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From the Chairman

by Brent Fields, UMDF Chairman

Milestones are important in any organization. They help us to measure significant events on a timeline of progress or call attention to a major development. Our cover story this month on #TogetherWeFightMito reminds me that our community is on approach of another important milestone.

As a parent of a child with mitochondrial disease, I know how frustrating it can be to have to make multiple doctors' appointments for the myriad of issues that we, as parents and caregivers, need to have monitored and treated. Adult patients also have told us that it is incredibly difficult to see one physician for one issue only to have to see a second or third for other medical needs. Most times, those additional medical visits are at other facilities on different dates and at different times. That is why coordinated patient care is a priority and an important pillar in UMDF's Roadmap to a Cure.

The Mitochondrial Medicine Society (MMS) also recognized this missing component and worked very hard to outline standards of care for the mitochondrial disease patient. In the fall, they released their consensus statement on the issue. These important guidelines will serve as our guidepost towards the creation of a Mitochondrial Care Network.

We challenged the staff, the board of trustees, and other mitochondrial disease patient groups to work together towards a strategic initiative that benefits the entire patient community. In fact, at the symposium in June last year, coordinated patient was major discussion topic among the above mentioned stakeholders.

The meeting last summer and the release of the MMS consensus statement led to additional strategy sessions between MMS, MitoAction, Foundation for Mitochondrial Medicine, and the UMDF. That important collaboration formed the idea for the Mitochondrial Care Network. You can read more about the proposed network in the newsletter. Progress on this is moving fast. As you read this, proposals are being submitted and reviewed from clinicians who may be interested being part of the pilot program.

Unifying clinicians towards the implementation of best practices for patients and families and overall better managed care for the community is our collective goal. I am delighted we have started the first leg of this journey – but we do have a ways to travel. It will be a happy moment when this mile marker on the Roadmap to a Cure becomes a milestone for our patient community

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Mitochondrial Organizations Launch Pilot Phase of The Mitochondrial Care Network

The Foundation for Mitochondrial Medicine (FMM), MitoAction, the Mitochondrial Medicine Society (MMS) and the United Mitochondrial Disease Foundation (UMDF) collectively announce an important new initiative to create a Mitochondrial Care Network (Network). The goals for the Mitochondrial Care Network, the first of its kind, are to formally unify clinicians who provide medical care to individuals with mitochondrial disease; to define, design and implement best practices in mitochondrial medicine; and to optimize management and care for patients with mitochondrial disease.

Any clinician in the United States who provides care to patients with mitochondrial disease can apply to join the pilot phase of the Network. Factors for consideration in the Network will include, but are not limited to, current and prior patient volume, multidisciplinary approach and hospital/center support. This exciting initiative offers clinical and scientific challenges, opportunities and rewards. Additionally, participants will play a pivotal role in identifying underserved patients and providing highly needed services for them, as well as contributing to an expanding knowledge base that promises better care for the future. The Request for Application can be found at: *https://goo.gl/qXaiP4*

Deadline for submission is February 28, 2018.

"We are excited about this major collaboration among the patient advocacy groups and the MMS because the Network will help continually improve the standard of care for mitochondrial disease in the United States," said Amy Goldstein, MD, President of the Mitochondrial Medicine Society. "The Network will be an organized group of individual Mitochondrial Medicine Centers (MMCs) that will build on current consensus guidelines for diagnosis and care and significantly improve patient outcomes by sharing knowledge. We strongly encourage all interested parties to apply."

The organizations recognize that without collective knowledge of treatment guidelines, clinicians approach patients in a trial-and-error manner. Working together, the groups plan for a better defined natural history of the disease and better understanding by clinicians, patients and patient families.

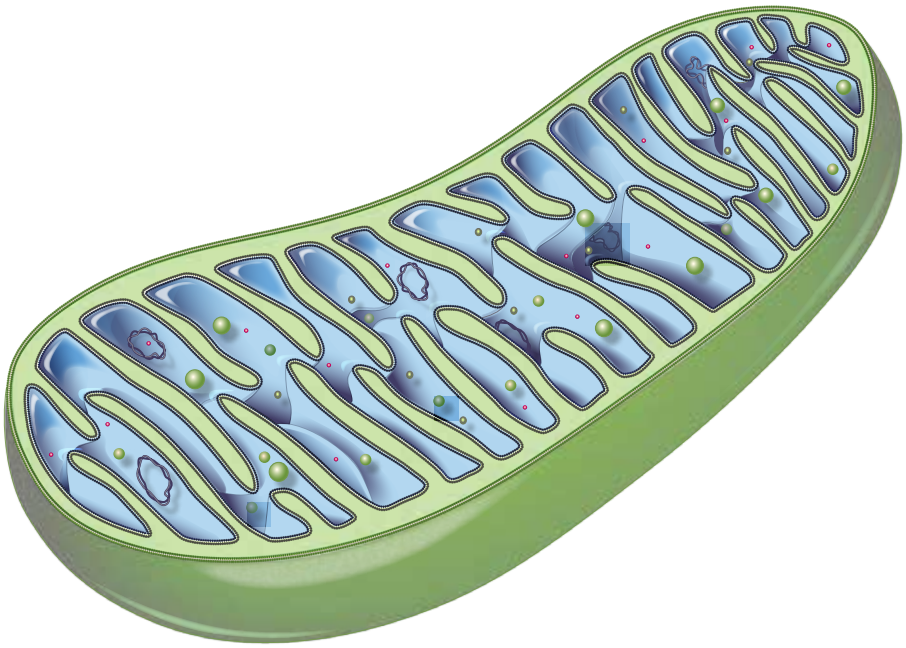
"By combining our efforts and information base, the Network will offer consistency for proper evaluation and diagnosis for primary mitochondrial disease and provide comprehensive medical care to individuals with primary mitochondrial disease," said Kira Mann, CEO of MitoAction. "We are enthusiastic that the Network will address the unmet needs of clinical care for many patients with mitochondrial disease and lead to an expanding knowledge base that will result in better care for the future."

