible about what needs to be done and I find that they are cooperative and flexible with me in return. I guess it's important for the patient to have a good bedside manner as well as the doctors and nurses and all those that care for us.

My mitochondrial disease is showing me that attitude is everything and makes a huge difference helping me to look at what I can do rather than what I cannot do. Smiling and laughing and appreciating others is important. Sometimes I like to give little surprises to these special people and sometimes they do the same for me. I'm like Olaf, I like to give and receive warm hugs and so do most of the people I work with.

Everyone likes to know they are appreciated and important and being polite and courteous are two things that mitochondrial disease does not take away. I may not be able to see, but I know when you are smiling. And when you smile you help me control my disease and help me stay part of the CURE!

Member Spotlight – The Lee Family

When Elizabeth and JungHo met it was as if fate intervened. They both moved to Raleigh, NC at a young age, Elizabeth from Wisconsin and JungHo from Korea and then Washington D.C. They both attended the same high school but did not know it. When they met as adults it was at one of JungHo's kickboxing classes at his Tae Kwon Do studio, they fell in love and were married. 14 months after their marriage their precious son, Aiden, was born and they could not have been happier. However, when Aiden was 11 months old he started to have medical complications, and he was admitted to the hospital. Five days later, he was released with the diagnosis of Adrenal Insufficiency. By the age of two, Aiden was suspected of having seizures, but the doctors could never catch them on an EEG.

During these struggles with Aiden, the Lee family was blessed twice more once in 2008 with a girl and again in 2010 with another boy. Aiden loved being a big brother to his siblings, and while health problems and doctor visits increased, he did not let that stop him from playing, laughing, and doting on his younger siblings. Shortly after their second son was born they were able to catch the seizures on the EEG, Aiden was diagnosed with Epilepsy. This was also when the Lee's received the news that Aiden had a mitochondrial disease, POLG1.

Elizabeth immediately went to the internet to research the condition. There was not a lot known about Polg1, then she found the UMDF website, and the UMDF became a comfort zone for her and her family as she did not know what the future would hold. Aiden was a happy and gentle little boy who loved life, he was able to go to school, play and did everything expected of a child, even participated in Tae Kwon Do lessons.

On May 3, 2014, Aiden had a severe seizure that lasted 13 hours before the doctors could stop it. He was put into a medically induced coma. He spent 3 months in the PICU at Duke Hospital before being moved to the step down floor, where he spent another month. Aiden was unable to talk, move, eat, or unable to do any of the things he so loved to do. Aiden was then transferred to Levine Children's Hospital to work on rehab, The Lee family spent one month there before realizing he had reached his limit. On November 6, 2014, Aiden passed away surrounded by his loving family.

The Lee family has taken their tragedy and turned it into helping all those affected with mitochondrial disease. They established a Family Research Fund through UMDF and have been actively fundraising. This past year JungHo's foundation, The Sidekick Foundation, turned one of their yearly events into a UMDF fundraiser, raising $20,000 for mitochondrial disease research! The Sidekick Foundation does a different fundraiser for a different cause every year, many times for local charities in their community. The Lee family however has been fundraising on their own for UMDF since Aiden was in Pre-School. The first fundraiser was a community walk hosted by Aiden's Montessori School.

This year's major event was for UMDF and it was one of the most unique fundraiser's to date. On September 24, 2016 during Mitochondrial Disease Awareness Week, an Adventure Race was held, MAD Dash. This race included physical as well as mental challenges, such as brain teasers. There was also a smaller scaled race for all the kids! This event promotes all of the things the Sidekick Foundation encompasses, it teaches the

participants about teamwork and giving back to their community. Aiden's brother Jaxson, had his own team, and won their group this year, making the day even more special. Jaxon said, "I'm happy my team won and I know my brother is happy too." Aiden's sister Samantha also has a very special connection to the event. "The MAD Dash was very special to me, I like how all my friends and family come together to raise money for UMDF. I want the money to help all the other families with mitochondrial disease so their family members can come home healthy," she said.

The Lee family works diligently to not only fundraise for research, but to raise awareness about mitochondrial disease. Their mission is to help all other mito warriors and their families so that they will never have to go through a loss like they have. Even though their son is no longer here, they do not want their connection with UMDF to go away, "It is a part of our family, and always will be", said Elizabeth.

The Lee's have lived the past two years with a piece of their family missing. Looking back, they are able to see all that Aiden taught them. He always saw the beauty in people and in the world around him, and he taught those around him passion, perseverance and love.

Roadmap Updates

by Philip Yeske, PhD - UMDF Science & Alliance Officer

The Trustees of UMDF long expressed an interest in creating a "Roadmap" for treatments and cures for mitochondrial disease. For many years, this was an unachievable desire, as the scientific knowledge base did not exist upon which to credibly develop such a plan. In recent years, the situation changed due to advances in scientific knowledge; so, in 2016, the UMDF staff and the Scientific and Medical Advisory Board of UMDF took on the challenge of creating a first version of the roadmap.

The first step was to define project scope. In this regard, all involved in the discussion felt it was imperative to broadly cover the patient experience with mitochondrial disease. We settled on a three-pillar approach: 1) Diagnosis, 2) Development of treatments and cures, and 3) Patient care. With these three pillars defined, the next step was to assess the current status of each, identify what resources are already in place, and describe gaps to be addressed by the Roadmap. A report summarizing this work was presented to the UMDF Trustees in June 2016 and adopted at that time as guidance for setting UMDF program priorities.

Two key existing assets within the Roadmap being stewarded by UMDF are the Mitochondrial Disease Community Registry (MDCR) and the Research Grant Program. The goals of each of these programs have been aligned with the Roadmap. Here are a couple of examples:

MITOCHONDRIAL DISEASE COMMUNITY REGISTRY (MDCR)

GOAL: Utilize MDCR as a tool for identifying and characterizing patients to improve diagnoses of mitochondrial disease (Diagnosis Pillar)

ACTION: Integrate and protect the collection of genetic testing data to facilitate research into defining various forms of mitochondrial disease

STATUS: UMDF is investing in the buildout of an electronic consenting process (eConsent) for a research study that allows MDCR registrants to approve the sharing of genetic testing results into a database that researchers will be able to query in aggregate. Consistent with the charter of MDCR, registrants that join the study will have full control of who can see their anonymous data, analyze said data and potentially contact them about other relevant research studies.

WHY THIS IS IMPORTANT: The most effective biomedical research requires health information, genetic data and biosamples. The MDCR eConsent process will be piloted on genetic data and then applied to creation of a robust patient biosample repository in follow-up work.

TIMELINE: Rollout of eConsent for genetic testing study will be in the second half of 2017.

