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UNITED MITOCHONDRIAL DISEASE FOUNDATION®

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Tyler Sauers and his father, Mike, of Buffalo, WY, take a stroll through the hotel hallways at Mitochondrial Medicine 2017 in Washington DC.

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From the Chairman

by Brent Fields, UMDF Chairman

Inspiring and amazing! Two words that come to mind as I reflect on Mitochondrial Medicine 2017 in Washington D.C. this June. Inspiring because this international meeting afforded me the great opportunity to meet and talk with so many of you. Your struggles, your stories, and your perseverance are nothing short of amazing. Whether you came to the symposia from near or far, I applaud your determination to gather information and learn more about this disease that impacts us all. You are the energy behind the UMDF!

As a parent of an affected child, I know how important it is to stay connected to the latest information on managing care, potential treatments and breakthroughs in therapeutic development. I've attended many international UMDF symposia. Each year, I am astounded by the progress that has been made since the last meeting.

In 2017, UMDF funded over $600,000 in scientific investments. While none of that would be possible without your support, your efforts are moving us faster down the roadmap towards a cure. We all want an easier diagnosis for mitochondrial disease. One of our grants this year funds research that, if successful, could lead to a simple, inexpensive blood test to diagnose mitochondrial disease. That would be nothing short of amazing!

I also want to talk about somebody who I personally find inspiring and amazing. Chuck Mohan started the United Mitochondrial Disease Foundation in the basement of his home in suburban Pittsburgh in 1996. He's served many roles – founder, volunteer, donor, executive director and Chief executive Officer. Now 21 years after serving all of those roles, Chuck has announced that he will transition out of that role in 2018. Chuck is not easily replaced. He and his wife Adrienne worked tirelessly to create a foundation to provide what they did not have while caring for their late daughter, Gina. Their dedication and devotion has provided all of us with a place to find education, support, better treatments and potential therapies. That is beyond amazing!

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Patient Advocacy Groups present Stealth BioTherapeutics Clinical Trials Update

The UMDF, MitoAction and the Foundation for Mitochondrial Medicine (FMM) joined together in August to enable Stealth BioTherapeutics to present the latest findings on its MMPOWER-2 study for patients with mitochondrial myopathy. Stealth is a Boston-based biopharmaceutical company developing therapeutics to treat mitochondrial dysfunction; its lead drug under investigation is elamipretide (previously known as Bendavia).

Primary mitochondrial myopathy (PMM) is a genetically-acquired mitochondrial disease characterized by signs and symptoms of myopathy (debilitating muscle weakness, easy fatigability, exercise intolerance and pain). The Phase 2 MMPOWER-2 trial was conducted to evaluate safety, tolerability and efficacy of treatment using elamipretide in 30 adult patients with PMM. An overall assessment of the top-line MMPOWER-2 results showed benefit across multiple endpoints and is supportive of continuation toward a Phase 3 study in this patient population.

Stealth also outlined a new multi-center study, which is also known as SPIMM-300, researchers will study the relationship between patients' genetic test results and the signs and symptoms of primary mitochondrial disease. As genes are the instructions for how the body functions and how proteins are made, abnormalities or variants in the genes can lead to proteins that do not work correctly and can cause disease. Learning more about how these variants relate to primary mitochondrial disease symptoms may help in developing future treatments. Regional differences in the genetic test methods and in the standard clinical care of patients will also be examined. Additionally, this study may help researchers identify potential patients for future clinical trials of investigational drugs to treat PMD.

Subject to certain exclusion criteria, the study accepts male and female patients, ages 16 to 65, with a clinical presentation of PMD and signs or symptoms suggestive of myopathy.

For more information about the study and its enrollment requirements, vists *http://www.umdf.org/current-clinical-trials* or *www.clinicaltrials.gov*

Australian Scientists Identify Gene as a Cause for Leigh Syndrome

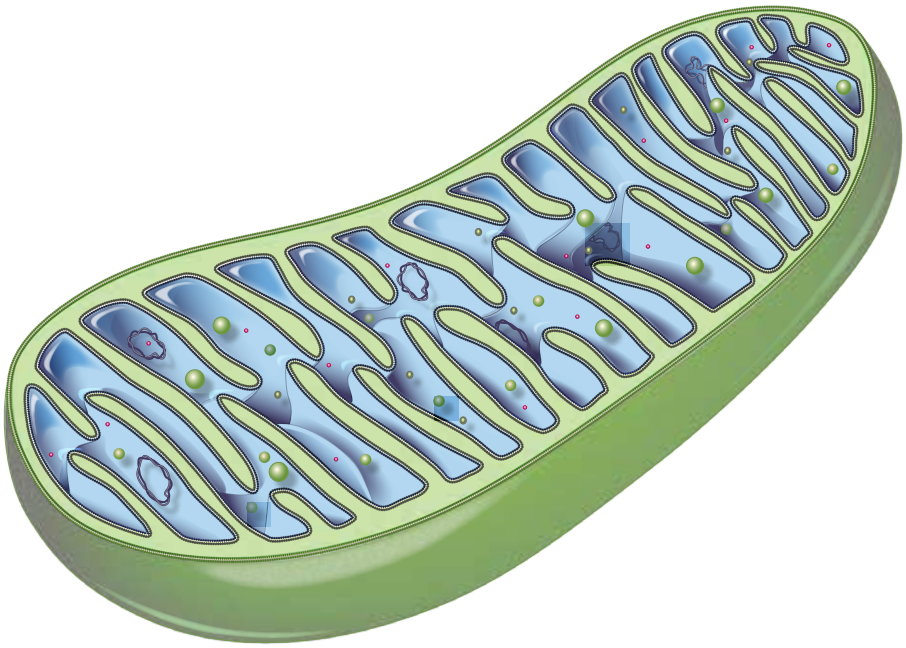
Scientists have developed a new strategy for diagnosing mitochondrial diseases and identified the *Mrps34* gene as a cause for Leigh syndrome. The study, "Biallelic Mutations in MRPS34 Lead to Instability of the Small Mitoribosomal Subunit and Leigh Syndrome," was published in *The American Journal of Human Genetics*.

The team is based at Murdoch Children's Research Institute (MCRI) in Australia. Mutations in a gene called *Mrps34* were found in six patients with Leigh syndrome from different parts of the world, including Australia, France, and the United States. The findings were only possible with researchers applying a different technique called quantitative proteomics. This process involves sampling all the proteins in a cell at once to identify any problems within the cells. Researchers used skin cells from the six patients. They believe that in using the quantitative proteomic approach, they might be able to improve diagnosis.

The research was supported in part by the Australian Mitochondrial Disease Foundation (AMDF).

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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Chuck Mohan Transitioning From Role

After serving the mitochondrial disease community for more than two decades, United Mitochondrial Disease Foundation Chief Executive Officer and Executive Director, Charles A. Mohan, Jr., has announced that he will transition from his role as CEO/Executive Director at the end of 2018. Mohan made the announcement to the UMDF Board of Trustees and to foundation staff. For more than 23 years, beginning in the basement of his home, Mohan began as a volunteer, a member of the Board of Trustees, and has been in his current role as CEO since 2006.

"What Chuck has accomplished at UMDF is nothing short of astounding," said Brent Fields, Chairman of the UMDF's Board of Trustees and CEO of Big Brothers and Big Sisters of Austin, TX. "When he was starting out in the 90's and talking about the disease, people would ask 'mito what'?"

Under Mohan's leadership, Fields says UMDF is funding scientists and researchers who are creating clinical trials and potential treatments for the disease. Patient voices are being heard at the NIH and in Congress.

"This is only possible because Chuck said yes two decades ago to creating, building and nurturing the success of the UMDF," Fields added.

Current Honorary Board Member, John DiCecco, has known Mohan and his family years before the UMDF's founding. "I believe Chuck to be a pioneer," DiCecco said. "Chuck's vision to create an organization that provides help, hope, answers, and research where there was none is such a powerful legacy for the entire community," DiCecco said.

Chuck Mohan said, "We began with one primary focus - to find a cure. We have sharpened that focus to three main objectives: Diagnostic Development, Coordinated Patient Care and Therapeutic Development. We've made much progress only to realize we have a long way to go and I look forward to staying involved in the "Quest Toward a Cure."

Mohan will remain CEO and Executive Director through the search process, scheduled to launch in November 2017. A successor is expected to be announced in May 2018. The Search Committee of UMDF has engaged executive recruiting firm Nonprofit Talent to assist with the search for the next CEO.

Additional Staff Transitions to Serve You Better

UMDF is working hard to ensure we serve our patients and families as best we can all while striving to maximize our program ratio and assure the best use of donor dollars. You may not be aware, but our Regional Coordinators support so much more than just our Energy for Life Walkathons. They support families and individuals interested in establishing Family Research Funds, other special events and connecting families in their region to others that can support them.

We are pleased to share that Margaret Moore will be transitioning from her Southeast Regional Coordinator role to Education and Support Associate. In Margaret's new role, she will continue to build upon and strengthen our UMDF Ambassador program and enhance our support and educational programming. Additionally, she will oversee the "Ask the Mito Doc", "Mito on Call", "Info" and "Connect" email accounts. Margaret will continue to support the Charlotte Energy for Life Walkathon with support from Nicole McCaslin.

Nicole McCaslin will continue to oversee the Northeast Region in addition to overseeing the Southeast Region's special events and supporting Margaret with the Charlotte Energy for Life Walkathon.

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"Day on the Hill" Recap

As part of Mitochondrial Medicine 2017 in Washington, DC, 239 representatives from UMDF, MitoAction, the Foundation for Mitochondrial Medicine (FMM), Miracles for Mito, and the LHON community participated in "Day on the Hill". The advocacy day showed a strong effort in uniting the mitochondrial disease community in speaking with one voice to elected officials.

With more than 300 meetings with members of Congress and the U.S. Senate, participants asked their elected officials to sign onto a letter, authored by Rep. Jim McGovern (D-MA-02), to National Institutes of Health Director Francis S. Collins. The letter asked the NIH to promote mitochondrial disease within the Environmental Influences on Child Health Program, (ECHO). It asked the NIH to competitively fund mitochondrial disease centers of excellence. It asked that the NIH include mitochondrial disease patients in the All of Us Research program to help develop individually, genetically based treatments. Finally, it asked the NIH to utilize new resources under the Cancer Moonshot Initiative to promote the link between mitochondrial function and cancer.