The Mitochondrial Care Network Governance Board will be responsible for final decisions on selecting Centers for the initial pilot phase and eventual expansion of the Network. Members of the Board include Laura Stanley, Executive Director for FMM; Kira Mann, CEO of MitoAction; Amy Goldstein, MD, Amel Karaa, MD, and Sumit Parikh MD of the Mitochondrial Medicine Society; and Phil Yeske, PhD, Science and Alliance Officer of the UMDF.

The Board seeks a diverse group of Centers for the pilot phase to determine the full scope, clinical priorities, implementation of standards of care and long term desired outcomes of the Network. For questions, please contact *info@mitonetwork.org*. More information on the partnering organizations is available at *www.mitochondrialdiseases.org*; *www.mitoaction.org*; *www.umdf.org* and *www.mitosoc.org.*

Stealth BioTherapeutics

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



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People With Parkinson's Have Two Crucial Brain Differences Compared To Healthy People

Scientists have identified two previously unrecognized differences between the brains of patients with Parkinson's disease and those with healthy brains. The new study found that those with the neurological condition have more errors in the DNA of the mitochondria, which leads to increased cell death. Cells that survive have more physical copies of mitochondrial DNA than should be expected in this area of the brain.

According to Dr. Joanna Elson, geneticist at Newcastle University in the UK, previous studies have shown that when patients get Parkinson's, the mitochondria in this area starts to change, and levels begin to drop off.

"Our study is a major step forward in gaining an enhanced insight into the serious condition," Dr. Elson said.

FDA Grants Fast Track For LHON Treatment

Stealth BioTherapeutics (Stealth), has announced that the U.S. Food and Drug Administration (FDA) has granted Fast Track designation for its lead candidate, elamipretide, for the treatment of Leber's hereditary optic neuropathy (LHON).

"We are pleased with the FDA's decision to grant Fast Track designation to elamipretide for the potential treatment of LHON," Stealth BioTherapeutics Chief Executive Officer Reenie McCarthy said. "This designation, a first for our ophthalmology program, is evidence of the serious need for new therapies for those suffering from this devastating rare mitochondrial disease." The designation from the FDA was announced in December, 2017.

The FDA's Fast Track program facilitates the development and review of drugs to treat serious conditions with unmet medical needs. The designation provides the opportunity for more frequent meetings with the FDA over the course of development and allows a drug company to submit individual sections of its New Drug Application (NDA) for review as they are ready, rather than waiting until all sections of the NDA are complete. The designation also increases the likelihood of eligibility for priority review and accelerated approval if relevant criteria are met.

Khondrion presents Phase II KHENERGY trial data supporting Phase III development of KH176 in mitochondrial disease

Khondrion, a clinical-stage pharmaceutical company focusing on small molecule therapeutics for mitochondrial diseases announced in December 2017, results from its KHENERGY study, a Phase II exploratory trial with oral KH176 in the m.3243A>G multisystem mitochondrial MELAS and MIDD syndromes and mixed phenotypes. The results of the trial were presented by Prof. Jan Smeitink, Khondrion's CEO, at the Dutch Life Sciences Conference.

"The final reporting of the KHENERGY study is planned for Q1 2018, but encouraged by the results, we wanted to share these preliminary data regarding safety and efficacy now", said Smeitink.

The KHENERGY study is a Phase II, single-center, double blind, randomized, placebo-controlled 2-way crossover trial involving 20 patients. Patients received KH176 in a 100 mg twice-daily oral dosing schedule for one month. Efficacy endpoints included objective, quantitative assessments as well as questionnaires evaluating the mood and quality of life of patients. The study also explored biomarkers associated with mitochondrial functioning.

The preliminary findings of this study related to adverse events showed a promising safety profile.

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 June 1, 2017 and January 22, 2018.

G. Monty Weathers
Lara Feigitsch
Stacey Mangni
Joshua Atkins
Rose Lingard
Kevin Graessle
Madelaine Martin
Meghan Gannon
Lisa Graves
Grace Caruso
Riley Inglesby

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*

Partner Highlight - Gillette Children's Celebrates 120 Years

Gillette Children's
Specialty Healthcare

This past fall, our partner Gillette Children's Specialty Healthcare in St. Paul, Minnesota, celebrated 120 years of existence, improving lives and helping patients worldwide.

On October 27, 1897, Dr. Arthur Gillette's first patient, Royal Gray, was admitted to the hospital for a severe spine condition. Gray was a 10-year-old boy from Pine City, Minnesota. Dr. Gillette provided excellent medical care to Royal Gray. More importantly, he worked with the Grays to make sure their son had the opportunity to live his best life.

Gillette Children's Specialty Healthcare cares for patients who have some of the most complex, rare and traumatic conditions in pediatric medicine. Children who have disabilities and complex medical needs—and their families—have been at the center of the hospital's mission for more than 120 years. Congratulations to our partner Gillette Children's Specialty Healthcare!

Member Highlight – Lilli Southern

Lilli Southern has been an active part of our mito community and supporting the UMDF since the passing of her brother Jack in 2012. When Lilli started college in 2016 at Butler University in Indianapolis IN, she formed an on-campus service group "Dawgs for Mito". The group has held several awareness and fundraising events, and they have volunteered at the Energy for Life Walkathon in Indianapolis.

On November 12, 2017, Lilli Southern was recognized at Butler University Men's Basketball game with the Chapman Champion award. This recognition is given to a student who shows consistent hard work, outstanding teamwork, dedication and overall excellence in their time at Butler University. Additionally, Lilli Southern has been selected for Butler's Top 100 Outstanding Student Recognition Program for 2018.

We are proud of your accomplishments, Lilli!