RESEARCH GRANT PROGRAM

GOAL: Identify and fund the most promising research opportunities related to the development of treatments and cures for mitochondrial disease (Treatment and Cures Pillar)

ACTION: Broaden the scope of opportunities considered and associated funding mechanisms

STATUS: In fall 2016 UMDF leadership decided to temporarily pause the Research Grant Program in order to consider a more efficient submission/review process as well as a fuller range of funding options. Academic grants will absolutely remain an important component of research funding. Other possibilities, including the funding of pilot clinical trials and internationally-partnered grants continue to be evaluated.

WHY THIS IS IMPORTANT: With limited resources and capital, it is critical for UMDF to carefully, but aggressively, allocate its research investments. For example, increased industry attention on mitochondrial disease provides an opportunity to advance early clinical studies as opposed to being focused solely on bench research. Partnering also continues to play an important role in the UMDF strategy to increase the impact of the foundation's research programs.

TIMELINE: The foundation anticipates announcing several academic research grant awards under the new programs at the June symposium.

These are just two examples of how UMDF is utilizing the Roadmap. The Roadmap is a "living document" and will continue to be updated over time. A next goal will be to partner with other mitochondrial disease-focused organizations to create a community-wide Roadmap that takes advantage of each respective organization's strengths and resources. Watch for further updates about the Roadmap in future articles.

Corporate Partnership Spotlight

TJX Companies is the leading off price retailer of apparel and home fashions in the US and Worldwide. Their stores include T.J. Maxx, Marshalls, HomeGoods and Sierra Trading Company. We have all been to one of those stores, right? Did you know that TJX and specifically TJ Maxx –Atlanta area, have been amazing supporters of the UMDF for 10 years? These folks are so much more than corporate partners, they are a part of the UMDF Family.

It all started in 2007 when Atlanta area UMDF Member, Katy Lyons, had a chat with her neighbor, Anna White. Anna, a TJ Maxx employee, told Katy about TJX Corporation grants. The UMDF applied, and received a grant and the relationship began. The employees of TJ Maxx in the Atlanta Market began to get to know our Atlanta area UMDF members and wanted to do more to help. At first, they helped by bringing toys to our local UMDF children at the Annual Atlanta Holiday partner. This event was once small but has grown to be a very large event hosted by UMDF Members Chris and Mary Swinn at The Peachtree Club in Atlanta. While the size of the event has changed- one thing has not...TJ Maxx. Every year they provide carefully selected gifts for our Members…

But that is not all…
Every week, all year long, the TJ Maxx associates in the Atlanta area raise money in their stores to donate to the UMDF at the Holiday Party and at the Energy For Life Walk in Atlanta. How do they raise the money? They have bake sales, host $1.00 dress down days every Friday, Saturday and Sunday, have packaged lunches with the proceeds going to the UMDF. They started out by doing this in 12 stores and now 38 stores participate. All of this is organized by a dedicated "Charity Champion". To date, including TJX Foundation grants, they have raised over $50,000 for the UMDF!

Chuck Mohan, UMDF CEO, Beth Whitehouse, UMDF Director of Development, Margaret Moore, UMDF Southwest Regional Coordinator and local Atlanta area families had the privilege of being invited to a TJ Maxx store in Atlanta for a meet and greet breakfast in September of 2016! It was an amazing experience to meet the TJ Maxx employees that work so hard for us all year long. It was so heartwarming to know that the associates care so much about the mitochondrial disease community. Thank you ,TJ Maxx , for all you do for the UMDF!

"For some of the mito families, this is one of the only times they are able to take their child(ren) out to a gathering. TJ Maxx has made it possible for us to not only have a gathering that includes food and Santa, but also enables us to provide gifts to all the children (siblings too) and adults who are living with mitochondrial disease. It warms my heart when I see a child choose a gift from the gift table and then go right to the TJ Maxx table to thank them for being so generous.

"My daughter, Emily, who is now 21 years old, has been attending this event since she was a young child. She has grown up around many of the TJ Maxx folks and every time they see her, she is greeted with loving hugs and smiles.

TJ Maxx has taken our families in as their own. They participate in many UMDF events. My family is thankful for their love."

Mary Swinn – Atlanta, GA

"After meeting the TJ Maxx team I can understand why success is not only measured on the P&L but also by building strong community relationships. TJ Maxx is a fantastic example of a company with a focus on community. Your support of our mission is greatly appreciated."
Charles A. Mohan, Jr., CEO/Executive Director

"I was so excited for the breakfast meeting we had. It was the opportunity to increase awareness and include more people from TJ Maxx in our efforts. It was like a fabulous family reunion. Both sides of my family meeting and joining forces. I love my employer TJ Maxx and I work with some of the finest people in the world. It was wonderful to see them bond with my MITO family."
Lori Mason ,TJ Maxx Charity Champion

Donor Spotlight

July 1 - December 31, 2016

The United Mitochondrial Disease Foundation thanks our many individuals, organizations, foundations and companies who so generously support our mission. We've updated our Donor Honor Roll Giving Society to now include cumulative giving throughout our fiscal year versus what was previously reported on a quarterly basis in our newsletters. We have also included the number of years of support to UMDF next to each donor's name. We strive for accuracy and completeness. Please send any questions or suggestions to info@umdf.org

Visionary Investors
$100,000 and above

William Wright Family Foundation (13)

Power Investors
$50,000 - $99,999

Stealth BioTherapeutics (3)

Energy Investors
$10,000 - $24,999

The Breslow Family (6)

CB&T Bank *

Charlotte Pipe & Foundry Company (5)

Mr. and Mrs. Chris Florio*

Mr. and Mrs. Pat Geary*

Gensight Biologics*

The George W. Bauer Family Foundation (1)

Mr. and Ms. Thomas Hefferon (19)

Mr. David Heikkinen and Dr. Ann Heikkinen (2)

Hilton Worldwide, Inc. (1)

Mr. Peter Kelley (11)

Kelley Management Consulting (8)

Lee Brothers Sidekick Foundation*

Mr. Sebastiano Lopresti & Family (5)

PNC Charitable Trust*

Rachael's Gift Inc. (6)

RA Kirby Foundation (1)

Reata Pharmaceuticals, Inc. (2)

Robert J. Bauer Family Foundation (2)

Tishcon Corp (9)

William S. Kallaos Family Foundation (3)

Hope Investors
$5,000 - $9,999

Austin Canvas & Awning (5)

Mr. Joseph Auth (13)

Barth Syndrome Foundation Inc.*

Mr. Brett Cohen and Dr. Gwen Cohen*

Mr. John Duffey*

Edison Pharmaceuticals Inc. (4)

Mr. Bruno Granville*

GVM Associates Inc.*

Mr. Walter Hawrylak*

Henry Lea Hillman, Jr. Foundation*

Jay Roberts Jewelers*

JDM Fund (7)

Kendra Scott Design (1)