Because of the effort by patients and families during 'Day on the Hill' eight other House Members signed onto the letter. They are:

- Rep. Hank Johnson (D-GA-04)
- Rep.Bill Foster (D-IL-11),
- Rep.Bill Keating (D-MA-09)
- Rep. Brenden Boyle (D-PA-13)
- Rep. Rick Nolan (D-MN-08)
- Rep. Stephanie Murphy (D-FL-07)
- Rep. Niki Tsongas (D-MA-03)
- Rep. Jackie Walorski (R-IN-02)

We will keep let you know when we receive a response from Dr. Collins on these requests.

Also, participants asked their House Members to join the Congressional Mitochondrial Disease Caucus. Because of their efforts, we added eight to the list. New caucus members are:

- Rep. Brenden Boyle (D-PA-13)
- Rep. Jackie Walorski (R-IN -02)
- Rep. Rick Nolan (D-MN-08)
- Rep. Bill Foster (D-IL-11)
- Rep. Thomas MacArthur (R-NJ-03)
- Rep. Ryan Costello (R-PA-06)
- Rep. Robert Pittenger (R-NC-09)
- Rep. Brian Fitzpatrick (R-PA-08)

Make plans to join us for our next Day on the Hill in June of 2019!

MMS Updates Patient Treatment Guidelines

For more than two years, the Mitochondrial Medicine Society (MMS) has been working on updating guidelines for the treatment and management of patients with mitochondrial diseases. This summer, the working group released those updated guidelines in a paper published in Genetics in Medicine, which is the official journal of the American College of Medical Genetics and Genomics. The paper can be seen at *www.umdf.org* and on the MMS website.

More than 30 experts from the United States, Australia, Canada, Great Britain, Italy and The Netherlands worked on this new consensus statement. However, the group urged that the new review does not replace its 2015 statement "Diagnosis and Management of Mitochondrial Disease". That statement was developed by mitochondrial disease experts from around the world and has proven to be most helpful in the treatment and care of patients. The revision includes a summary of the recommended screening guidelines and specialist consultations including cardiology, endocrinology, gastroenterology and hematology to consider.

The review also provides information on medications that must be used with caution or those that should be avoided by mitochondrial disease patients.

The authors list the following in that category:

- Valproic Acid
- Statins
- Metformin
- High-dose acetaminophen
- Selected antibiotics including aminoglycosides, linezolid, tetracycline, azithromycin and erythromycin

The authors hope the recommendations made in this review will raise awareness about the multiple symptoms of mitochondrial disease and help clinicians in the diagnosis and treatment of patients.

The MOTOR Study

A study of omaveloxolone (RTA 408) in mitochondrial myopathies

MOTOR is a double blind, placebo-controlled, multi-center Phase 2 study of the safety and efficacy of omaveloxolone (RTA 408) in mitochondrial myopathies

About the Study

- Treatment: Omaveloxolone or placebo capsules taken by mouth once daily
- Approximately 8 visits to the study site over 16 weeks
- Primary endpoint: Change in peak workload, measured on a recumbent bicycle
- Cost of travel may be reimbursed

Criteria for Participation

- Between ages 18 and 75
- Exercise intolerance with genetically confirmed mitochondrial disease (testing may be provided)
- Willing to discontinue some medications
- Not pregnant, planning a pregnancy, or breastfeeding

Recruiting Study Center Locations

- Los Angeles, California: UCLA
  Perry Shieh, MD
- Dallas, Texas: Institute for Exercise Medicine
  Ronald Haller, MD
- Houston, Texas: University of Texas Houston
  Mary Kay Koenig, MD
- Houston, Texas: Baylor College of Medicine
  Fernando Scaglia, MD
- Akron, Ohio: Akron Children's Hospital
  Bruce Cohen, MD
- Pittsburgh, Pennsylvania: University of Pittsburgh
  Gerard Vockley, MD
- Philadelphia, Pennsylvania: CHOP
  Marni Falk, MD
- Boston, Massachussetts: Mass General
  Amel Karaa, MD
- Copenhagen, Denmark: University of Copenhagen
  Karen Madsen, MD

MOTOR

Contact information for participating study centers can be found on the clinicaltrials.gov listing

REATA PHARMACEUTICALS

Go to www.clinicaltrials.gov/ct2/show/NCT02255422 for more information

Version 1; September 2016

Study looks at Health Care Costs of Mitochondrial Disease

A recent study published in **Molecular Genetics and Metabolism** indicates that treatment of patients with mitochondrial disease is expensive. The study, "Hospitalizations for mitochondrial disease across the lifespan in the U.S.," makes an urgent appeal for better therapies.

The study was conducted by researchers at Children's Hospital of Philadelphia. It compared costs of patients with mitochondrial disease with hospitalization costs across the United States. The study found that most mito patients pay more for treatment than other patients. It also found that mito patients suffer higher than normal rates of other disorders and in hospital mortality rates. A National Institutes of Health funded study also found that actual costs and disease burden could be higher than estimated. In some cases, hospitals discharge patients without registering mitochondrial disease on their record.

"There have been few systematic investigations of the public health burden of mitochondrial disease, even as these disorders are being diagnosed more frequently than ever before," Dr. Shana E. McCormack, study author and CHOP pediatric researcher, said in a press release. "We hope that our initial efforts to define the scope of this problem will lead to better medical practice in helping patients and families."

Study co-author Dr. Marni Falk says she hopes this information will lead to improved funding for research and drug development.

"In addition, understanding the true health burden may help healthcare providers and administrators to refocus efforts to prevent death and severe disability from mitochondrial disorders," added Dr. Falk, who is also the Executive Director of CHOP's Mitochondrial Medicine Center.

Aubrey's Army & Miles for Myles: Linked by Faith, Friendship and Mito

They say that you meet no one by accident, and that couldn't be truer for the Manning and Bell Families. Their story begins before they even realized that it had started. Here is their story of how faith, coincidences and mito brought them together.

Before the Pittsburgh walk in June 2017, Monica Manning posted a message to a mom's board on Facebook for her local town asking about local vendors to get shirts made for their team. Jamie Bell, also a member of the group, saw the post, recognizing 'Energy for Life' walk immediately, as they had signed their team up for the walk as well. The two instantly connected via Facebook Messenger and realized all that they had in common, from growing up in West Virginia, to moving to Columbus, Ohio, just miles apart, to their kids going to the same preschool. During their hours of talking that night, Monica told a story about Myles' last event he was able to attend: his preschool's Christmas program. That's when Jamie realized they had already met!

Back in 2015, at the preschool Christmas Program, Jamie recalls seeing a little boy in a wheelchair, whose feeding tube was leaking. Jamie had noticed and let the nurse who was caring for Myles know. At that moment, the two realized that their children, through Brady and Maxton, who were in the program, and Myles and Aubrey, had brought them together. Jamie and Monica met for the first time face to face at Monica's painting fundraiser for her team. Since then, their husbands and children have met and had an instant connection.

"With this connection, not only was our fundraising to keep the memory of our son Myles alive but to also support others like Aubrey and her family who we have come to know," Monica said.

"I think God knew that we both needed a friend that knows what it's like to see your child suffer and fight the way these kids have and continue to do," Jamie said.

Aubrey's Story

As told by her mom, Jamie:

When Aubrey was 4 months old, we noticed that her growth was slowing and she wasn't smiling or laughing. After what seemed like hundreds of appointments and tests, including a normal brain MRI and normal Whole Exome Sequencing (including the extra portion for the mitochondria), it seemed that mitochondrial disease was completely ruled out. Finally in September 2016, shortly after her 2nd birthday, a new blood test (GDF 15) confirmed that there was a problem with Aubrey's mitochondria and her symptoms fell under the umbrella of Leigh's Disease. Then on December 21, 2016, our world came crashing down around us when Aubrey became very ill with RSV. The ICU doctor sat Chad and I down and told us that our daughter needed intubated and put on a ventilator right away and that there was a very good chance that her heart would stop when they did that. Hearing that and seeing the code team in the doorway with the crash cart, ready to resuscitate if they needed to, was the scariest moment of my life. We spent the next 13 days at Nationwide Children's Hospital, 10 of those in the ICU. Those days were so eye opening for us in so many ways. Not only did we see firsthand the seriousness of mitochondrial disease, but we also saw the lack of knowledge about mitochondrial disease. Here we were in one of the top pediatric hospitals in the country with this new and terrifying diagnosis and WE were the ones educating the medical team on how to properly take care of Aubrey. One physician was actually printing off information from the UMDF website to educate himself! So when I read about the Energy for Life Walk in Pittsburgh, I knew this was something we wanted to be part of. We realized (the hard way) that raising awareness is so incredibly important and crucial for research and treatment that will hopefully lead to a cure one day. Aubrey is the toughest little girl I know and has been through more in her 3 years of life than any person ever should in a lifetime. So until there is a cure, Aubrey's Army will continue to fight!

Myles' Story

As told by his mom, Monica:

Our team is Miles 4 Myles from Galloway, Ohio. Our Family & Friends walk in memory of our sweet son, Myles Edward Manning. Myles was a typical and healthy 7.5 year old boy in the first grade, a basketball player, a Boy Scout, a kid full of laughter & smiles and an awesome big brother. Prior to April 2015, Myles showed no signs of any illness until overnight he became very ill, complaining of his "brain hurting" and blurry vision with nausea. After rushing him to the ER at Nationwide Children's Hospital in Columbus, OH, we learned he was actually experiencing severe status seizures and he was put into an induced coma for nearly two months while the doctors tried to figure out what was wrong. After many tests during his five month hospital stay, he was diagnosed with Alper's Syndrome, a Mitochondrial Disease, and the diagnosis came from a muscle biopsy test followed by the whole genome genetic testing at Baylor University. The physicians and neurologists were in close communications with the Mitochondrial Disease specialist in Akron making sure we made the right decisions caring and treating Myles and were very clear yet sympathetic with our family in this difficult time. Myles was a very strong and brave young boy whose battle was short. He was able to come home in September 2015 but his quality of life had drastically changed. Our family's perspective on life changed as well as we cherished what we didn't know would be the last few month's with our son. After just 9 months from becoming sick, Myles received his angel wings in January 2016 and our hearts ache for him every day. No parent should ever have to experience this. Our family, friends and community has been shocked by how sudden this disease came on Myles with no warning and that there is no cure. We have participated in the Energy for Life Walk in Pittsburgh for two years now raising awareness to our communities through our story about Myles and fundraisers applying our motto to make "MYLES of a Difference".