Advocacy Highlight – Lillian Dejean

Lillian DeJean, 15 years old from Lafayette, Louisiana, was recently selected to serve as a self-advocate on the Louisiana State Developmental Disabilities Council. Lillian is the first youth to serve as a member of this council.

Lillian's responsibilities as a member of the council include representing the interests of citizens with disabilities at community events, meeting with legislators in session and at their local offices regarding the concerns of constituents with disabilities, and attending quarterly meetings to collaborate with self-advocates, parents of children with disabilities, and disability related agencies.

"I'm excited because this perspective hasn't been explored," Lillian says. "I am in a position to speak for children with disabilities that can't speak for themselves. Parents are an important part of their child's care in advocating for them, but the perspective of the child is unique and vital."

Congratulations Lillian!

Lillian Dejean and her mother, Nicole

Leslie Westbrook/The Advocate

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In 2017, two of our long-term UMDF Ambassadors in Chicago stepped down from key volunteer positions after bringing their energy and leadership to the Chicago Energy for Life Walk and Support Group Meetings for over the past 14 years. We'd like to introduce you to **Gail Wehling** and **Cheryl Lawson** and thank them for their years of dedicated volunteer service to the UMDF.

Ambassador Spotlight

Gail Wehling

Gail Wehling first connected with the UMDF in 2001 as an affected adult. She helped form the Chicago Chapter in 2003 and has been a key resource since that time. Cherie recalls the day she got a call from Gail to say they were going to start a support group in Chicago. "All these years later she is always there to answer a question or guide us. She has been very supportive of the walk and at the ready with information for newbies. She is one special lady." Gail is most proud of the fact that the Chicago Support Group has held consistent support meetings for 13 years. "The best and most extraordinary thing that has happened to me has been meeting all these exceptional individuals and families. They inspire me every day with their strength, courage, determination, resilience, and grace under some of the most difficult circumstances. We have not only become close friends, but are a family - a very special family" says Gail. Gail is looking forward to reading, attending educational classes and art exhibits with her free time. But, Gail plans to continue to be involved in the Chicago area, and she will continue her involvement with the UMDF Adult Advisory Council Team (AACT). "As long as I am able, my mission and lifework will be for the UMDF".

Cheryl Lawson

Cherie Lawson has been involved with the Chicago Chapter since its formation in 2003. Cherie's son Andrew is affect and has been her motivation and inspiration. "The upside to having a chronic illness in my life has been making an extended family." Cherie launched the first UMDF walkathon in Chicago and has served as the event's Chair through 2017. In those years, the Chicago Walkathons chaired by Cherie have raised over $840,000! Cherie says that she is very proud of the walkathon in Chicago and how it has changed and grown. "I love seeing all the teams start gathering on walk day. I always try to make my way around the park to speak with each team and meet the new families." Cherie has been a fixture at local support group events as well. She plans to continue to be involved with the Chicago support group and the walk. She is looking forward to traveling more, perhaps to visit some other walk cities to participate in their events.

We are thankful to these outstanding and long-term volunteers, for their contributions and continued support of the UMDF mission. UMDF Board Member Patrick Kelley was present at the Chicago walkathon this past fall to help thank and recognize Cherie and Gail. "In many ways, the Chicago Chapter is here today because of their tireless work. The UMDF is better and stronger because of their involvement and commitment."

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

Research Fund Spotlight: Emma Grace Boggs

On September 24th, 2015, God blessed The Boggs family with their second beautiful baby girl. She had some complications at birth, but after they brought her home, she appeared to be healthy and was developing normally. They first noticed a problem when they tried to introduce baby foods and she refused to eat anything. They did everything in their power to try to get her to eat and even took her to the STAR Institute in Denver. She was not gaining weight nor growing well, but the doctors always attributed that to her poor intake. She had low muscle tone and started taking steps around 15 months but didn't walk until 19 months. Despite all the family's efforts at trying to get her to eat, she ended up having surgery for a G-tube placement. They thought the G-tube was the answer to their prayers because once she started getting the proper nutrition, she started to get stronger and have more energy. Then came the shocking and devastating diagnosis of Surf1 Deficiency Leigh Syndrome that has knocked the Boggs family to their knees.

Leigh's Disease is a progressive neurometabolic disorder with a general onset in infancy or childhood, often after a viral infection, but can also occur in teens and adults. It is characterized on MRI by visible necrotizing (dead or dying tissue) lesions on the brain, particularly in the midbrain and brainstem. The child often appears normal at birth but typically begins displaying symptoms within a few months to two years of age, although the timing may be much earlier or later. Initial symptoms can include the loss of basic skills such as sucking, head control, walking and talking. These may be accompanied by other problems such as irritability, loss of appetite, vomiting and seizures. There may be periods of sharp decline or temporary restoration of some functions. Eventually, the child may also have heart, kidney, vision, and breathing complications. There is no cure for Leigh's Disease. Treatments generally involve variations of vitamin and supplement therapies, often in a "cocktail" combination, and are only partially effective. The prognosis for Leigh's Disease is poor. Depending on the defect, individuals typically live anywhere from a few years to the mid-teens. Those diagnosed with Leigh-like syndrome or who did not display symptoms until adulthood tend to live longer.

Good news is that Emma continues to make amazing progress. We truly believe she is going to defy the odds and be our little miracle. Although this has been a shocking and devastating diagnosis for our family, it has given us a new perspective on life. Emma has been our greatest teacher. She has taught us to put all our trust in God and to live in the present, cherishing each precious moment with our loved ones. She teaches us every day of the important things in life including how to treat the ones we love by never hesitating to give a hug, steal a kiss and hold a hand. Emma serves as a reminder to live life to the fullest because tomorrow is never a guarantee.