Mr. and Mrs. Gordon Kidd (15)

Mr. and Mrs. David Langer (19)

Mrs. Molly Auth Manning (12)

NIEHS (3)

Nikos S. Kefalidis Foundation, Inc.(10)

Pearce Family Foundation*

Raptor Pharmaceuticals (3)

Mr. and Mrs. Matthew Richardson (4)

Mr. and Mrs. Brent Staples (6)

Dr. Annette St. Pierre-MacKoul MD (3)

VOYA (2)

The WaWa Foundation*

Dr. and Mrs. Harry Weinrauch (11)

Friends
$1,000 - $4,999

3M Foundation (1)

Mr. and Mrs. Randy Adams*

Akron Children's Hospital (4)

Allegis Group Foundation (3)

Alternative Christmas Fair Evergreen Lutheran Church*

American Junior Golf Foundation*

Anthony Baldi & Associates (3)

Appalachian Disc Golf Association*

Arena Sports, Inc. (1)

Auction Masters (1)

Mr. and Mrs. Tim Babiarz (2)

Babiarz Law Firm P.A. (9)

Mr. and Mrs. Tommy Baker (10)

Mr. Anthony Baldi (5)

Ball Corporation (6)

Baxter International Foundation (4)

Baxter Pharmaceutical Solutions LLC (3)

Mr. and Mrs. Mike Bech (11)

Dr. and Mrs. Robert Belfer (10)

Mr. John Belk (5)

The Bell Family*

Mr. and Mrs. Andrew Benney*

Bernard's Salon & Day Spa*

Bill & Melinda Gates Foundation*

Bradley Arant Boult Cummings (3)

Mr. Mark Braverman (4)

Mr. and Mrs. George Breslow (13)

Broadway Dental Inc.*

Dr. and Mrs. Jerry Butler*

Mr. and Mrs. Lyle Caddell*

CBRE Charlottesville*

Dr. David Charney (9)

Mr. and Mrs. Ronald Christenson (5)

Clear Lake Community School District*

Coconut Point Ford*

Dr. and Mrs. Bruce Cohen (16)

Mr. John Cohen (4)

Mr. James L. S. Collins (6)

July 1 - December 31, 2016

Ms. Holly Collins (3)
Mr. and Mrs. Scott Connell (12)
Corporate Office Properties Trust (9)
David & Paula Kirsch Family Fund (5)
Mr. and Mrs. William Diaz (2)
Mr. and Mrs. Willard Dickerson (19)
Mr. and Mrs. David Dobke (7)
Mr. James Dooley (2)
Dooley Gasket and Seal, Inc. (3)
Dr. Cynthia Dorroh and Mr. Craig Topham (3)
Mr. and Mrs. Justin Ebert (2)
Electrical Associates (3)
Mr. and Mrs. Joseph Ernst (6)
Mr. and Mrs. Michael Foglia (2)
Ms. Angelina Foglia (2)
Footprints Case Management*
Ms. Nathalie Garner*
Dr. and Mrs. Andy Geer*
Mr. Sigmund Gjesdal*
Gloria & Frank Pipp Family Foundation (14)
Gold Fisch Designs*
Ms. Patricia Gordon and Mr. Alan Koreneff (5)
GraceWorks Unlimited (1)
Mr. and Mrs. David Gray (14)
Mr. Brian Greene*
Haggerty Family Foundation*
Mr. and Mrs. Michael Hall (4)
Mr. Hooper Hardison (8)
Mr. and Mrs. Edward Hardison (6)
Mr. and Mrs. Joel Hasfjord (8)
Mr. Louis Hazel (9)
Henrico Fraternal Order of Police - Lodge #4*
Mr. David Hess*
The Hessler Family (1)
Prof. William Highsmith Jr. (6)
Mr. and Ms. Tom Hodge (5)
Horwitz*
The Hunt Michael Hollis Fund (4)
Illinois Tool Works Foundation (ITW) (11)
Independent Health*
Mr. and Mrs. Darren Jackson*
Ms. Jodi Johnson*
Mr. and Mrs. Glenn Jorden (2)
The Joshua & Luke Welch Charitable Fund*
Dr. and Mrs. Glenn Kalick DVM (16)
Mr. and Mrs. Mark Kaminsky (4)
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Keystone Pharmacy*
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Mr. William McMenamy Jr (8)
Ms. Gail McNutt*
Mechanical, Inc. (7)
Mr. and Mrs. Charles A. Mohan Jr. (21)
The Mouk Family (5)
Mr. and Mrs. Jim Mowrer (10)
Mr. and Mrs. Francis Mroz (7)
Mr. and Mrs. Eric Mullin (5)
Mr. David Neill (5)
Mr. Jason Nemeth*
Northern Virginia Plant People*
Mr. Martin Packouz (13)
Mr. and Mrs. Neal Palmer (16)
Partners In Education - Narragansett Elementary School*
Pediatric Dentistry of Ft. Myers*
PriceWaterhouse Coopers (2)
Puget Sound Kidney Centers (4)
Mr. and Mrs. Gregory Ray (1)
Mr. and Mrs. Andrew Reardon (3)
Drs. Fred and JoAnn Reckling (2)
Mr. Stephen Red*
Mr. and Mrs. Taylor Reid*
Revenue Optimization Solutions LLC (3)
Ms. Megan Ritchey (1)
Roanoke City Public Schools (7)
Roofing Consultants Ltd (12)
Rothlisberger Family Charitable Fund*
Mr. David Russell*
Ruth Camp Campbell Foundation (1)
Mr. and Mrs. Don Ryan (4)
Ryan Companies Us Inc. (5)
Scharff Charitable Foundation (3)
Mr. Nick Schneider*
Dr. and Mrs. Mark Schnitzler PhD (10)
Mr. and Mrs. Brett Schoenecker (14)
Showalter Construction Co (5)
Mr. and Mrs. Jerry Shuck (15)
Mr. Larry Sidwell*
Mr. David Smith (3)
Mr. Matthew Smith*
Mr. Robert Snell (6)
St. Paul Episcopal School (2)
Ms. Patty Stageman (5)
Star Fuel Centers, Inc. (13)
Mr. and Mrs. Peter Stathakis (3)
Mr. Robert Stevick (9)
Mr. and Mrs. Douglas Szopo*
TD Bank (4)
TJ Maxx - District 4101 (4)
Mr. and Mrs. Robert Tranbaugh (10)
United Heritage Charity Foundation*
Mr. and Mrs. Li Kan Wang (3)
Mr. Brent Warner (1)
The Whitehouse Family (2)
Dr. Terry Yochum (15)
Mr. John Zetterower (6)

( ) Number denotes years of support to UMDF *(*) indicates new donor* To become a UMDF donor, please visit us online at *www.umdf.org/donate*

Family Research Funds

The Sterchi family started their Family Research Fund through UMDF in June of 2012. Since that time the family has managed to raise over $83,000 towards important mitochondrial research. They have managed to do that through both personal donations and hosting wildly successful special events. Their son Brady was diagnosed with MELAS in 2009.