Hannah's Story

We want to introduce you to 16-year-old **Hannah Jordan** of Oklahoma. Hannah has a mitochondrial disease. Like most affected individuals, Hannah faces medical treatments. Currently, she wears a backpack that houses a pump that provides a constant flow of a metabolic formula to keep her body functioning.

Despite all odds, Hannah is a competitive cyclist and is one of the top ranked juniors in the United States. She was invited to the Olympic Training Center in Colorado Springs. The camp is "for athletes that USA Cycling have identified and would like to develop for international competition." Hannah is an inspiration to all of us and a born fighter.

Her mom, Alicia recently sat down with UMDF and answered some questions for our Spotlight Section:

Tell us about your family.

Our family motto is "Odds Don't Determine Outcome". We have personally been through more than most could imagine. We have been through a house fire, and countless long and innumerable hospitalizations. We have found that if we put God in the center then, we are all stronger than we realized. Hannah's family includes a 17 year old brother, Braxton who was diagnosed with autism, seizures, MR and a multitude of other issues. I was diagnosed with a very aggressive form of MS that included several times of being paralyzed and a pretty significant list of other major health challenges that surrounded that diagnosis. My husband, Chad helped keep us glued together even though our health issues often caused his anxiety to get out of hand.

Tell us about Hannah's diagnosis with mito.

We eventually ended up with almost every organ system being affected with Hannah before mitochondrial disease was suggested. She also had a rare metabolic defect which still remains unidentified and a growth disorder as well. At that point, Hannah was a very sick little girl. She was in a severe catabolic state several times and that turned into an alarm for several specialists that we needed to take a better look at the whole picture. There were times she slept 20 hours a day. She even had low muscle tone which, is shocking now. They made the diagnosis based on our family's abnormal mito marker labs, family history, my abnormal muscle biopsy, abnormal labs and multiple other abnormal tests. They put her G-tube in at six years old after a long hospitalization and stay in the ICU. They found that she did not respond to the drug Glucagon and it was very hard to get her blood sugars to elevate. She left that hospital stay with her G-tube and it has been there ever since.

How does Hannah overcome her mito to ride her bike?

As a family, we work very hard on keeping her well. We demand that she gets proper rest, and that is one of the paramount elements. We turn into "germaphobes" pretty quickly so we aren't all passing things around. Her wellness is very important. She has a mind, body, and spirit approach to her health. She also rides with a very fast men's race team. Being with people that are stronger than you makes you stronger as a person and as an athlete. She doesn't get sedentary for too long or she starts having issues. About a year ago she stopped riding for a few months after having her third set of eye surgeries. She did

not rebound well and lost five pounds of muscle in one month. Nothing changed except her exercise routine. We learned not to do that again. When she first started riding we tried to have her re-tested to see if we could get her off her continuous feeds through her g-tube. After some testing was completed at Boulder Center for Sports Medicine, we discovered she was still burning carbs at a rate of 95% and 5% protein. You should be burning 50% carbohydrates and 50% protein. In ten minutes of riding her blood sugars were in the low 60's. To her frustration, she did not succeed in getting rid of her g-tube. Thankfully that hurdle didn't keep her from her dreams.

How does Hannah keep her mito healthy?

We make as many dietary smart choices for Hannah as possible. In all honesty, she has never eaten well and didn't really eat until she was six years old. If it was a calorie, we cheered. That backfired later because she had a limited diet as she has gotten older and healthier. We have had some major challenges with reflux using blended diets at times.

Recently, we have been utilizing a product we love called Kate Farms. Lately we are running it through her g-tube most of the time. She could tell a difference immediately in how she felt and can drink it by mouth too, if she wants. The great thing is that they have a higher calorie (500 calorie enteral formula) blend that is fantastic when she is having a bad calorie day. She was on nonstop feeds of D 12.5 of maltodextrose at 50-120 ml an hour prior to that.

We are also big fans of Generation UCan and Cocoa Elite. UCan is a super carbohydrate that was designed for a kid with a glycogen storage disease. Elite endurance athletes discovered it's benefits and started using it to fuel and it is now being used with great success by many. We bolus it through Hannah's g-tube before she rides. Hannah is still very volume sensitive and can only handle so much at a time. UCan is a huge benefit for that reason. For her, it is critically important because she can't start vomiting while she is on a ride or she very quickly can go into a severe ketotic/acidotic state.

Cocoa Elite is another very important arsenal in our line-up. Hannah loves how it helps with her muscle recovery. It has flavonoids that have been found to have a positive affect on cardiovascular health. Many elite athletes have been began using it after discovering it aids in their muscular recovery.

Lastly, she is also on a mito cocktail that includes COQ10 and some other supplements.

What is Hannah competing in next in order to achieve her goal of the Olympics?

She will continue racing as she gains physical strength and racing skills. She just upgraded to a Category 3 racer after only learning to ride a bike two years ago. That is a pretty amazing feat for a young rider with no athletic background. Her new race category will frequently include competition against pro and semi-pro women. This will be a new challenge for her.

Hannah became a National Champion just a few days ago by completing the National Hill Climbing Championship. She walked away with a shiny gold medal and new Stars and Stripes jersey by cycling all the way to the very highest summit of Pikes Peak. I was told that she was the youngest female cyclist to ever conquer that.

This very moment, she is residing at the Olympic Training Center. She was identified based on her statistics as a young elite athlete that the Olympic Training Center (OTC) is interested in developing for international competition. This a huge honor for any athlete. OTC did not know her history when she was invited to attend which made her very ecstatic that she was invited on her merit as an athlete. Of course they were shocked when they heard the rest of her story. After she finishes up this Women's Talent ID Camp, we will be home for a few days and then head to Texas for three days of racing at Hotter N' Hell at Wichita Falls, Texas. This will likely prove to be the most challenging field as a new cat 3 racer.

Every single race is dictated by the support Hannah receives by those that have loved and believed in her. We do as much as possible to cut corners everywhere we can. Thankfully for the awesome support of those that have backed her with equipment (Trek, Oakley, Hed, Bontrager, Borah Team Wear, and Phat Tire Bike Shop) she has been able to compete at a high level. As a family, we take one race at a time due to financial reasons. At this point, Hannah needs help obtaining coaching, with racing fees and traveling expenses to get to the next level. We have had to trust God every step of the way and are so grateful for those that have invested in her potential

Thankfully, Hannah is extremely mentally strong because of what she has been through. That same mental strength has helped her immensely to develop strength and gain momentum as an athlete.

Follow Hannah on Facebook at *https://www.facebook.com/hannahjordancyclist*

Ambassador Spotlight

The UMDF is about *coordination, communication and collaboration*; coordinating our efforts and resources, communicating our needs and abilities and forming collaborations to bring information, patients, medical professionals and resources together to enhance progress for treatments and cures.

UMDF Ambassadors are an essential part to those efforts. They are a resource for members seeking to gather information. They coordinate efforts with other members to make a difference in their local areas, and they are key collaborators with the UMDF regional and national staff.

If you would like more information about the UMDF Ambassador program, please contact us at *connect@umdf.org*.

Hi! My name is Dana Ritterbush and I have served as a Support Ambassador in Omaha, Nebraska, since 2013.

On February 3, 2013, my husband and I welcomed our first child, Jonah. Shortly after Jonah's birth, he was taken to the Neonatal Intensive Care Unit due to concerns about a low temperature. We were told that Jonah would likely be away for a few hours. On day two of Jonah's stay in the NICU, we were approached by the neonatologist who broke the news to us that they suspected Jonah had a metabolic disorder – a disorder in which many children do not survive.

Throughout Jonah's 67 days of life, he underwent echocardiograms, abdominal ultrasounds, renal ultrasounds, chest x-rays, MRIs, IVs, a central line and constant blood draws. He had numerous tests, all of which came back normal. Jonah had significant difficulty with oral feedings and he was fed through a nasogastric feeding tube. Despite being fed through the NG tube, he continued to have difficulty putting on weight. Jonah's breast milk was fortified to add additional calories into his diet, but he did not tolerate the fortifier and he had issues metabolizing the calcium. Jonah also had issues with unexplained spitting up and diabetic symptoms requiring frequent injections of insulin. His blood tests showed abnormal levels of amino acids, organic acids and lactic acid. The nurses and the doctors often commented on Jonah's "low tone." Jonah had the doctors stumped. He "wasn't going by the books" for any specific disorders. A Mitochondrial Disease was suspected.

Jonah was never able to come home from the hospital. On April 11, 2013, resting peacefully in our arms and surrounded by family, Jonah lost his battle with Mitochondrial Disease. Jonah's cause of death was Severe Lactic Acidosis and an Inborn Error of Metabolism. A muscle biopsy that was taken after Jonah died, confirmed a Mitochondrial Disease and genetic testing has confirmed Alper's Disease.

Shortly after losing Jonah, I visited the UMDF's website to learn more and to look for any understanding of what happened to my son. I reached out to the UMDF to express interest in becoming a Support Ambassador. We started the Jonah Ritterbush Research Fund and we successfully hosted the 1st Annual EFL in Omaha, in September of 2014. I am thankful for the UMDF and the opportunity to be a part of the mission to find a cure, as well as help others affected by a Mitochondrial Disease in the Omaha area.

Our love for Jonah is our driving force and we will never stop fighting for him. He is greatly missed and shines through in all that we do to help the Mito community.

The UMDF staff says:
"For the past year, I have had the privilege of working with Dana and the rest of the Ritterbush family. They are champion advocates in spreading awareness about Mitochondrial disease, fundraising for research and assisting families who have received a newly confirmed diagnosis. I can see Dana's passion and commitment to finding a cure for mitochondrial disease and it is inspiring. She is a true example of a UMDF Ambassador."
Jessica Rios- Regional Coordinator, Central

Research Fund Spotlight: Noah Shulman

Evan and Kristelle Shulman welcomed their beautiful son, Noah Alan Shulman, into this world on December 29, 2016. Ecstatic to start their new life as a family of three, they took baby Noah home on New Year's Eve. Unexpectedly, their world was about to change and come crashing down.

At just under two weeks of age, they knew something was wrong. Noah was not feeding well, not gaining weight and was overly sleepy. On January 12th, he was admitted to the hospital and an extensive workup began. After nearly a month in the NICU, Noah was finally diagnosed with a rare mitochondrial disease.