Emma has proven to be a fighter ever since the day she came into this world and she fights with such grace and always with a smile on her face. We have always referred to her as "The Boss". Ever since she was born, all the doctors and nurses in the NICU called her "The Boss" and she has lived up to this name. She bosses everyone around and she even refers to herself as "The Boss" because she can't say Boggs just yet. One morning she awoke and rolled over and said "Emma Boss here"! She is wise beyond her years and her light shines so bright upon everyone she meets. She has a passion for life and loves her family so much. She and her big sister Riley share such a special bond. She is truly an angel and we are so proud to be her parents. Emma and Riley are our whole world and we will go to the ends of the earth for them. We will do everything in our power to give them the life they deserve - one that is filled with love, laughter and happiness.

Through research, the medical world will one day have the resources to help children like Emma win the battle against mitochondrial disease.

We ask for your continued love, support and prayers. Please help us to spread awareness that will lead us to a cure.

Donate to the Emma Grace Boggs Family Research Fund at *www.umdf.org/emmagraceboggs.*

Please contact us if you are interested

New Analysis of Nutritional Interventions for Mitochondrial Disorders

Defects in mitochondria, the tiny structures that power our cells by functioning as biological batteries, cause an array of complex, often life-threatening disorders that can affect any and all organs and systems. In the absence of validated, effective drug treatments, patients with mitochondrial disease often take a variety of vitamins and supplements, substances that are largely unstandardized, unregulated, and unproven.

Experts in mitochondrial medicine propose to remedy that situation, calling for systematic scientific studies in cells and animals to lay the foundation for clinical trials of precise nutritional interventions for patients with energy deficiency diseases.

"We're aiming to raise the bar for clinical treatments," said Marni J. Falk, MD, executive director of the Mitochondrial Medicine Frontier Program at Children's Hospital of Philadelphia (CHOP). Falk co-authored a new analysis of nutritional interventions for mitochondrial disorders published Nov. 3 in the Annual Review of Pathology: Mechanisms of Disease. "Our major objectives were to review the basic scientific evidence for compounds already being used in mitochondrial disease patients and to advocate a framework for rigorously evaluating their safety and efficacy in this population."

The review article represents the collaborative effort of expert co-authors from eight centers, including first author Adam J. Kuszak, PhD, of the Office of Dietary Supplements of the National Institute of Health (NIH). The current effort grew out of a 2014 NIH meeting focused on developing an evidence base for nutritional interventions in primary mitochondrial disorders.

"Our analysis made it clear how much more we need to learn about developing effective nutritional treatments for mitochondrial disease," said co-author Zarazuela Zolkipli-Cunningham, MBChBD, a neuromuscular specialist and attending physician in CHOP's Mitochondrial Medicine Frontier Program. "There's a large gap between the compounds that patients are routinely using and the degree to which those compounds have been scientifically tested."

For instance, Zolkipli-Cunningham pointed to an "astounding variety" of the supplement coenzyme Q10 (CoQ10), sold over the counter in diverse versions and dosages. It is marketed as an antioxidant to reduce biological damage from reactive oxidant molecules.

However, she pointed out, there is no definitive evidence for health benefits from CoQ10. Moreover, there are no standardized formulations for this supplement, so patients may receive widely varying ingredients from one product to another. A third consideration is that a given supplement may act differently in a healthy consumer than in an individual with a mitochondrial disorder, because defects in mitochondria have wide-ranging effects on cellular function. Finally, supplements may act very differently across different subtypes of mitochondrial disease.

"Anything that affects cellular function is biologically acting as a drug, whether you obtain it from a pharmacy or a health food store," said Falk. "However, unlike prescription medications, which are closely regulated and standardized by the U.S. Food and Drug Administration, vitamins, dietary supplements, and medical foods are considered in our country to be in a separate regulatory category with much less stringent requirements. Their manufacturing standards are not as tightly regulated, and their claims are limited to optimizing general public health, not to treating specific diseases. So we know a lot less about their safety and efficacy in patients."

In the current study, the authors review the main types of nutritional therapies used in patients with mitochondrial disease. These include micronutrients, such as vitamins and vitamin-related substances like thiamine (B1), riboflavin (B2), nicotinic acid (B3, also known as niacin), and folic acid. Other nutritional therapies include metabolic-modifying agents such as L-arginine, creatine and CoQ10, cellular signaling-pathway modulators such as resveratrol, and macronutrient modifications such as the ketogenic diet.

Moving Toward More Precise Nutritional Therapies

The authors recommend that all nutritional interventions undergo rigorous testing, and detail the range of laboratory models available for such tests, including specific cell culture systems and experimental animals: C. elegans microscopic worms, fruit flies, zebrafish and mouse lines genetically engineered to model different forms of human mitochondrial diseases. Preclinical studies in these cell culture systems and laboratory model animals, the authors say, should be used to advance precise treatments for different types of mitochondrial disease, setting the stage for clinical trials in both children and adults.

The Mitochondrial Medicine Frontier Program at CHOP already tests potential therapies in cells, worms and zebrafish, with the goal to discover precision therapies targeted to the distinct genetic disease affecting each patient. Partnering with external sponsors, the program will shortly have four active phase 2 or phase 3 clinical trials under way in mitochondrial disease patients. The program has also now hired a dedicated dietitian, possibly the first dietitian in the U.S. focused on harnessing all aspects of nutrition to improve health in patients with mitochondrial diseases.

"As we move toward translating research leads into therapies to test in individualized, patient-centered clinical trials, we need to focus on improving health outcomes that are meaningful to patients," said Falk. Patients each typically have more than a dozen symptoms, most commonly muscle weakness, chronic fatigue, exercise intolerance and balance problems--hallmarks of the energy shortages stemming from malfunctioning mitochondria.