Q: Tell us a little bit about your family.
A: We are are family of 4 from southern Indiana. Haylee is a 12 year old 6th grader and Brady is a 9 year old 3rd grader.

Q: When did you find out Brady had a mitochondrial disease?
A: We took Brady to his local pediatrician because he had a fever and was extremely lethargic. By the time we got there, he was seizing and had a temperature of 106.3°. He was rushed to the local ER and had a blood sugar of 10. He was then taken via helicopter to Riley's Children's Hospital in Indianapolis. He was placed in a medically induced coma for 9 days and remained hospitalized for another 11. Once released, we were called within 4 weeks with the diagnosis.

Q: When did you decide to start your Family Research Fund?
A: Essentially, we had a death in the family and $10,000 was left to the UMDF. We were contacted about starting a research fund. Since then, we've been involved in fundraisers every year.

Q: What made you decide the Research Fund was the right option for your family?
A: We wanted to do what we could for all people affected with Mito.

Q: This is the second year you've held the Brady's Bunch 5k and Fun Run. Why did you decide to hold that type of event?
A: Our support system was ready to tackle a new type of fundraiser.

Q: What is your favorite part of the event planning process?
A: The opportunity to bring awareness to the community.

Q: What is the best part of event day?
A: Meeting new people that don't know us but came out to support our family.

Q: What has been the most rewarding part of the entire process?
A: The amount we have been able to donate in Brady's name.

Q: What advice can you give to other families who are thinking about starting a Family Research Fund?
A: It's a good way for a family that has been affected with the disease to make a meaningful difference. It provides an avenue to help with the challenging situation. Whether it is one dollar or a million dollars, every dollar counts.

UMDF education

Bringing the experts to your community

Mitochondrial Medicine 2017

The United Mitochondrial Disease Foundation invites you to be part of our annual Symposium, Mitochondrial Medicine 2017: Washington, DC. Patient and Family meetings and our LHON sessions begin June 30 and end in the afternoon of July 1. Sessions for our scientific and medical community begin June 28th and end on July 1. This year, the symposium will be held at the Hilton Alexandria Mark Center in Alexandria, Virginia.

Whether this is your first symposium or you are a past attendee, the UMDF symposium is designed to enable you to network with many families and individuals who, like you, are seeking more information about mitochondrial disease. The UMDF symposium offers unique access to many of the top mitochondrial specialists through session presentations, Ask the Mito Doc panels, and on a one-to-one basis through the Doctor Is In forum.

Patients and families are given many opportunities to network with other families as well as with some of the top mitochondrial specialists from around the world. The two day patient/family program offers two tracks to meet a variety of needs – affected adults, parents, caregivers, teens, and an opening session targeted for all attendees to prep them for the entire conference.

The Leber Hereditary Optic Neuropathy (LHON) Program will also be available this year on Friday, June 30 for those mitochondrial families interested in attending those sessions as a track choice.

On Thursday, June 30, 2017, we will conduct the 4th Day on the Hill. Patients and families are encourage to sign up and participate for this advocacy event. This is an opportunity for the mitochondrial disease community to gather together and let our voices be heard in the halls of Congress. Once you are signed up, UMDF will provide transportation to and from Capitol Hill, will make appointments for you with your elected officials, and provide you with the background and resources needed to tell your elected officials how mitochondrial disease impacts your life. Currently, more than 100 patients and families are participating. If you would like to take part, visit *www.umdf.org/symposium/day-on-the-hill* and sign up.

An all-time favorite with past symposium attendees, "The Doctor Is In" offers patients and families the opportunity to meet with some of the top specialists in mitochondrial medicine one-on-one. Hours of operation and sign-ups are provided at registration and posted in the networking room.

Another favorite session of our attendees is the Ask the Mito Doc panel. We offer two panels — one dedicated to affected adults and the other for parents/caregivers of affected children. The panels consist of mitochondrial specialists who have experience with adults and/or pediatric mitochondrial patients. Questions are submitted ahead of time and some are taken from the floor depending on time.

As part of a grant from the Edith L. Trees Charitable Trust, the UMDF is pleased to offer FREE Teen Sessions again this year and is extending the age limit through 25 years of age. The Teen/Young Adult Sessions are intended for mitochondrial teens and/or young adults between the ages of 13-25. If an interested individual falls outside of this age range and wants to participate, please contact us at connect@umdf.org and explain your situation. *Please note – a parent/guardian will need to complete the registration form for minors participating in all symposium teen activities. The sessions will be held on Friday and Saturday as noted in the schedule. Lunches and banquet celebration will be provided for a fee.*

To see our complete list of patient and family sessions, our scientific and medical session, or to register, visit *www.umdf.org/symposium*. We would also strongly encourage you to make your hotel reservations soon as rooms book fast.

Scholarships

UMDF will offer scholarships to help defray costs associated with travel, lodging and registration fees. Please visit *www.umdf.org/symposium/scholarships* to see eligibility requirements and to download an application.

Discounted Rates

NEW this year! Through the generosity of **PNC Charitable Trust, Tishcon and Reata Pharmaceuticals**, the UMDF is pleased to offer special discounted rates through May 27, 2017.

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 application deadline is April 15, 2017.

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.

Cancellation Policy

All cancellations will be subject to a service charge based on registration fee amounts - a small percentage must be deducted to cover processing charges to the foundation. A written notification of cancellation must be made to UMDF in order to process any refunds. No refunds will be issued after Friday, June 30, 2017.

Permission to Use Image

By registering for this conference, you have given the UMDF permission to use images (photo and/or video) from this event for educational and promotional purposes for this and future conferences. If you do not want your image to be used, please submit a letter by July 1, 2017, to UMDF, Attn: Cliff Gorski, 8085 Saltsburg Road, Suite 201, Pittsburgh, PA 15239.

Marketing Opt-Out

As a 'thank you' to exhibitors for participating and financially supporting our efforts, the UMDF has agreed to provide them with the names and addresses of our attendees. If you do not want your name and address to be released, please submit a written letter prior to the conference to:

UMDF Development Department
8085 Saltsburg Road
Suite 201
Pittsburgh, PA 15239

FAMILY PROGRAM REGISTRATION FORM
Mitochondrial Medicine 2017: Washington DC
Hilton Alexandria Mark Center - Alexandria, Virginia
June 30 - July 1, 2017

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 FAM1705

Registration Code: FAM1705

REGISTRATION FEES

	Early Bird	After May 27	
☐	$115	$225	Individual Registration
☐	$225	$450	Family Registration *(2 adults/same household)*
☐	$42	$85	LHON Program **Only**
☐	$65	$65	Additional Friday Night Banquet tickets *(per ticket)*

Individual and Family Registration Rates include syllabus, daily continental breakfast, refreshment breaks, two lunches and Friday's banquet. **Daily rates are available online.**

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 ☐ 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.