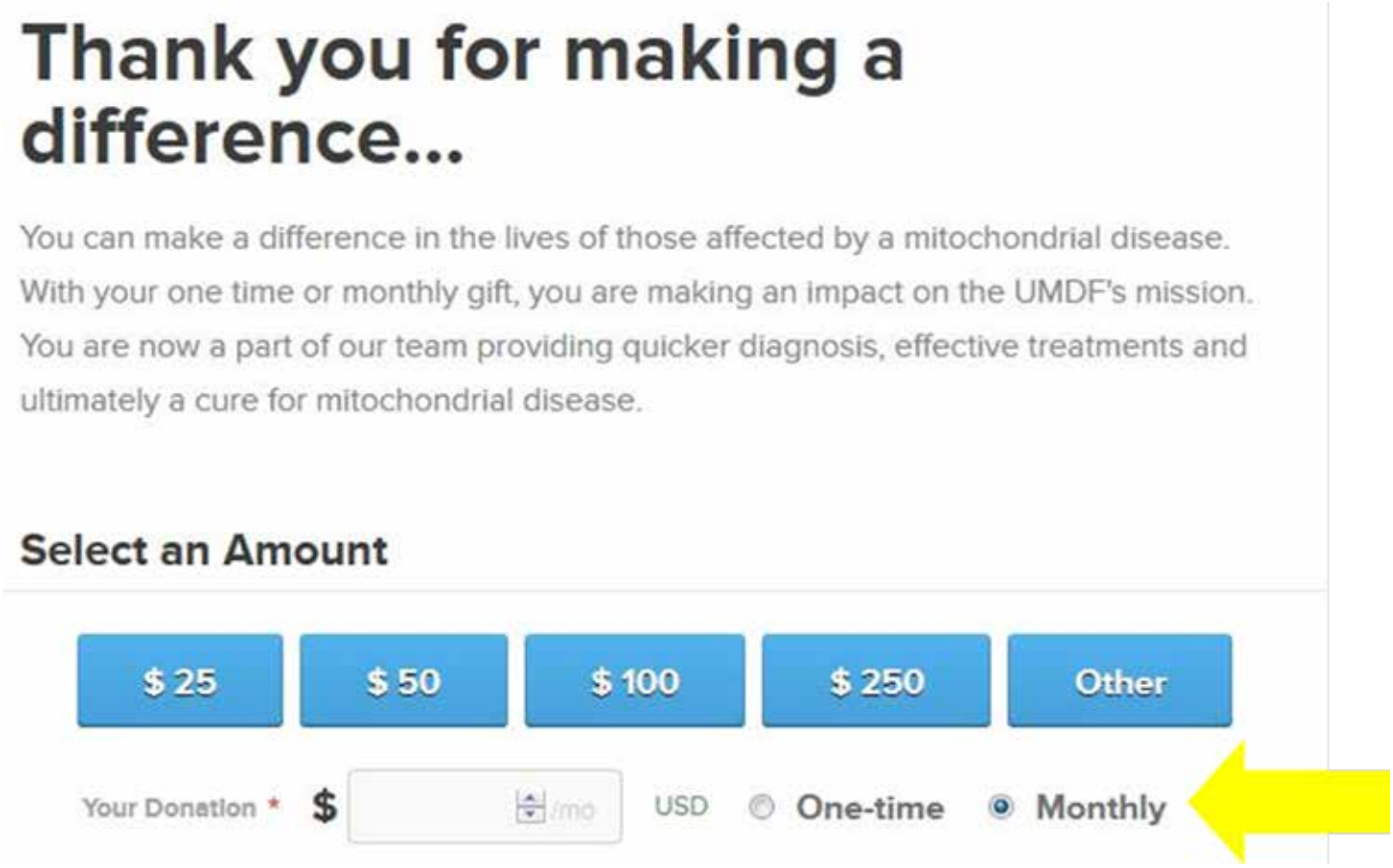
Despite all of Noah's struggles, he never stopped smiling. His eyes would draw you in from across the room, spreading his love infectiously to all who saw him. Nurses, doctors, housekeepers, respiratory therapists – all formed an eternal bond with Noah – each of whom could always be found in Noah's room "partying" the night away – reading him books, playing peek-a-boo, and dancing with him to his favorite songs. Noah loved all.

Sadly, on March 21, 2017, at just 11 weeks old, Noah peacefully passed on to become Evan and Kristelle's little angel. Below, they share why they decided to establish a Research Fund with the UMDF in his memory and the impact he had on all who knew him.

> The fund was established to honor our son, Noah. There is a tremendous need to raise awareness for this disease that so few have heard about, yet so many suffer from. We believe awareness is the key to a cure. Thus far, most of our fundraising has been through social media and word of mouth.
>
> Recently, we added a new modality of fundraising to our campaign, thanks to an amazing nurse. Hyun Sun is one of Noah's nurses who has been with him from the beginning. At Noah's side for his most difficult moments, the bond that she developed with our son is unimaginable. At all hours of the night, she would be at his side and was so much more than a nurse. When she was not caring for his medical needs, she was reading him books, talking to him, playing with him, dressing him in cute outfits, etc.
>
> After Noah's passing, Hyun Sun introduced us to her husband, Joel. We met up with them several times for social events throughout the summer. Each time, we shared stories of Noah and discussed the impact he had on everyone in a short amount of time.
>
> Last month, Hyun Sun and Joel revealed that they were starting their own campaign to help raise money for Noah's research fund through a series of running races and a website, ***www.runformito.com***. Kristelle and I will be joining them in a 10k race this Labor Day. In preparation, we designed t-shirts to represent our cause. We also designed silicone wrist bands to distribute and help raise awareness.
>
> I would be remiss if I did not mention the other amazing nurses whom we have kept in touch with. We have a group chat with several of them and from time to time, each of them will chime in with a reminder of Noah or story to share. The support and love we have received from his entire care team was second to none and each of them has forever become a part of our family.
>
> It is our hope that Noah's legacy will live on through his research fund. If we can help just one baby, one child, one family, then our goal is complete.
>
> "He who saves a single life, saves the world entire..."

Donate to the Noah Shulman Family Research Fund at *www.umdf.org/noahshulman.*

Please contact us if you are interested in partnering with the UMDF in the establishment of a Family Research Fund.



Be an Energy Booster Through Recurring Giving

Convenient, affordable, and impactful—recurring giving gives you the opportunity to make a sizeable difference through small donations over time. Anytime you donate to UMDF online, you have the option to set up monthly donations and become a UMDF Energy Booster. Enter your preferred billing information and have your donation, at whichever level you choose, deducted or charged monthly. You can also sign in anytime and change your monthly giving amount or stop donations. It's easy! Visit *www.umdf.org/donate*. Consider the impact you could make with smaller monthly gifts.

$10 per month – For the cost of a magazine subscription, you could help a newly diagnosed patient find answers, or invest in resources for patients and families like UMDF's online videos, articles, and "Ask the Mito Doc" forum.

$50 per month – For what you might pay for a streaming TV service, you could give a mito warrior a life-changing experience. Help send an affected family or adult to UMDF's international symposium, filled with up-to-the-minute medical information and invaluable doctor/patient networking opportunities.

$100 per month – For the price of a wine-of-the-month club membership, you could advance mitochondrial research and medicine. Support UMDF's Roadmap to a Cure, and change the landscape of therapies and care for patients across the globe.

If you are interested in making a quarterly or annual recurring gift, please contact the UMDF office directly at 1-888-317-UMDF (8633). We will be happy to work with you on your recurring gift preferences.

Join our community of Energy Boosters and sustain our work toward treatments and cures. Your recurring donations, big or small, add up over time and create a huge impact!

Corporate Partnership Spotlight

KENDRA SCOTT

Who are they?
Kendra Scott Designs began its relationship with the UMDF in June of 2015. Since that time, Kendra Scott Designs has hosted 26 *Kendra Gives Back* events for the UMDF. They have helped the UMDF raise over $16,000! These events are fun, social gatherings where our UMDF membership can be celebrated. They are typically 3 hour "parties" where our families get together, have snacks, and look at beautiful jewelry while creating awareness of mitochondrial disease in the community at large. The bonus is that 20% of the sales in that store during the party come back to the UMDF!
If you would like to host an event at a Kendra Scott store in your Community- please contact *events@umdf.org*

About Kendra Scott Designs
Kendra Scott is a loving mom, a driven entrepreneur, and a passionate designer who believes the truest form of success is giving back in a meaningful way.
As a creative mind with a love of natural gemstones, Kendra designed her first collection of jewelry in 2002. The foundation of Kendra's success has been her infectious energy and entrepreneurial spirit, which took her from a $500 project in the spare bedroom of her home to a billion dollar fashion brand loved globally. Known for her unique use of color and quality materials, Kendra has created collections of timeless pieces that have won over loyal fans, media and celebrities alike.

Since she began her company, Kendra has lived by three core values: Family, Fashion and Philanthropy. She created a brand and culture that authentically values giving back and making a positive difference in the community. The Kendra Scott company maintains a focus on its customers and the causes close to their hearts, abiding by the mantra "What Matters to You, Matters to Us."

Kendra Scott Staff says…
The event was so incredibly successful!! I do not say this lightly, but this was truly the most touching, inspirational partnership I have ever had with this company. The families just made this the success it was and I can't thank you enough for bringing them into the Kendra Scott family :)

I'm SO excited to share that we are able to donate $1482.44!! This is the largest donation we've ever been able to provide out of our Boca Raton store and I couldn't be happier that it is for UMDF!

UMDF Members say…
My husband and I were in awe after learning about Kendra Scott's involvement with the UMDF. We greatly appreciate the opportunity and the hospitality that the Kendra Scott Boca Raton team offered. This was our first Mito fundraiser, and we were able to raise funds and awareness. Thank you, Kendra Scott and your team!

Roadmap to a Cure

Perhaps you have seen the big Roadmap poster. Maybe you heard a discussion about it. Maybe you are unfamiliar with it. At Mitochondrial Medicine 2017 in Washington DC, UMDF launched what we call the Roadmap to a Cure.

The 'Roadmap' focuses on three pillars of support, each designed to develop better patient outcomes. The 'pillars' are Diagnosis, Therapeutic Development, and Patient Care. Each involves members of the scientific community, the medical community and industry. The most important part of these pillars is you. The patient community is at the center of each.