"Our toolbox is so much better than what was available 20 years ago," said Falk, who draws on next-generation sequencing technologies that have enabled researchers to identify approximately 300 different genes in which inherited mutations cause mitochondrial disease. She then integrates the broad advances that have been made in understanding the biology of how mitochondrial dysfunction from different genetic causes leads to disease, together with cutting-edge laboratory techniques and a growing cadre of model systems that can now be readily exploited to prioritize precision-medicine strategies to test in clinical trials. "But of course, we still have a lot of work ahead of us to develop proven, effective nutritional and drug therapies for the diverse array of mitochondrial disease."

Update on the Roadmap to a Cure

by Phil Yeske, PhD, UMDF Science & Alliance Officer

The year 2018 is underway and it's "full steam ahead!" with the UMDF's Roadmap to a Cure initiative. Here is a brief update on key program priorities aligned with each of the Roadmap's three pillars:

PILLAR: IMPROVED DIAGNOSES

Program: Mitochondrial Disease Sequence Data Resource (MSeqDR)

What it is and why it is important: MSeqDR is a genetic data repository that allows researchers to explore the multitude of genetic variants that make up mitochondrial disease. Because mitochondrial disease is a collection of rare disorders, it is critical that a central data repository be established to facilitate faster and better diagnoses.

Key Priorities: To-date few patients have had the opportunity to request the data resulting from genetic testing, and then put said data to work with researchers in a way which represented their own wishes. Soon UMDF will launch a new process by which participants in the Mitochondrial Disease Community Registry can do all those things, unleashing a flood of new data to researchers interested in exploring mitochondrial disease.

Status: The MSeqDR project will be led by Dr. Marni Falk at the Children's Hospital of Philadelphia, which is in the final stages of reviewing and approving the project. We expect to make this important new program available to the mitochondrial disease community sometime in the first half of 2018.

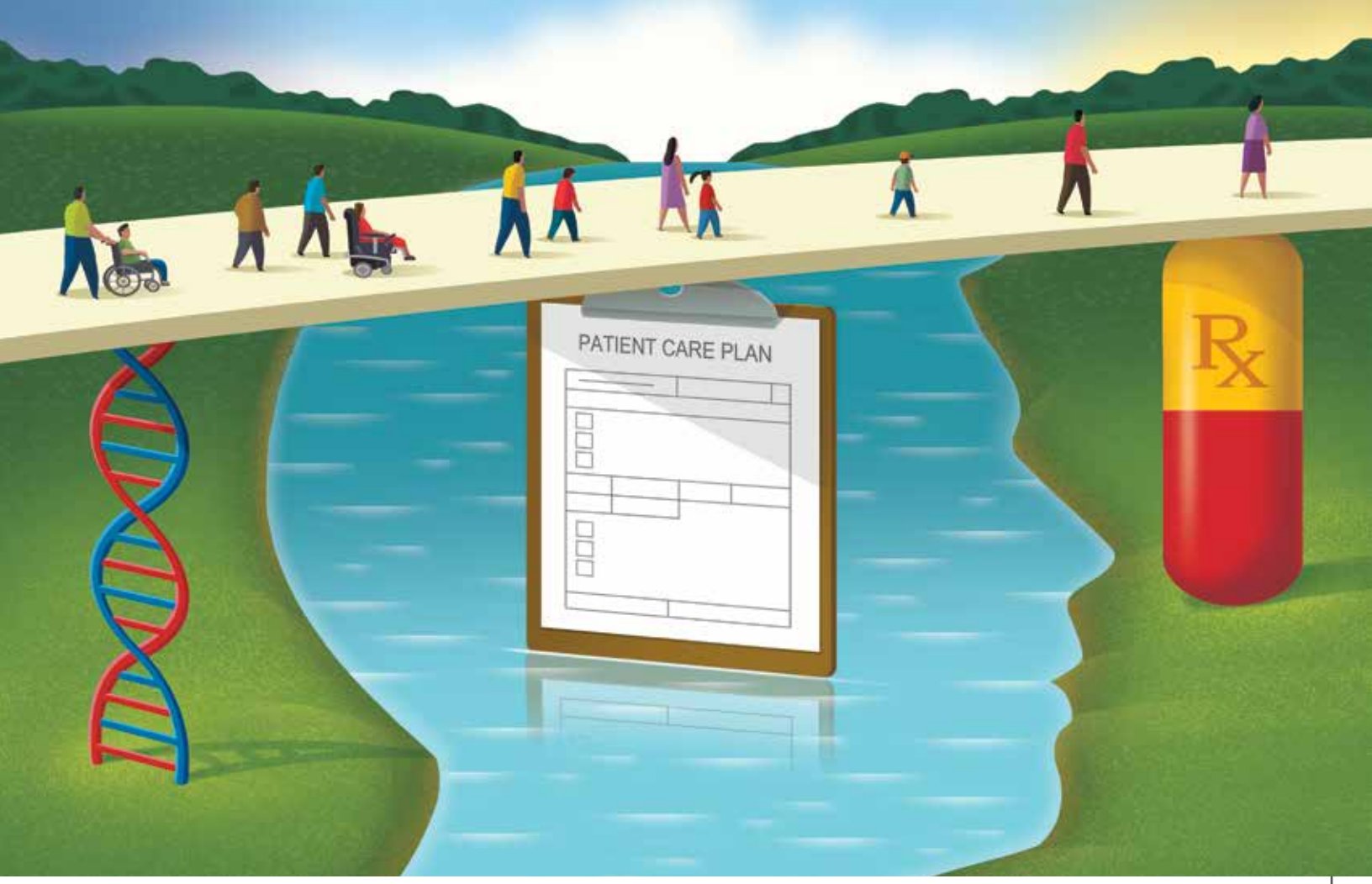
PILLAR: THERAPEUTIC DEVELOPMENT

Program: Mitochondrial Disease Community Registry (MDCR)

What it is and why it is important: We often refer to MDCR as the backbone of the Roadmap because it provides a direct line between the mitochondrial disease and research communities from diagnosis to clinical care. MDCR allows the broad capture of opinions and data that advance biomedical research based fully on the unique wishes of each individual participant.