Online registration closes Wednesday, June 10, 2017

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

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)

PLEASE PRINT CLEARLY

Last Name _____ First Name _____ MI _____

Degree/Suffix _____ Specialty _____

Address _____

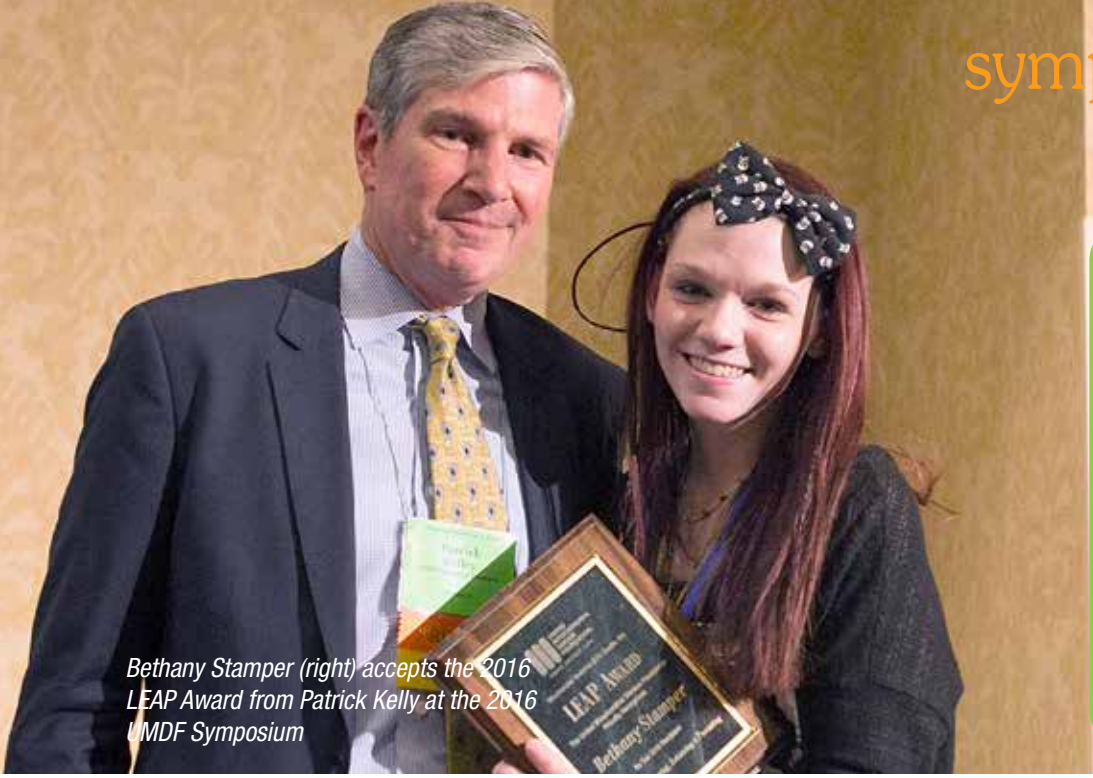
City _____ State/Province _____

Country _____ Zip/Postal _____

Email: _____

Phone: _____ Fax _____

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



Bethany Stamper (right) accepts the 2016 LEAP Award from Patrick Kelly at the 2016 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 30, 2017.

The awards will be presented at the UMDF's annual symposium on June 30, 2017. 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/r/2017Energy

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 fundraising 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/r/2017Heartstrings

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/r/2017LEAP

UMDF national

News from the national office.

Are you looking for someone to connect with? Connect with a UMDF Ambassador, an affected individual/family member who would be happy to network with you. To get started, complete our contact form at *http://secure.umdf.org/RegionalContact* or call us toll-free at 1-888-317-8633.

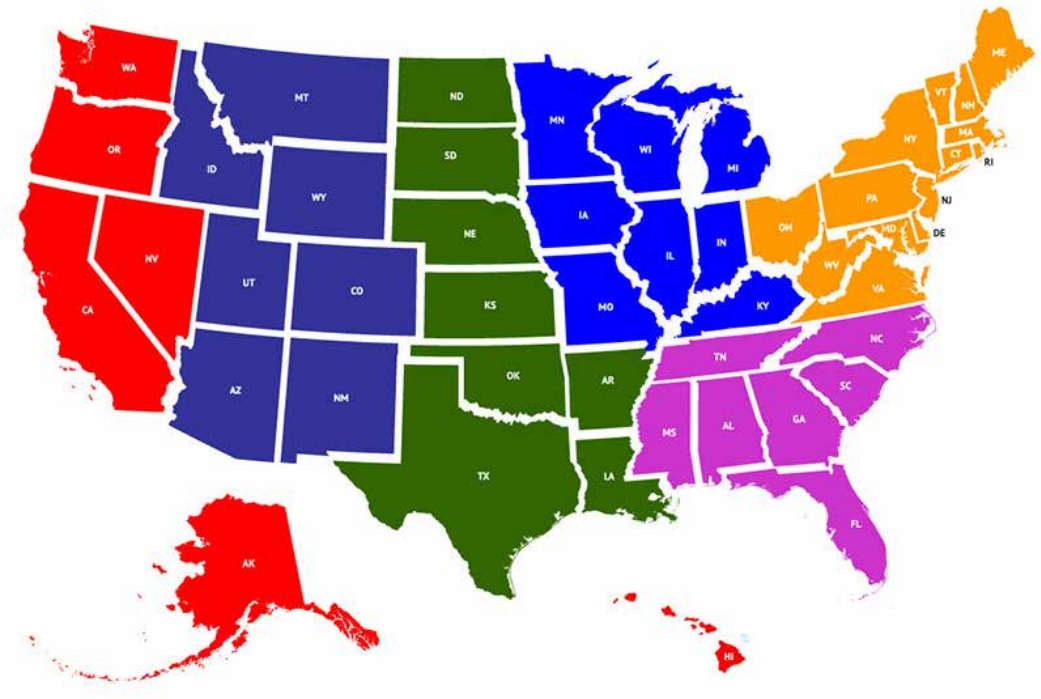
WHAT UMDF REGION DO YOU LIVE IN?

Northeast Region
Nicole McCaslin
Nicole@umdf.org
www.umdf.org/regions/northeast

Southeast Region
Margaret Moore
Margaret.Moore@umdf.org
www.umdf.org/regions/southeast

Great Lakes Region
Anne Simonsen
anne.simonsen@umdf.org
www.umdf.org/regions/greatlakes

Great Lakes Region 5
Jessica Rios
jessica.rios@umdf.org
www.umdf.org/regions/central



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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.