I. DIAGNOSIS

Genetic testing presents an excellent opportunity to achieve a specific diagnosis, but less than half of all cases are successfully diagnosed by genetic testing. Other testing methods include muscle biopsies, fibroblasts or buccal swabs. Brain imaging, exercise physiology and various lab measurements of mitochondrial function show some promise. In general, the pathway to diagnosis is not standardized.

UMDF wants to create a better diagnostic scenario for mitochondrial disease patients, there is a clear need to broadly identify and characterize patients based on health information, genetic testing and biosamples.

II. THERAPEUTIC DEVELOPMENT

There are no licensed therapies for mitochondrial disease in the United States. Numerous investigator-initiated trials have been conducted, but in general, there is a notable absence of well-controlled studies within the field. Industry sponsored clinical trials are rapidly increasing in number.

The UMDF's role is to coordinate stakeholders in academia, government and the drug development industry to address important topics such as validated outcome measures, patient-report outcomes and regulatory guidance. These steps are necessary in gaining treatments and cures for mitochondrial disease more efficiently and quickly.

III. PATIENT CARE

Mitochondrial disease patients receive care from a relatively small number of knowledgeable specialists. The many different types of mitochondrial disease and the many symptoms associated with each challenge even the most knowledgeable of doctors. The result is clinical care that is often inconsistent. Additionally, insurance reimbursement for rare disease care is an increasingly challenging situation.

UMDF's goal is to take advantage of a national focus on"personalized medicine" affording an opportunity for the mitochondrial disease community to help develop the programs and tools that will advance optimized patient care in the 21st century. We are committed to collaboration that leads to standards of care and Centers of Excellence models.

You will be hearing more about the "Roadmap." Our aim is to update you as progress is made in each area as we move closer to potential treatments and cures for mitochondrial disease.

Go Pro for Mito

by Wendy Loyd

We held the Third Annual Go Pro for Mito Golf Tournament on June 16, 2017 at Maple Ridge Golf Course in Columbus, GA.

Our first tournament, in 2015 raised $19,500 for the UMDF, followed by $22,000 in 2016. This year our tournament raised $40,000 for the UMDF!!! Our number of golfers has grown each year – 48, 72 and now 107.

My young adult daughter, Chelsea, is affected by mitochondrial disease. Chelsea takes every opportunity to spread awareness about mitochondrial disease at the tournament. People are truly interested in understanding more about the disease. She is very open to answering all questions presented. The comments and feedback have been extremely positive and we truly believe we will continue to grow this event and increase the amount we send to UMDF each year.

The golf committee is primarily a group of my co-workers at Synovus that want to make a difference in the lives of people with mitochondrial disease. We have some others that have expressed interest in joining our committee which will help us continue to expand and grow the tournament. As the parent of a child with mitochondrial disease, this means so much to me. $40,000 from our community is huge.

We are already planning for next year and hope to send the UMDF another large donation. Thanks for all you and the others at UMDF do to support us. If you are a golfer and anywhere near Columbus, Georgia when we are hosting our tournament maybe you can join us!

UMDF

events

The energy providing education,support and research.

Past Fundraisers Benefitting the UMDF

May 7, 2017 GA Atlanta area volunteers Annett Cotte, Debbie Parsons and Katie Parsons hosted a "Kendra Gives Back" event at the Kendra Scott Store at Lenox in Atlanta. A good time was had by all and they raised $518.65 for the UMDF!

May 11, 2017 A Panera Bread Fundraiser was held in St. Louis by Olivia and Liam's Crew to fundraise for the St. Louis EFL.

May 20, 2017 The annual Walk Day Subway Fundraiser was another success for the St. Louis EFL. The event raised $200. Thank you Olivia and Liam's Crew for organizing this all-day fundraiser!

May 20, 2017 A Lula Roe fundraiser was held to benefit Oliva and Liam's crew and the St. Louis EFL. The event raised $264 and we are expecting additional donations!

May 31, 2017 A t-shirt and awareness bracelets fundraiser was held by River Strong to benefit the Indianapolis EFL. The team sold over 80 t-shirts raising $461.47! They have raised $100.50 selling awareness bracelets and plan to continue at the Indy walk this September with a goal to raise $100 additional!

June 2017 The Family of sweet Lacey Jayne Wade sold mito awareness t-shirts and window clings in honor of Lacey and to celebrate her life. Lacey passed away at just 3 ½ months old on September 19, 2014. The Wade Family donated $280 from the fundraiser to Makenzie Lawrey's Million Dollar Mission with the UMDF.

June 4, 2017 Southeast Florida Volunteers Heather Thomas, Gina Hagan and Jessica Lieberman hosted a "Kendra Gives Back" event at the Boca Raton Kendra Scott Store. They raised $1,566.55 for the UMDF! Way to go, Ladies!

June 16, 2017 The 3rd annual Thomas' Golf for a Cure was held in Bridgewater, MA at the Olde Scotland Links Golf Course. It was another great year with the event raising $20,000! Thank you, Jason Schmid (Thomas' uncle) for your amazing support!

June 16, 2017 The 3rd Annual "Go Pro for Mito" Golf Tournament was held at Maple Ridge Golf Course in Columbus, GA. They had a record turnout of 107 golfers and raised $40,000 for the UMDF. A special THANK YOU to Wendy and Chelsea Loyd for making this happen for the UMDF!

June 24, 2017 Gwen Lewis hosted an "Energy For Life Ride" event at CycleBar- Dunwoody in the Atlanta area. 18 attendees raised $475 for the UMDF! Thank you Gwen!

June 24, 2017 The annual Nicholas J. Torpey Butterfly Classic was held in Saint Clair Shores, MI.

June 29, 2017 The Annual Bingo Night Fundraiser was held each Thursday evening in the month of June by Friends of Kristen's Angels of the Chicago EFL. This year the event raised $920. Thank you Friends of Friends for your support each year!

July 2017 12 year old Jared Leit, who has CPEO/KSS, is an amazing artist. For his Bar Mitzvah, he asked family and friends to make donations to the UMDF, and in return they would receive a thank you note created by his own art! Jared was able to raise over $2,600! Way to go, Jared! To read more about his story and see some of his artwork, visit: www.umdf.org/jaredleit.

July 28, 2017 Lee Lyons held a "Shades for Sage" party in honor of her brother, Sage Lyons. There were over 400 people in attendance and they enjoyed an evening filled with music, food, a silent auction and fun! This event raised $16,107 for the UMDF and donations are still coming in!

July 31, 2017 The University of Notre Dame Office of Information Technology Student Solutions chose the UMDF as their benefitting charity for July. Dress-down days, a pizza lunch, cheesecake Friends to benefit

Kids gather around to win prizes at the 3rd Annual Carter's Memorial Celebration

University of Notre Dame Office of Information Technology Student Solutions go green for UMDF

sales and various awareness activities raised $1,000 for the UMDF.

July 31, 2017 An Arbonne Fizz Sticks Fundraiser was held by our Minnesota Corporate Partner Sara & Ryan Wright, Independent Arbonne Consultants. They donated 100% of their commission for each box of Fizz Sticks sold to benefit the Minnesota EFL.

August 2017 Dancing for Donations of Tampa, FL will be donating all of their proceeds for the month of August to UMDF- Thank you, Sara Battaglia!

August 4, 2017 The 10th Annual Run4Raley was held in Philo IL was attended by nearly 200 runners. Over $15,000 was raised for the UMDF! Thank you, Kirby family!

August 4 & 11, 2017 Saving Samantha held two Dine-To-Donate fundraisers at the Fairmont MN Perkins to benefit the Minnesota EFL. Perkins donated 15% of their totals sales 4pm-10pm on the two evenings, raising $560!

August 5, 2017 This year's 3rd annual Carter's Memorial Celebration held in Athens, PA included a softball tournament! Mom and Dad (Brittany and James) and big sister, Alayna, hold this event each year in memory of their son and brother, Carter James Lackey. Their event has raised over $2,800!

August 6, 2017 The Collins Family hosted the Inaugural "Mimosas for Mito" at The Ivy-Buckhead in Atlanta, GA in memory of their son, Graham. This brunch event featured a band, a silent auction, a raffle and mimosas! Over 100 people attended and raised $10,300 for the UMDF!

August 8, 2017 The 7th Annual BP Expressway/DQ Fundraiser was held by our Minnesota Corporate Partner Staples Oil Co. at the BP/DQ location in Jackson MN. The event includes multiple fundraising and awareness events, including 100% of the sales from the Dairy Queen donated, a raffle for Minnesota Twins tickets, a free-will donation and live radio promotions throughout the day. This year's event raised $6,481! Thank you Staples Oil Co.!

August 12, 2017 Jays Monsters of the Minnesota EFL held a Coffee, Community, Music & Bake Sale fundraiser at Community Grounds in Columbia Heights MN. The event raised $327 for the Minnesota EFL.

August 20, 2017 Samantha and Jaxson Lee hosted a Lemonade Stand in Wake Forest, NC in memory of their brother, Aiden. The raised $265. Proceeds will benefit the Aiden Lee Research Fund.

August 23, 2017 The 23rd Annual Fitz Invitational was held at CreeksBend Golf Course in New Prague. The UMDF was selected as the Charity Hole non-profit by the Fitzsimmons family. The event raised $965!

Upcoming Events

September 8, 2017 The 4th annual Birdies for the Blind will be held in Gardner, MA at the Gardner Municipal Golf Course. This event is held in honor of Shane Stewart. Good Luck, Shane!

September 15, 2017 The 4th annual Carlos Alberto Memorial Golf Outing will be held in Miamisburg, OH, at the PipeStone Golf Course. Shotgun start at 1pm, followed by dinner, raffles and awards at 6pm. Join in as a single, or a team of 2 at www.umdf.org/GolfForCarlos. Contact Cristina Rue (cristinarueg@gmail.com) with any questions!

September 16, 2017 The Cutiff Family will be hosting their Annual Mitochondrial Disease Awareness Walk in Anderson, SC. They hold this event in memory of Samuel Cutliff and the proceed will benefit The Samuel Cutliff Research Fund.

September 23, 2017 Elizabeth Lee will host a "Kendra Gives back" event at the Kendra Scott store in North Hills Mall in Raleigh NC, to benefit the Aiden Lee Research Fund.

October 7, 2017 Arizona's Dinner in Dark to benefit the UMDF's LHON Project Fund will be at Maggianos Little Italy in Scottsdale, AZ. The event will feature a three-course meal enjoyed by guests who will be blindfolded at the door. Join us for this amazing evening at Dinner in the Dark.