Key Priorities: To date, we have used MDCR to encourage participation and to share basic information (currently over 2,100 registrants!), but ultimately we are preparing for much broader utility. As more and more approaches for targeting mitochondrial disease with novel therapeutics MDCR will be an important mechanism for aligning patients with researchers and for eventually recruiting participants in clinical trials. The rare nature of mitochondrial disease means we must turn over every stone in every corner of the world to identify and characterize our community, and so a global network of registries is under development.

Status: UMDF is partnering with like-minded patient groups around the world to develop the global registry network on the



PEER platform on which MDCR is based. Our goal is to create a harmonized system of data collection that will facilitate scientific queries. It takes time and money to build such an infrastructure, but the payoff will come as mitochondrial disease research projects and subsequent clinical trials rapidly grow in number in 2018 and succeeding years.

PILLAR: OPTIMIZED CLINICAL CARE

Program: Mitochondrial Care Network (MCN)

What it is and why it is important: Treating mitochondrial disease is a challenge for both patient and clinician due to the complexity of the disease. Recent advances in establishing diagnosis and care guidelines are improving the situation, but a large opportunity still exists for clinical care centers to more formally share best practices and continually improve the care our patients receive. UMDF is partnering with other patient groups and the Mitochondrial Medicine Society to launch a Mitochondrial Care Network in the United States with a shared goal of achieving steady continuous improvement in the level of mitochondrial disease clinical care.

Key Priorities: US-based clinicians actively seeing mitochondrial disease patients have been solicited to apply for the pilot phase of the project in which a small number of centers will work together over the next 12-18 months to further develop the long-term goals of MCN. The project is meant to be broadly inclusive and not limited to only the largest mitochondrial care centers. **Status:** Applications are being accepted in the first calendar quarter of 2018, and pilot phase sites will be announced sometime in the second calendar quarter of 2018. An update will be provided at the UMDF's Mitochondrial Medicine Meeting in Nashville this June.

Beyond these programs we are also actively structuring a targeted set of investments this year in Leigh Syndrome research that will span all three pillars of the Roadmap. Please keep in mind that each of the programs is a large, complex undertaking that requires effective partnering as well as significant capital investment. Your donations are what make it possible. Thank you for your support and we look forward to sharing further exciting updates as 2018 progresses!

We continue to invite families to send us personal stories about how they live with their mitochondrial disease. This story is from a very special friend that I first met at the St. Louis EFL on June 3, 2006. I saw this large gathering of children all dressed in blue shirts that read, "We support Jenny." As I approached the group I discovered, in the middle of the crowd of blue shirts, a little girl in a wheelchair with a shirt that said, "I'm Jenny."

I have had the privilege of keeping in touch with Jenny and have used her story as an example for many, including me, on how to be part of the cure and not a victim of this disease.

I want to thank Jenny and her parents, Joni and Mark, for reminding me how nice Holland can be.

"When you change the way you look at things the things you look at change!" - Dr. Wayne Dyer

Chuck Mohan

Jennifer's Story

We have been blessed with two amazing daughters. Nikki (age 22) and Jennifer (age 20) were born almost exactly two years apart and have been very close their whole lives. However, their lives couldn't be more different. While Nikki has had the "typical life" (currently a graduate student), Jennifer has Leigh's Disease.

It has been suspected since she was 2 and proven a few years ago when genetic testing found two different point mutations of the NDUFV1 gene, one from mom and one from dad. NDUFV1 encodes the part of Complex I that binds NADH, starting the electron transport chain. Leigh's usually presents early in the first year of life and survival is short for most. Our girl had other ideas.

Jennifer had mild symptoms from about age one. She was very slow to develop physical skills such as crawling and walking. However, the neurologist did not show concern as she was progressing, and no diagnosis was made. At 2 ½ she was a "normal" little girl who loved to talk, run, and play with her sister and friends. At 33 months (March 2000), she suddenly stopped walking. After an MRI, significant damage in the fine motor control region of her brain was discovered and she was diagnosed with a mitochondrial disease. At this point, we started our journey. The shock that our girl had a disease we had never heard about and whose life wouldn't be typical was overwhelming. We had no idea where this journey would take us. Looking back, we are glad we did not know as one step at a time is how we needed to take life then and continue to take life today.

A muscle biopsy was done a couple of weeks later to help determine which type of the disease was affecting her. On April 1, 2000, the day after the biopsy, Jennifer's body began to shut down under stress. We have since learned she is very sensitive to anesthesia. She began taking very deep breaths, farther and farther apart. By the time we rushed to the hospital she was breathing less than two times per minute. She has placed on a ventilator at the PICU at St. Louis Children's Hospital. We will never forget that night, leaving her in the PICU and not knowing what was going on. We had thought her diagnosis meant her not being able to fully participate in life, not that her life might end.

Our little girl surprised everyone and after a week Jennifer was off the vent, in a regular hospital room, and we planned to head home the next day. However, she struggled that night and would not sleep. By morning, she stopped breathing and once again an emergency intubation was performed. She was placed back on the ventilator in the ICU. Later that day, the doctors gave us the diagnosis of Leigh's disease telling us it was one of the deadliest forms of mitochondrial disease. They gave us little hope of her ever getting out of bed again and asked if we wanted to move forward with treatment or not. We told the doctors that she was in God's hands and to continue treatment hoping she would get better. And she did!

After two months in the hospital Jennifer came home, but she came home a different child. She had a trach and a ventilator at night to help her breath. She could eat very little and was fed through a G-tube. She could no longer walk, stand or even sit on her own, and did not have the muscle control necessary to speak. Every sickness left her weaker than before and months if not years of hard work are required to recover lost skills. Gradually even these skills have all gone. However, Jennifer has always had a great attitude and caring heart.