The UMDF focuses on coordination, communication and collaboration.

We bring people and resources together to make an impact on diagnoses, treatments and a cure for mitochondrial disease.

UNITED MITOCHONDRIAL DISEASE FOUNDATION®

Adult Advisory Council Team

On behalf of the UMDF Adult Advisory Council Team (AACT), may this New Year brings you good health and a peace filled life.

We are often asked, "How can I help?" There are many different ways, but the most important and impactful way is to enroll in the UMDF Mitochondrial Disease Community Registry (MDCR.)

First, it's vital to establish a patient registry to show that mitochondrial disease is, in fact, a real disease that effects adults in addition to children. Second, and most powerful, you become a part of its path to a cure.

To read more about the MDCR and if you haven't enrolled yet or haven't finished entering your information, we highly encourage you to do so by visiting *www.umdf.org/registry*.

2017 promises to be another busy year. We have highlighted some upcoming special events so mark your calendar now. We hope to see you along the way!

AACT is honored to represent and serve you. We'll be in touch throughout the year with news and announcements of special interest. We very much welcome your feedback and don't hesitate to contact us anytime at AACT@umdf.org.

Happy New Year!

UPCOMING REGIONAL SYMPOSIA

Regional Mitochondrial Medicine 2017: St. Louis, Missouri
Clinical/Medical Sessions: Friday, March 10, 2017
Patient/Family Sessions: Saturday, March 11, 2017
Washington University School of Medicine
Eric P. Newman Education Center
Seminar B
320 S. Euclid Ave
St. Louis, MO 63110

To register, visit
www.umdf.org/symposium/greatlakes

Regional Mitochondrial Medicine 2017: Birmingham, AL
Join us for an exciting collaborative effort:

- Foundation for Mitochondrial Medicine
- The United Mitochondrial Disease Foundation
- University of Alabama Birmingham

Clinical/Medical Sessions: Friday, April 7, 2017
Patient/Family Sessions: Saturday, April 8, 2017
Friday Location –
Grand Bohemian Hotel
2655 Lane Park Road
Birmingham, AL 35223

Saturday Location –
Birmingham Botanical Gardens
2612 Lane Park Road
Birmingham, AL 35223
To learn more, visit
www.umdf.org/symposium/southeast

Regional Mitochondrial Medicine 2017: San Antonio, Texas

Clinical/Medical Sessions: Friday, November 3, 2017
Patient/Family Sessions: Saturday, November 4, 2017
The University of Texas Health Science Center at San Antonio
7703 Floyd Curl Dr.
Academic Learning and Teaching Center (AL&TC)
3.303
San Antonio, TX 78229

To learn more, visit
www.umdf.org/symposium/central

Mitochondrial Medicine 2017: Washington DC
Hilton Alexandria Mark Center
5000 Seminary Road
Alexandria, VA 22311 USA

Scientific/Clinical Program: June 28 – July 1, 2017
Day on the Hill: Thursday, June 29, 2017
Patient/Family and LHON Program: June 30-July 1, 2017
Register at
http://www.umdf.org/symposium/registration

To view the full UMDF Calendar visit
www.umdf.org/events

Adult Advisory Council Team (AACT)

Jennifer Schwartzott, AACT Chair, New York
Gail Wehling, AACT Co-chair, Illinois
Devin Shuman, YA Coordinator, Washington
Kailey Danks, Toronto
Whit Davis, Pennsylvania
Rev. David Hamm, Maryland
Christy Koury, North Carolina
Joy Krumdiack, Washington
Terry Livingston, Florida
Deb Makowski, Arizona
David McNees, Ohio
Linda Ramsey, New York
Sharon Shaw, Arizona
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 whom will assess and evaluate, provide advice and guidance, and make recommendations to UMDF on all adult related issues and needs.

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: I have had tests for mitochondrial disease and on my genome everything was positive and I thought that was the end of the concern of having a form of mitochondrial disease. A more in depth testing has come back showing evidence of cox deficient fibers and they are now checking for s specific mutation that my uncle was diagnosed with which I believe will take months before I know any more. Having cox deficient fibers showing in a test. Does that mean I have a mitochondrial condition? Or can it show up in perfectly healthy individuals as well? Thank you for your time.

A: Without knowing the specifics of your case (or your age) - I can try and provide some general information.


Sumit Parikh, MD

I am presuming that you mean your "genome testing" came back normal when you say that it was "positive." I would be curious to know what DNA test you had - as there is no one mitochondrial DNA test and even if all available DNA testing in blood is completed - we are only able to confirm a mitochondrial disease diagnosis in patients where we are highly suspicious of one about half of the time.

The muscle biopsy is often sent for a variety of tests - some where we look at the muscle under a microscope - but additional tests of how mitochondria are working (biochemical tests) and how the mitochondrial DNA looks (Genetic tests) are also routinely performed. You mention "COX-deficient fibers." In this situation - the mitochondria look different under the microscope. This finding can be seen in mitochondrial disease, as a part of normal aging and in other genetic and muscle/nerve disorders - so it is a non-specific finding by itself. This piece of information would be used by your physician along with other test findings to help decide the likelihood of your having a primary mitochondrial disorder.

Sumit Parikh, MD

Q: My daughter is 8 years old, and was diagnosed with CUD when she was about 3 months. She has been taking L-Carnitine since she was 4 months old. Lately she has had symptoms of extreme hunger. She even goes so far as to sneak food in her room, eat it and hide the evidence. She has also been having stomach pain, and burps that smell like rotten eggs, and at times nausea, vomiting, and diarrhea. Do you think that any of these symptoms have to do with her CUD?.

A: These are not symptoms of CUD per se. However, the odor and GI and some of the GI symptoms may be due to carnitine. Excess appetite could be a symptom of low blood sugar, but it would unusual with good metabolic control. You should contact your metabolic physician for an appointment.


Jerry Vockley, MD, PhD

Jerry Vockley, MD, PhD

Q: What are the prospects for genetic therapy using CRISPR/Cas9 gene editing technology to treat mitochondrial disease? It seems that mitochondrial diseases that are the result of a single autosomal genetic mutations (e.g., those involving mitochondrial aminoacyl-tRNA synthetases) are well-suited to CRISPR and that CRISPR technology is on the cusp of moving from the realm of academic research to the realm of practical treatment.

A: CRISPR/Cas9 (more generally known as gene editing) technology is indeed a promising prospect for correcting genetic disorders, including those causing mito disease. In fact, the UMDF is already sponsoring a research project to explore gene editing of the mitochondrial chromosome. While exciting, it must be kept in mind that any clinical use is still likely years away.