October 7, 2017 The 2nd Annual Luca's Legacy Golf Tournament will be held in Fredricksburg, VA, at the Cannon Ridge Golf Club. Registration and Lunch begins at noon with a shotgun start at 2pm. You can sign up your team of 4, or as a single player at www.umdf.org/lucagolf2017.

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

October 12, 2017 C.U.R.E. Ride riders will be biking from Santa Barbara to San Diego on October 12 through the 15th. You can join in on the fun or make a donation in support of our riders! The C.U.R.E. Ride benefits the UMDF's LHON Project Fund.

October 15, 2017 A Pumpkin Carving Contest will be held in Pompano Beach, FL, hosted by Jessica Lieberman. Bring your BEST Pumpkin Face ideas and join us for this fun contest!

October 28, 2017 Sydney Breslow will host a "Kendra Gives Back" event at the Kendra Scott store at North Hills Mall in Raleigh, NC, The proceeds will benefit The Logan Sloan Aronson Research Fund in Honor of Sydney Breslow.

November 18, 2017 The 2nd Annual "Mad Dash" will be held at Joyner Park in Wake Forest, NC to benefit the Aiden Lee Research Fund.

May 6, 2018 If you are running in the Pittsburgh Marathon – please be sure to join our Fundraising Team! UMDF is a Contributing Charity for the 2018 Pittsburgh Marathon! For information, please visit www.crowdrise.com/umdfpitt2018.

For a full list of events, visit ***www.umdf.org/events!***

Upcoming Symposia

The UMDF is proud to enhance our educational programming through regional symposia. Below is a list of the locations for 2017 and 2018 within the United States. The regional program will offer a full day of CME activity on a Friday and a half day of sessions for patients/families on the following Saturday.

UMDF Mitochondrial Medicine Northeast Regional Symposium
Akron Children's Hospital
Akron, OH
Course Chair: Bruce H. Cohen, MD
Friday, October 13, 2017 & Saturday, October 14, 2017
www.umdf.org/symposium/northeast

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
www.umdf.org/symposium/central

UMDF Mitochondrial Medicine Pacific Regional Symposium
UC San Diego
Department of Neurosciences
La Jolla, CA
Course Chair: Richard Haas, MD
Friday, February 23 & Saturday, February 24, 2018
www.umdf.org/symposium/pacific

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

Our Fall 2017 EFL Walk season is underway! 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:

San Francisco Bay Area
www.energyforlifewalk.org/sanfrancisco
Houston
www.energyforlifewalk.org/houston
Tampa Bay
www.energyforlifewalk.org/tampabay
Dallas/Fort Worth
www.energyforlifewalk.org/dallasforthworth
Nashville
www.energyforlifewalk.org/nashville
Atlanta
www.energyforlifewalk.org/atlanta
New England
www.energyforlifewalk.org/newengland
St. Louis
www.energyforlifewalk.org/stlouis
Cincinnati
www.energyforlifewalk.org/cincinnati
Pittsburgh
www.energyforlifewalk.org/pittsburgh
Minnesota
www.energyforlifewalk.org/minnesota
Indianapolis
www.energyforlifewalk.org/indianapolis

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 Fall Walks! Please help us reach those goals by supporting one of these amazing cities!

Saturday, September 16 - West. New York
www.energyforlifewalk.org/westernnewyork
Saturday, September 16 - Kansas City
www.energyforlifewalk.org/kansascity
Saturday, September 16 - Detroit
www.energyforlifewalk.org/detroit
Sunday, September 17 - Chicago
www.energyforlifewalk.org/chicago
Saturday, September 23 - Delaware Valley
www.energyforlifewalk.org/delval
Saturday, September 23 - S Wisconsin
www.energyforlifewalk.org/southerwisconsin
Saturday, September 30 - Central Texas
www.energyforlifewalk.org/centraltexas
Sunday, October 1 - Seattle
www.energyforlifewalk.org/seattle
Saturday, October 14 - Charlotte
www.energyforlifewalk.org/charlotte
Saturday, November 18 - Southwest Florida
www.energyforlifewalk.org/southwestflorida

EFL Wrap-Around Events

September 9, 2017 A princess themed "Kendra Gives Back" will be held from 12:00-3:00 pm at the Kendra Scott Store at SouthPark Mall.

September 10, 2017 A Cause an Effect Chipotle fundraiser will be held in Edina MN 4:00-8:00pm to benefit the **Minnesota EFL**. Thank you to team Renewing Hope for organizing this fundraiser.

September 17, 2017 A Kendra Scott Design Party will be held in **Indianapolis** for affected families, individuals and those who have lost a loved one. Limited spots are available to attend the design party. Please contact Great Lakes RC Anne Simonsen for additional details.

September 26, 2017 A Kendra Scott Gives Back Night will be held at the Indianapolis location 5:00-8:00pm. Enjoy sips, sweets and jewels. See the creations from our mito families who participated in the design party. And 20% of sales will be donated to the **Indianapolis EFL**.

October 2, 2017 Boardwalk Billy's Annual Golf Tournament to benefit **EFL Charlotte** and Team Brady Bunch.

October 8, 2017 Bowling for Lila will be held at Park Lanes and will benefit **EFL Charlotte** and Team Life for Lila.

October 12, 2017 A Black Light Yoga event will be held at 7:00pm at Soul Studio in St. Louis Park MN. Proceeds will benefit the Aiden's Red Wagon of the **Minnesota EFL**.

Hosting an event to benefit your EFL Walk Team?? We want to know about it! Email *events@umdf.org* or your Regional Coordinator today!

Symposium Recap

Nearly 350 patients and families joined approximately 270 scientists and clinicians in the Washington, DC area for the UMDF's Mitochondrial Medicine International Symposium. The symposia was held from June 28 – July 1, 2017. Patients from as far as Hawaii to as nearby as Virginia participated in the two day patient and family meeting at the Hilton Alexandria Mark Center Hotel in Alexandria Virginia. In addition to the patient and family platform, members of the LHON community held their annual meeting in conjunction with the symposia.

A number of topics were presented including those on exercise, nutrition, and palliative care. Teen sessions, underwritten by the Edith L. Trees Charitable Trust, provided activities and a prom. The symposium was also a gathering place for adults and young adults who battle mitochondrial disease.

If you were unable to attend this year, UMDF has placed some of the closed captioned presentations on our website and at *http://www.umdf.org/multimedia-library*.

SAVE THE DATE!

Mitochondrial Medicine 2018: Nashville
Sheraton Music City, Nashville, TN
Scientific/Clinical Sessions: June 27-30, 2018
Patient/Family and LHON Sessions: June 29-30, 2018

For more information, visit *www.umdf.org/symposium*

Summary of Presentations

The following are brief summaries of some of the presentations from Mitochondrial Medicine 2017 by Kierra Perry:

AMPK: Restoring Metabolic Balance and Mitochondrial Homeostasis; *Presenter: Reuben J. Shaw, PhD, The Salk Institute for Biological Studies, La Jolla, CA, USA.*
AMPK is an enzyme that plays a key role in cellular energy homeostasis, further recognized as the "metabolic master switch". In studying the enzyme's implications, roughly 2,000 proteins that AMPK phosphorylate were identified in the human genome. When applied to mitochondrial disease, a prolonged AMPK response can be recognized in chronic illness/mitochondrial damage. Activation of AMPK was hypothesized to directly activate PGC1α (for mitochondrial biogenesis), and ULK1 (for mitophagy). Involvement within the AMPK pathway further analyzed novel roles of MFF, the first discovered substrate of AMPK. Variables such as exercise, dietary restrictions, glucose levels, and physiological stress created decreased ATP production, further triggering the AMPK pathway as measured through mouse models. The emerging idea from these studies aims to understand how AMPK acts as a control center of mitochondrial biogenesis. These studies show researchers how AMPK bridges a gap between the complex mechanisms of mitochondrial homeostasis and restoration of natural metabolic balance.

Damage Control: How PINK1 and Parkin Survey Mitochondrial Fidelity and Respond with Selective Autophagy; *Presenter: Richard J. Youle, PhD, National Institutes of Health, Bethesda, MD, USA.*
PINK1, a mitochondrial kinase, along with Parkin, a ligase activated by PINK1, were described to influence the molecular mechanisms of autophagy, mitochondrial quality control and the progression of neurodegenerative disorders. The mechanism responsible for innate cellular destruction of damaged mitochondria is recognized to have a near-identical presentation to manifestations of Parkinson's disease. Findings from relative mouse model strains highlighted PINK1 and Parkin's ability to mediate a quality control pathway essential to various aspects of mitochondrial stability. Upon evaluating strains of Parkin KO mice with controlled L-DOPA administration, a near 100% restoration rate of motor control/movement was identified in just 30 minutes after treatment. In both PINK1 and Parkin KO models, cytokine serum levels spiked post exercise, indicating a substantial inflammatory response; a new conditional phenotype. In the future, it is possible ideas as such can be translated to a clinical perspective to better treat both mitochondrial disorders and Parkinson's disease.

Defining the Molecular Basis of Mitochondrial Disease; *Presenter: Robert W. Taylor, PhD, DSc, FRCPath, Wellcome Centre for Mitochondrial Research, Newcastle University, UK.*
Diagnosis of mitochondrial disease relies heavily on genetic testing and comprehensive biochemical assays. Identifying novel disease genes associated with a range of mitochondrial OXPHOS defects were described best analyzed via next generation sequencing strategies, including both whole exome and whole genome. Next generation sequencing tools are being developed to accurately identify mtDNA depletion syndromes, complex I deficiencies, and generalized disorders of mitochondrial protein synthesis. Furthermore, these studies aim to define the prevalence, natural history, and genotype/phenotype correlations associated with mitochondrial disease and mitochondrial gene expression. Many challenges exist for next generation sequencing. The generalized setbacks in the scientific community were explained in addition to specific challenges relating to identification of mitochondrial disease variants, as they most often present heterogeneous in nature. Understanding mitochondrial disease mechanisms remains the primary goal of this research, with patient care/longevity at the core of providing accurate genetic advice.