In 2005, we became involved with the UMDF and wrote this in a letter asking for support from friends and family for the first St. Louis area fund raiser:
"Five years have passed since Jenny was diagnosed with Leigh's, a mitochondrial disease, and we began the role of parents of a disabled child. One minute we cannot believe it has been five years and the next it seems as if it has been much much longer. Even though we try to live a "normal" life, every decision we make is affected by Jenny's disability. When she is doing good, we are doing good. When she is not, we are not. Five years ago, we were told that Jenny would probably be bed ridden and on a ventilator for the rest of her short life. We had temporarily lost all hope. However, we refused to give up and she is now a thriving seven-year-old. Yes - even though Jenny cannot talk, eat, walk, sit up or even roll over by herself - she is thriving. Jenny loves life and her attitude warms all those around her. She is a special child. She is in first grade now and loves school and her friends. Her friends love her also. She is not treated badly because of her disabilities but instead treated special. Everyone who gets to know Jenny grows because of the experience. Unfortunately, I am describing the good times. During the bad, she is home from school. We constantly worry about getting her enough nutrition since she cannot keep anything down. She loses the strength she gained from months of therapy in just a few weeks. After one bout with pneumonia she even lost the ability to smile. So far, she has always been able to recover from the bad times though usually not fully. Each time her high point is just a little lower. The smiles have come back every time."

Moving forward on our journey to today. We are nearing 18 years since her diagnosis and we are blessed to still have those smiles! We also have a small "ICU" in an addition we built on our house to manage Jennifer's care, two wheelchair vehicles, an elevator, extensive equipment for her care too long to list, in-home nursing most days, and therapists and teachers both in and outside of our home.

We met Chuck Mohan at that first event in St. Louis. We didn't know his story at the time. He was just from the UMDF with turtle in tow. Chuck gave so much of himself to all of us affected by Mito even after Mito was done taking from him. We are eternally grateful for all he has done for us and for all affected by these diseases. In no small part for his gentle nudges that there was and is hope and help available for people with Mito.

During our journey, we have chosen to avoid the conservative do nothing approach many clinicians recommended. Instead seeking out caregivers willing to treat with caution. However, choosing to treat the unknown has its risks. There is good reason for the oath "First, do no harm." Jennifer has been hurt instead of helped many times by the most well-meaning doctors who just wanted to do something. An abnormal metabolism won't always respond as expected to standard medical treatments. A good balance between caution and willingness to treat is hard to find but worth the search. With nearly two decades of hindsight we can see that, overall, the choice to seek treatment has been the right course. We have learned to never give up and to continue moving forward on the journey one step at a time.

Life has certainly not turned out as we expected when Jennifer was born a normal child 20 years ago. Our lives have been forever changed. We don't know if it would have helped 18 years ago, but Emily Perl Kingsley's poem "Welcome to Holland" rings true to us today. We have

been blessed to be able to "enjoy the very special, the very lovely things about" Jennifer.

> ***WELCOME TO HOLLAND***
> ©1987 by Emily Perl Kingsley
>
> I am often asked to describe the experience of raising a child with a disability to try to help people who have not shared that unique experience to understand it, to imagine how it would feel. It's like this….
>
> When you're going to have a baby, it's like planning a fabulous vacation trip to Italy. You buy a bunch of guide books and make your wonderful plans. The Coliseum. The Michelangelo David. The gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting.
>
> After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later the plane lands. The stewardess comes in and says, "Welcome to Holland."
>
> "Holland??" you say. "What do you mean Holland?? I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy. But there's been a change in the flight plan. They've landed in Holland and there you must stay. The important thing is that they haven't taken you to a horrible, disgusting, filthy place, full of pestilence, famine and disease. It's just a different place.
>
> So you must go out and buy new guide books. And you must learn a whole new language. And you will meet a whole new group of people you would have never met. It's just a different place. It's slower paced than Italy, less flashy than Italy. But after you've been there for a while and you catch your breath, you look around… and you begin to notice that Holland has windmills…. and Holland has tulips. Holland even has Rembrandts. But everyone you know is busy coming and going from Italy…and they're all bragging about what a wonderful time they had there. And for the rest of your life, you will say "Yes, that's where I was supposed to go. That's what I had planned."
>
> And the pain of that will never, ever, ever go away…because the loss of that dream is a very, very significant loss. But…if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things…about Holland.

Team Highlight
Team Addison, EFL Detroit

The 2018 Energy for Life Walkathon season is in full swing. The walks are critical to funding needed research, providing education opportunities and supporting families. There are so many different ways to get involved to support a walk in your area and we would like to recognize one of our fall walk teams that is doing just that.

It was the second year of participation in their local Energy for Life Walkathon for The Szopo family of Northville, Michigan. The past year was pivotal for their daughter, Addison. She had made many developmental strides, and it energized and empowered Addison's family to get involved with the walkathon. Mom Leah shared her family's story and struggles with her employer of five years, SpringCM. The company immediately rallied around Team Addison.

"I kicked off our fundraising efforts with Addison's story and an explanation of what we will be doing during Mito Awareness Week." Flyers, posters, mito facts, pictures of Addison and fundraising tallies were sent in company-wide emails.

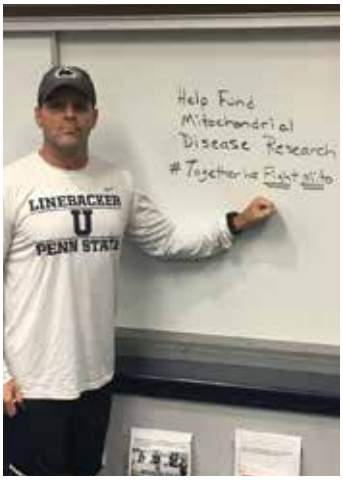
"The response was astonishing. I owe our fundraising success to my amazing co-workers and the volunteer organizers that encouraged me to share Addison's story and talk about mitochondrial disorders and the impact that it has had on our family" say Leah.