Jerry Vockley, MD, PhD

Q: I am 48 and have LHON plus. I have been having horrible GI issues as well as many other issues for six months, still trying to get resolved. Fever, chills, weight loss, extreme abdominal and back pain, malnutrition, black stools, nausea, heartburn, and more. My palliative nurse found thrush yesterday. I'm worried about an infection throughout my entire body now but can't get my doctor to respond. Any advice I can print and pass on for urgently testing and treating an infection such as thrush that could be candida in the blood stream? Thank you.

A: A systemic fungal infection is very serious and can certainly be deadly so it is unlikely that this is causative for this patient's issues. She definitely needs to seek assistance from her local doctor.

Fran D. Kendall, MD


Fran D. Kendall, MD

Q: Through genetic testing using a sample of a muscle biopsy taken 4 years prior, I have been told I have a large single 12kb mitochondrial deletion with a heteroplasmy level <15%. My assumption is since my symptoms have progressed significantly since the muscle biopsy, my heteroplasmy level has increased over time. I am an adult whose main symptoms include muscle fatigue/weakness throughout body and muscle fasciculations which have progressed with time, light headedness/mental fatigue as well as focus issues/blurry vision occasionally in one eye primarily. Exercise appears to lower my baseline (creating what seems to be more significant fatigue and weakness which never gets better). My questions are as follows: I was curious if any of the medicine being tested in current pharmaceutical trials (Stealth Biotherapeutics, Reata Pharmaceuticals, etc.) could be beneficial to someone who has such a large single deletion (i.e., with having so much genetic material deleted in a percentage of mitochondria, is it still theoretically/physically possible to still somehow receive benefit from these medications being tested)? ?Are there any treatments recommended for adults with large single mito dna deletions? I am taking supplements (CoQ10 - 300mg morning and 300mg at night, Creatine 5 grams/day, Vitamin E 400 IU/day, Vitamin C 500 mg/day, Vitamin B2 100 mg/day, Vitamin D3 2000 IU/day, Alpha lipoic acid 300 mg morning and 300mg night) which don't seem to help. I have tried light exercise which seems to permanently lower my baseline after engaging, which I notice a few days later after engagement. ? Is muscle fasciculation, typical with those having large single deletion in mitochondria? Any research of large single mitochondrial dna deletions that I should follow? Any doctors in US that specialize in adult patients with large single deletion in mitochondria? Thanks very much. I would greatly appreciate all or any subset of these questions answered. Please let me know if you need any additional information.

A: I am sorry to hear about your medical condition. Currently, as far as I know, the only clinical trial looking at mitochondrial myopathy is the study by REATA. The Stealth study has closed but what we hear, they will be having a Phase III study on their medication opening soon. You can always check clinicaltrials.gov and look for mitochondrial disease studies. Since your heteroplasmy is so low, likely the normal mtDNA present should be working and providing enough energy. There are no studies showing any vitamin therapy to be universally helpful (Cochran Report). Certainly, in any particular patient the vitamins may give some help. Muscle fasciculation is rare in mitochondrial disease and mostly only seen in young children with mtDNA depletion syndromes. Muscle fasciculations are often seen in other types of myopathies, especially ALS. But there are benign forms seen in viral illness. At your age, I would follow the mtDNA induced CPEO literature as single deletion disorders expressed during adult age ranges are usually related to CPEO. I do not know of any physician specializing in large mtDNA deletions."

Russell Saneto, DO, PhD

Russell Saneto, DO, PhD

Q: Recent labs (OAT) support that my 7 year old autistic son has at least some degree of mitochondrial dysfunction (high succinic, methylglutaric, and ketone and fatty acid oxidation). He responded well to Levocarnitine (990 mg twice per day), which also improved GI motility even though it continues to be a problem. He also takes 200 mg of ubiquinol per day and 1 gram of vitamin C twice per day with good results. He continues to struggle with hypotonia related issues: he has extreme difficulty pedaling a bike with training wheels, becomes exhausted after biking a few feet, unable to bike up slight inclines, unable to balance, etc. Per his doctor's recommendation, I recently began supplementing him with 1000 mg of kre-alkalyn twice per day. I have noticed some improvement with his speech with this and it also seems to have a calming effect. However, the improvements last for 2 hours maximum. I believe that he would benefit from a much higher dose eventually. My big concern is that the creatine supplementation has made him physically weaker and exhausted. Any idea of why this could be?

A: I am sorry to hear about your son. There are no universal treatments that are beneficial for all patients with mitochondrial disease. This is unfortunate and many of us are trying our best to uncover best treatments for this disorder. In a recent review article, in a very well respected journal The Cochrane Collaboration, a review of the literature was performed on creatine and its use in muscle dystrophies and metabolic myopathies (which would include mitochondrial myopathies). The authors looked at all studies investigating the effects of creatine on muscle performance. There was no significant effect on metabolic myopathies. So, although in some patients a beneficial effect might be seen, overall there is no significant effect on the majority of patients. So, what you are noticing with your son sounds very much like there is little benefit and maybe even a loss of muscle performance. This would be congruent with what the review study in the Cochrane Collaboration reported. I am sorry.

Russell Saneto, DO, PhD

UMDF

events

The energy providing education, support and research.

Fundraisers Benefitting the UMDF

September 2016 M&T Bank employees in Fayetteville, NY held a fundraiser throughout the month of September in honor of Caroline Payne and raised $2,856.

October 7, 2016 Frank and Larry's held a motorcycle run in Fond Du Lac, WI raising over $400 in memory of Kennedy Burgess.

October 7, 2016 Wyalusing Elementary School in Wyalusing, PA held a jeans day and raised $178.

October 17, 2016 The Ribeiro Family in La Mirada, CA hosted a Pizza Party and raised $307.

October 31, 2016 Stockwell Elementary School in Bossier City, LA held a jeans day and raised $440.

November 3, 2016 Narragansett Elementary School held a walk in memory of Elias Wezowicz and raised $1,258.

November 3, 2016 The Hall family hosted a Craft Brew Fundraiser in honor of Nina Hall and her Activate team, "Unstoppable Nina". They raised over $2,000.

November 5, 2016 The annual Fall into a Cure event was held at Breaux Vineyards in Purcellville, VA. The event was bursting with many fun auction items. They raised over $19,000.

November 6, 2016 The annual Bowling for Mito was held once again in Delaware, raising over $2,000.

November 12, 2016 Mary Richards participated in an "Alternative Craft Fair" in Evergreen, CO. She sold beautiful handmade turtle jewelry and raised $2,285.

November 15, 2016 The Knoxville Disc Golf Association held a tournament fundraiser and raised $1,100.