Late Breaking: Effects of Elamipretide in Adults with Primary Mitochondrial Myopathy: a Phase 2 Double-Blind, Randomized, Placebo-Controlled Crossover Trial; *Presenter: Amel Karaa, MD, Harvard Medical School & Massachusetts General Hospital, MA, USA.*
Primary mitochondrial myopathy (PMM) is a mitochondrial disease presented with symptoms such as debilitating muscle weakness, exercise intolerance, easy fatigability, and generalized pain. In the phase 2 trials presented, patients receiving the drug elamipretide showed a positive outcome, measured by ability to walk an average of 20 meters more than those receiving placebo during the trial. A new patient-reported outcome tool was developed by Stealth Biotherapeutics specifically for use in this trial in order to accurately assess elamipretide efficacy. This tool, referred to as the Primary Mitochondrial Myopathy Symptom Assessment (PMMSA), found patients reporting that they overall "felt better" after the 4-week trial. PMMSA reports further documented subsequent increases in patient ability to exercise alongside experiencing more mobile lifestyles.

Recruitment is ongoing for observational studies in advance of phase 3 trials. Information regarding enrollment and eligibility can be found at stealthbt.com and/or clinicaltrials.gov.

Role of Mitochondria-Associated ER Membranes in Alzheimer's Disease; *Presenter: Eric A. Schon, PhD, Columbia University Medical Center, NY, USA.*
The endoplasmic reticulum (ER) and the mitochondria exhibit an intricate communication system tethering at specific ER sub-domains, best understood as mitochondria-associated membranes (MAMs). This study described the mechanism in which up-regulated MAM function associated with increased mitochondrial dysfunction leads to Alzheimer's disease (AD). Bioenergetic defects in mouse models with AD preceded any amyloid plaque accumulation. It was thus shown that mitochondrial (respiratory chain defects) seem to be of more influence in progressive AD than what research had previously analyzed. In one described experiment, treatment with desipramine showed potential in rescuing the bioenergetics deficits seen in AD mouse model. A second experiment, treatment with myriocin was described to reverse respiration defects and rescue mitochondrial phenotypes in PS-DKO cells. These experimental conclusions represent a novel marker of physiological activity in mitochondrial dysfunction and AD. However, treatment results do not necessarily suggest that an equivalent therapeutic benefit can be translated in treating mitochondrial diseases.

NAMDC Updates *Presenter: Michio Hirano, MD, Columbia University Medical Center, NY, USA*
NAMDC, The North American Mitochondrial Disease Consortium aims to collect information on mitochondrial disease patients and families with a national effort to grow NAMDC's Clinical Patient Registry. With diagnostic criteria drastically changed in the past 10 years, genetic tools for diagnosis now present as several variables including biochemical data, clinical features, gene sequencing, and more. NAMDC continues to develop diagnostic criteria, natural history studies, pilot projects, fellowship programs, survey studies, and expansion of the biorepository at 16 clinical sites spanning the US & Canada. Looking forward, NAMDC highlighted possibilities such as dietary supplements in clinical trials. Such studies would appropriately examine effectiveness of the non-FDA approved supplemental treatments widely used by mitochondrial disease patients. From the pre-established "mito-cocktail", nicotinamide riboside, CoQ10, and PQQ are among the first three supplements to undergo clinical testing. NAMDC concluded by emphasizing the importance of patient participation towards the group's respective patient registries. Registries can be accessed via www.umdf.org/rdcrn/ as well as www.rarediseasesnetwork.org/cms/NAMDC.

Clinical Aspects (Hilary Vernon, MD), Cardiolipin (Michael Schlame, MD), and Molecular Physiology of Barth Syndrome (William T. Pu, PhD).
It is estimated that fewer than 10 patients are diagnosed with Barth Syndrome (BS) in the US every year. Abnormalities of CL have been linked to many common diseases, however BS remains the only mendelian disorder. This X-linked disorder is caused by pathogenic variants among the tafazzin (TAZ) gene. Among various clinically significant findings from three research labs focused on BS, Michael Schlame's lab found the drug Resveratrol (RSV) able to stabilize cardiolipin (CL) in BS patients. RSV has also been known to improve fatty acid oxidation and respiratory chain defects. Research presented by William Pu further suggested that excess ROS generated in BS could describe calcium abnormalities that relate to cardiomyopathy/arrhythmias frequently observed in patients. Notable research presented by Hilary Vernon included vast biochemical analysis, such as how CL levels serves as a significant indicator of phenotype severity. The combined ideas represent a comprehensive review in which BS may present, further offering various promising methods of treatment and diagnosis. An over-arching idea found from BS research explains how chemical equilibrium ultimately regulates what inside a cell is made. Specific examples include CL levels, phospholipid stability, etc. Presenter Michael Schlame elaborated on this finding, concluding his research efforts reject the theory behind enzyme supplementation therapy. Notable explanation for this theory followed how when fundamental equilibrium within relative cellular processes is not ideal, an unstable structure will be unable to respond appropriately to supplementation.

Mechanisms of Maternal Mitochondrial Inheritance; *Presenter: Ding Xue, PhD, University of Colorado, Boulder, CO, USA.*
Maternal mitochondrial DNA (mtDNA) inheritance remains a conserved mechanism across species. C-elegans were used as a tool to describe how paternal mitochondria are eliminated upon fertilization. Research revealed sperm mitochondria exhibit higher rates of mtDNA mutations, due to increased oxidative damage during fertilization. As a result, paternal mitochondria become depolarized and marked for selective degradation. Delayed removal of mutant paternal mitochondria slows cell division, likely explaining the evolutionary benefit of exclusive maternal mitochondrial inheritance.

Mitochondrial Donation; *Presenter: Doug Turnbull, MBBS, MD, PhD, FRCP, FMedSci, Wellcome Centre for Mitochondrial Research, Newcastle University, UK.*
Mothers with mtDNA mutations who wish to have genetically-related children may now consider this opportunity. The presented research revealed that offspring may present with varying levels of heteroplasmy, not necessarily warranting disease if proper treatment is associated. Newcastle provides counseling, ovum donation, prenatal diagnosis, pre-implantation genetic diagnosis, and mitochondrial donation via spindle transfer and pro-nuclear transfer techniques. In the UK, parents who undergo pronuclear transfer have an extensive 18-month follow up to ensure the child is following appropriate developmental milestones and continues to grow in a healthy manner.

Mitochondria and Autism-A Small Phase I/II Clinical Trial of Antipurinergic Therapy in Autism; *Presenter: Robert K. Naviaux, MD, PhD, University of California, San Diego, CA, USA.*

Initiation of the cell danger response (CDR) occurs as a universal metabolic response to injury in humans. In studying the CDR, it was found that roughly 1000 genes associated with the response correlate with increased sensitivity to sound and light, decreased appetite, and sensory motor deficiencies associated with autism. Understanding CDR and forming subsequent treatment options offers the potential for patients to partially or fully escape the autism spectrum scale. Blind randomization to a single dose of saline (placebo) or suramin (drug) was presented as a study model in autism patients. Developmental milestones such as formation of sentences occurred in every patient who received suramin treatment, while no improvement was seen in all patients who received placebo. Theoretically, if clinical trials prove successful, treatment could be seen effective and applicable for both autism and mitochondrial disease patients experiencing a mito crisis.

Advances in Use of Designer Nucleases to Reduce Mutant mtDNA Levels; *Presenter: Carlos Moraes, PhD, University of Miami, FL, USA.*

Designer nucleases such as ZFN and TALEN have been adapted to cleave mtDNA, highlighting novel implications of treating patients with mitochondrial disease. Mitochondrial-targeted nucleases, or mitoTALENs were shown effective in reducing mutant mtDNA transmission in respective mouse models representing mtDNA diseases. An experiment with mitoTALEN C5027T injection in TA muscle restored tRNA heteroplasmy levels. Current setbacks to this therapy (related to systemic viral delivery to mice) are the presence of DNA repeats preventing TALENs from providing their most effective/desired mechanism of delivery. This research further suggested mitoTALENs pose the ability to reduce mtDNA mutant load in embryos, potentially providing an alternative to mitochondrial replacement therapies. Current strides in designer nucleases were shown to serve as a potential treatment option for disease. However, ideas are currently restricted to mouse models while DNA editing techniques continue their advance to the human population.

Mitochondrial Stress Signaling in Disease, Aging, and Immunity; *Presenter: Gerald S. Shadel, PhD, Yale School of Medicine, New Haven, CT, USA.*

Analysis of the rare disease Ataxia-Telangiectasia was used to link pathways of mitochondrial stress, deafness, and innate immune signaling. Upon developing the first mouse model of a human pathogenic mtDNA point mutation known to cause deafness, research revealed how mtDNA stress triggers an innate, antiviral immune response. Analysis of major mitochondrial transcription factor TFAM showed an ability to selectively repress transcription at a given mtDNA promoter. TFAM knockouts further revealed enhanced viral resistance when studied in vitro and in vivo. Further research will expand on these studies for its ultimate application towards treating common human diseases as they relate to mitochondrial and metabolic pathways.

LEROS- A Long Term, Multicenter, Open-label Clinical Study Using Idebenone in LHON; *Presenter: Xavier Lloria, Santhera Pharmaceuticals.*

The main defect of LHON presents with mtDNA mutations causing reduced energy production and oxidative stress within retinal ganglion cells. There is an approved treatment for LHON that is currently not approved in the US, however research efforts in Europe have shown benefits from treatment as they relate to disease improvement and stability. A 900mg/day dose of Idebenone, a benzoquinone structurally similar to CoQ10 was tested in 250 LHON patients for 12 months. In this research, efficacy measurements were concluded successful if patients experience no disease progression upon trial conclusion. A comprehensive interpretation on drug efficacy following treatment remains controversial due to lack of placebo use throughout the studies. More information about the ongoing phase 4 trial can be found at clinicaltrials.gov, Identifier: NCT02774005.

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Summary of International Clinical Trial Readiness Meetings; *Presenter: Michio Hirano, MD.*

In a brief summary regarding standard of clinical trial assessment measures, the following factors were described as universal models: When analyzing adult patients with mitochondrial myopathies, the most relevant clinical tests can be limited to the 6 minute walk test (6MWT), times up and go test (TUG), sit to stand test, and timed water swallow. In children, the standard is considered the 6MWT, TUG, and the sit to stand test. Clinical trial standards remain just one among many comprehensive efforts currently being made to unite the mitochondrial disease community across the globe. The US population of patients in the NAMDC registry was reported to total 1045. This research concluded the US falls behind in numbers when compared to that of other countries with identical patient registry systems. A combined effort from the UMDF, patients, families, physicians, care centers, researchers, and more serve to continue building the patient registry and setting universal standards of care for the mitochondrial disease community.

Symposium Awards

At its annual symposium, Mitochondrial Medicine 2017: Washington DC, the United Mitochondrial Disease Foundation honored several volunteers for their efforts and dedication in supporting the organization and the patients and families it represents.

The Heartstrings award is presented to an individual under the age of 18 who has invested their time, demonstrated their talents, effectiveness and generosity in raising money or donations to enable the UMDF to continue its mission.

Katherine Chung, 17, from Gaithersburg, MD, wanted to give back to the mitochondrial disease community after her sister, Kristen, passed away in April. Katherine, her late sister, and their mother, were diagnosed with MELAS. Katherine made t-shirts last April to honor her sister. She sold all of them with the proceeds donated to the UMDF.

The UMDF Energy Award recognizes an individual who embodies the spirit of the UMDF mission, which is 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 Energy Award was presented to **Liz Kennerley**. The Solebury, PA native is on a mission to cure mitochondrial disease. She is sure the best way that she can help find a cure is by sharing her story with members of the House and Senate on Capitol Hill. Liz has battled mitochondrial disease her entire life. She is a remarkable young woman who has worked to put together mitochondrial disease educational platforms at various conferences, congressional briefings, and other advocacy meetings.

The LEAP Award recognizes an individual living positively with mitochondrial disease, highlighting the person's accomplishments and volunteer service.

UMDF presented the LEAP Award to 18 year old **Michael Malecha** of Northfield, MN. During a routine physical in 2015, it was discovered that Michael was having some vision problems. Within five days, Michael was diagnosed with LHON. He would lose a major portion of his vision within 6 to 8 weeks.

Michael takes time each month to speak to youth, his peers and adults about his daily challenges. He serves as a role model for other teens and young adults battling mitochondrial disease. In August of 2016, his vision returned to 20/20. Physicians don't know if his vision will stay, but Michael lives his life not taking his eyesight for granted.

The UMDF honored **Carrie Mullin** of Pittsgrove, NJ with the prestigious Stanley A. Davis Leadership Award. The award is presented to a UMDF volunteer leader who exemplifies dedication to the UMDF. The award was created to honor the organization's late Board Chairman, Stanley A. Davis.

Mullin and her husband, Eric, have two children, AJ, age 12 and Patrick, who is 9. Patrick battles mitochondrial disease. Since 2012, her Energy for Life Walk team, Patrick's Parade, has raised over $50,000 in the Delaware Valley. She will chair the walk this fall. She has coordinated numerous fundraisers and has been a UMDF Northeastern Regional Support Ambassador since 2013. Since Patrick was diagnosed in 2012, Carrie has made it her personal mission to raise awareness for mitochondrial disease with the hope of finding a cure.

UMDF
education

Bringing the experts to your community

Research Grant Awards

As part of its Roadmap to a Cure Initiative, The United Mitochondrial Disease Foundation (UMDF) announced it invested a total of $625,000 in 2016-2017 towards the development of diagnostic tools, therapeutic treatments, and patient care. At its annual symposium, Mitochondrial Medicine 2017, $300,000 in research grants were presented as part of the initiative:

Brent Fields, Dr. Prashant Mishra, Alan Breslow, Sherri Breslow, and Sydney Breslow

Brent Fields, Will & Nicole Dalton, Dr. Rustum Karanjia, Jamie, Steve, & Brady Sterchi

Prashant Mishra, MD, PhD, from the University of Texas Southwestern Medical Center, was presented with the Chairman's Award. His project, *"Identification of SLC Family Members as Predictive Biomarkers for Mitochondrial Disease,"* will investigate the potential use of specific solute carrier (SLC) family members in blood serum as novel predictive biomarkers of mitochondrial disease. If successful, this approach could serve as a means to improved diagnoses as well as therapeutic development.

The UMDF's New and Early Principal Investigator Award was presented to **Rustum Karanjia, MD, PhD.** Dr. Karanjia's planned research at the Doheny Eye Center and at the University of Ottawa is entitled "Photopic Negative Response as an Objective Biomarker in Mitochondrial Disease". Many forms of mitochondrial disease have eye-related symptoms. Dr. Karanjia believes his research will validate utilizing Photopic Negative Response as a biomarker for optic nerve function in mitochondrial disease patients. To test this theory Dr. Karanjia will measure eye cell electrical activity in patients over an 18-month time frame.

Brent Fields, Melissa Walker, Michael Hall

Melissa Anne Walker, MD, PhD was presented with the UMDF's Postdoctoral Fellowship Award. Dr. Walker is based at the Massachusetts General Hospital lab of Dr. Vamsi Mootha. Dr. Walker is working on the development of a blood test that will measure mitochondrial activity in a patient. Her preliminary data points to a simple test that can be done as part of a physical exam. This could lead to a simple and inexpensive diagnostic test.

Additional Scientific Investments

In addition to the UMDF Research Grant program, an additional $325,000 was allocated this fiscal year toward other scientific investments designed to bring diagnostic tools, therapeutic development and coordinated care to patients. For example, the UMDF invested $200,000 in further development of the Mitochondrial Disease Community Patient Registry (MDCR). The registration of patients and the capture of their clinical information is critical to the success of the clinical trial process. Without patient participation, clinical trials cannot proceed and treatments and potential cures are stalled. Funding provided by the UMDF to MDCR will enable the continued development of a robust global platform where patients, caregivers and family members around the world can safely and securely share their health and genetic information.

The North American Mitochondrial Disease Consortium (NAMDC) received $50,000 in UMDF funding in 2016-2017. NAMDC was established to create a network of clinicians and clinical investigators in North America who follow sizeable numbers of patients with mitochondrial diseases and are involved or interested in mitochondrial clinical research. NAMDC has created a clinical registry for patients, in the hopes of standardizing diagnostic criteria, collecting important standardized information on patients, and facilitating the participation of patients in research on mitochondrial diseases.

Since its inception, UMDF has provided funding to NAMDC and has successfully advocated on Capitol Hill to keep NAMDC funded through grants from the National Institutes of Health.
With a strategic goal of funding key biomedical research infrastructure UMDF continued to support at a $75,000 level the Mitochondrial Disease Sequence Data Resource Consortium, also known as MSeqDR. MSeqDR was launched at the UMDF Symposium in Washington DC in 2012 as a NAMDC pilot project. It is a global effort to create an international genomic database for the mitochondrial disease community. This database increases the ability of clinicians to diagnose primary mitochondrial disease and provides them with tools to mine valuable information from many mutations in a secure, confidential atmosphere. UMDF has also successfully advocated on Capitol Hill for funding for MSeqDR, leading to follow-on grants from the National Institutes of Health.

UMDF is the largest non-governmental funder of mitochondrial disease research. With the 2017 Research Grants, funding for MDCR, NAMDC, and MSeqDR, the UMDF has provided close to $12 million to-date in scientific funding for our Roadmap to a Cure.

Funding for the UMDF Scientific Portfolio is provided by the generous contributions of our donors, by those who participate in the Energy for Life Walk and other events, and by those who have created family research funds.

UNITED MITOCHONDRIAL DISEASE FOUNDATION®

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To represent and serve the unique needs of the affected adult mitochondrial 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/or needs.

Adult Corner Page

MITOCHONDRIAL MEDICINE 2017 WASHINGTON, DC

If you were unable to join us in Washington D.C., UMDF recorded many of the patient and family sessions.
You can watch and listen those sessions by visiting www.umdf.org/multimedia-library and scroll down to Symposia Presentations.
Visit the AACT website at www.umdf.org/aact
Watch your email for upcoming webinar details!

Dr. Sumit Parikh's Diet Tip

"Eat from the circumference of the grocery store".
- Sumit Parikh, MD, Cleveland Clinic

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, email info@umdf.org or call us toll-free at 1-888-317-8633.

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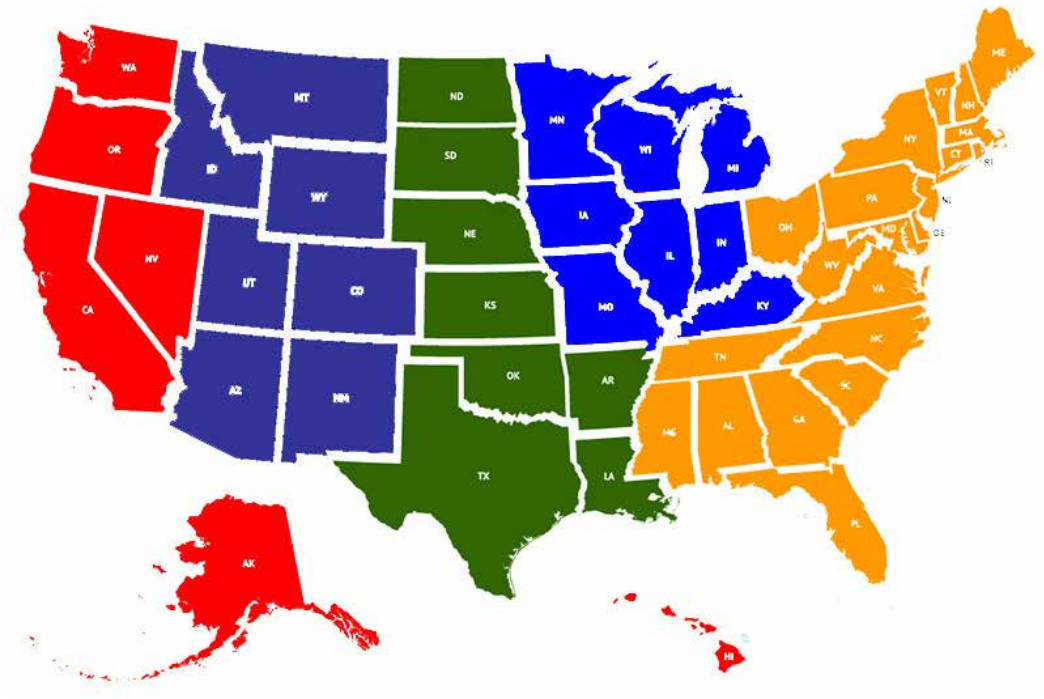

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UMDF Mission

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.

ADVANCING MITOCHONDRIAL MEDICINE

We are a Swiss specialty pharmaceutical company committed to developing medicines to meet the needs of patients living with mitochondrial disorders and other rare diseases.

Our focus is on the development of treatments for neuromuscular and neuro-ophthalmological diseases that currently lack treatment options and our passion is on improving patients' quality of life.

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