November 17, 2016 The Cousins for a Cure Event was held in Voorhees, NJ at The Mansion. The event was stunning, from auction items, the live auction, and special guest speaker, CNN's Michael Smerconish. The event raised over $180,000 for the Logan Sloane Arronson Research Fund in honor of Sydney Breslow.

December 3, 2016 The annual Carter's Christmas craft fair and vendor show was held at the Athens High School in Athens, PA. The event boasted the most vendors to date and raised $2,950.

February 1, 2017 St. Bernadette's Catholic School in Monroeville, PA hosted their annual Coins for a Cure fundraiser through the month of January raising over $1900!

Upcoming Events

April 1, 2017 The 6th annual Jackson Culley Mito What?! 5k and Fun Run will be held in Millington, TN.

April 8, 2017 The 2nd Mito Dawgs Family Fun Day will be held in Athens, GA by the University of Georgia club the Mito Dawgs! The event will have games, a bouncy house, and snacks!

April 22, 2017 The Brady's Bunch 5k and Fun Run will once again be held in Vincenne's, IN with proceeds benefitting the Brady Sterchi research fund.

May 7, 2017 The UMDF is proud to be a contributing charity for the 2017 Dick's Sporting Goods Pittsburgh Marathon. You can still join our team! Check out our CrowdRise page at *https://www.crowdrise.com/UMDFPitt2017*

June 16, 2017 The annual Go Pro for Mito golf tournament will be hosted at Maple Ridge Gold Course in Columbus, GA.

June 16, 2017 The annual Thomas' Golf for a Cure will be held in West Bridgewater, MA.

June 17, 2017 The second Annual Luca's Legacy golf outing will be held in Crozet, VA at the Old Trail Golf Course. Proceeds will benefit the Luca Florio research fund.

June 24, 2017 The annual Nicholas J. Torpey Butterfly Classic will be held once again in Saint Clair Shores, MI. Proceeds will benefit the Nicholas J. Torpey research fund.

October 8, 2017 Fundraising is already underway for the 40th anniversary Bank of America Chicago Marathon! UMDF is lucky enough to be a partner charity, check out our CrowdRise page today! *https://www.crowdrise.com/UnitedMitochondrialDiseaseFoundation1*

EFL: Seattle

Upcoming EFL Walkathons

Our Fall 2016 EFL Walk season has come to a close! A special thank you to each and every one of our teams, walkers, volunteers, donors, committee members and sponsors who supported an EFL Walk this year!!!

THANK YOU TO:

Energy for Life Minnesota
www.energyforlifewalk.org/minnesota
Energy for Life Indianapolis
www.energyforlifewalk.org/indianapolis
Energy for Life Western New York
www.energyforlifewalk.org/westernnewyork
Energy for Life Kansas City
www.energyforlifewalk.org/kansascity
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Energy for Life Omaha
www.energyforlifewalk.org/omaha
Energy for Life Southern Wisconsin
www.energyforlifewalk.org/southerwisconsin
Energy for Life Akron
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Energy for Life Seattle
www.energyforlifewalk.org/seattle
Energy for Life Charlotte
www.energyforlifewalk.org/charlotte
Energy for Life Southwest Florida
www.energyforlifewalk.org/southwestflorida

P.S. It's not too late to make a donation or to double your donation with a matching gift! Ask your employer if they match gifts!

We are looking forward to our Spring Walks! Please help us reach those goals by supporting one of these amazing cities!

Saturday, April 8 – San Francisco Bay Area
www.energyforlifewalk.org/sanfrancisco
Saturday, April 8 – Houston
www.energyforlifewalk.org/houston
Saturday, April 22 - Tampa Bay
www.energyforlifewalk.org/tampabay
Saturday, April 29 - Dallas/Fort Worth
www.energyforlifewalk.org/dallasforthworth
Saturday, April 29 – Nashville
www.energyforlifewalk.org/nashville
Sunday, April 30 – Atlanta
www.energyforlifewalk.org/atlanta
Sunday, April 30 – New England
www.energyforlifewalk.org/newengland
Saturday, May 20 – St. Louis
www.energyforlifewalk.org/stlouis
Saturday, May 20 – Cincinnati
www.energyforlifewalk.org/cincinnati
Saturday, June 10 – Pittsburgh
www.energyforlifewalk.org/pittsburgh

Upcoming Symposia

The UMDF is proud to enhance our educational programming through regional symposia and below is a list of the locations for 2017 within the United States. The regional program will offer a full day CME activity on a Friday and a half day of sessions for patients/families on the following Saturday.

UMDF Mitochondrial Medicine
Great Lakes Regional Symposium
Washington University School of Medicine
St. Louis, MO
Course Chairs: Christina A Gurnett, MD PhD and Marwan Shinawi, MD
Friday, March 10 &
Saturday, March 11, 2017

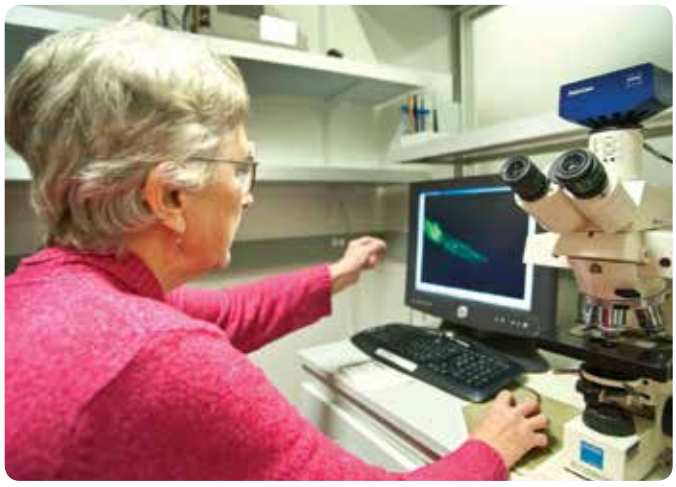
UMDF/FMM Mitochondrial Medicine
Southeast Regional Symposium
University of Alabama
Birmingham, AL
Course Chair: Bruce H. Cohen, MD
Friday, April 7 &
Saturday, April 8, 2017

UMDF Mitochondrial Medicine
Northeast Regional Symposium
Akron Children's Hospital
Akron, OH
Friday, October 13, 2017 &
Saturday, October 14, 2017

UMDF Mitochondrial Medicine
Central Regional Symposium
University of Texas Health Science Center
San Antonio, TX
Course Chair: Sidney W. Atkinson, MD
Friday, November 3 &
Saturday, November 4, 2017

For details on all UMDF Symposia visit www.umdf.org/symposium.

A leader in studying mitochondrial disease, Seattle Children's research is being applied to state-of-the-art patient care, diagnosis and treatments.



Visit www.seattlechildrens.org/mito-research to learn more about how you can help advance mitochondrial research at Seattle Children's.

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Research Institute