Team Addison's impact was sure felt in Detroit! The team raised over $8,500 and SpringCM signed on as a Corporate Partner to provide additional financial support.

In sharing their story this year, The Szopo family was showered with support and open hearts. They were able to impact others, educate about mitochondrial disease, and support the overall UMDF mission.

Your story could do the same!

To find a walkathon near you, please visit www.energyforlifewalk.org. For fundraising ideas, support to grow your team or to discuss how to get involved, please reach out to us!



#TogetherWeFightMito

Have you seen or been a part of the #TogetherWeFightMito movement on social media? From the Selfie Challenges with our entire community and public figures, to selling bracelets and t-shirts, to pumpkin decorating contest, the campaign has brought our community together in new, fun ways. But do you know the story behind this campaign and their connection to the UMDF?

Together We Fight Mito is a grassroots campaign led by a group of parents whose children are diagnosed with mitochondrial disease. One day, not long ago, one of the moms from this group was discussing how the recent international attention for a very beloved mito angel made her feel less alone in her own journey. A lady in the group named Laura responded with a simple, yet powerful hashtag, #TogetherWeFightMito.

And from that meaningful hashtag, this awareness campaign was born. The campaign co-founders recognized an urgent need for energizing the international patient community through a public awareness campaign to educate the world about the many forms of mito, and the fact that there are currently no cures for this progressive and often fatal disease. They believed we are stronger together, and that we can raise greater awareness and research funding when affected individuals and families pool their efforts and combine their voices for change.

The co-founders reached out to the UMDF and asked for support of their campaign with a website, with promotion and that funds raised would benefit research grants and the scientific investment initiatives of the UMDF mission. Director of Development at the UMDF Beth Whitehouse says of the group "We are so thankful for this group of mito-warrior moms and dads, coming together to raise funds and awareness for mitochondrial disease. In just four short months these amazing individuals have raised over $4,500. It's so important for our families to realize they are not alone in this battle, and the more we can do together, the more progress will be made to find effective treatments and cures. It only takes one individual to lead a charge, but together we are powerful and we will make change happen."

Co-Founder Glenda McCoy says although most of the moms have never met one another in person, they feel connected through their similar stories and struggles. "We are just normal parents trying to save our children. Most of us have never met - we found one another online through our children's personal awareness pages. Each day we shout out our stories to anyone who will listen with the hope that today will be the day that the world listens and funds this critical research."

The group feels that together, they are stronger and they are spreading hope. "Hope that the most common disease most people have never heard of becomes a household name; hope that people realize that curing mitochondrial dysfunction helps us all; hope that this becomes the next frontier of medicine; hope that the world can come together to raise our collective voices to fund mitochondrial disease research before our children die" says Glenda.

And they are very excited about growing this campaign more in 2018, expanding their efforts to raise more awareness and research funding. Watch for a fun and meaningful way to participate in the #TogetherWeFightMito campaign during Valentine's Day. And check out the UMDF page supporting the initiative at https://www.umdf.org/togetherwefightmito.

Symposium Preview

Mark your calendars for June, as UMDF brings the national symposium to Nashville. Scientist and clinicians will gather June 27-30, 2018. The UMDF Patient/Family will be held June 29-30, 2018. LHON program runs June 28-29.

For patients and families, 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. You will also be able to speak with our exhibitors about new products and diagnostic testing that is available.

Our Scientific and Clinical sessions are being led this year by Vamsi Mootha, MD, from the Harvard Medical School. The theme this year will be Mitochondrial Chemical Biology. Bruce Cohen, MD, from Akron Children's Hospital, is the CME Chair for the symposium.

SAVE THE DATE!

Mitochondrial Medicine 2018: Nashville
Sheraton Music City, Nashville, TN
Scientific/Clinical Sessions: June 27-30, 2018
LHON Sessions: June 28-29, 2018
Patient/Family Sessions: June 29-30, 2018

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

Corporate Partnership Spotlight

tishcon corp.
&
epic4health .com

Who are they?
Tishcon Corp was founded in Westbury, New York by Raj Chopra, Vipin Patel, and others in 1976 as a contract manufacturer of bulk products, mostly for mail order marketers and wholesalers. Over the years, the company has expanded to offer a full line of services: from packaging and labeling to new product development. A second location, in Salisbury, Maryland was established in 1984.

Presently, co-founder Raj Chopra serves as CEO. Chopra has an extensive background in various disciplines related to the nutriceutical, vitamin and supplement industry. In addition to previous work with contract manufacturers, Chopra has expertise in formulation, product development, nutrition research, and food technology. This range of experience has given the company a strong basis for expanding its product offerings.

Today, Tishcon operates facilities in New York and Maryland. Westbury is the home to softgel encapsulation, Research and Development (R&D) laboratories and quality control (QC) testing labs. The Salisbury operation houses encapsulation and tableting operations, as well as all bottling, packaging and distribution. A satellite laboratory in Maryland handles QC testing during production. All purchasing and accounting functions are also located in the Salisbury facility. Tischon is well known for their product Q-Gel (Coenzyme Q10) and nutritional supplements that support the mitochondrial disease patient cocktail.

What do they do for UMDF?
Tishcon and Epic 4 Health have been long-time, generous partners in the fight against mitochondrial disease and have supported UMDF since 2008. Tishcon continues to support our National Symposium educational and scholarship fund and is one of the founding members of our Industry Advisory Council. Marko Rosa and George McShane have attended each of our symposia providing support and product information to our families.

Tishcon says…
It is absolute privilege to be a part of UMDF family. Meeting patients and families at Symposium and hearing their personal challenges, success, and dedication to the cause, has been both motivating and humbling experience. UMDF professional staff members are absolute pleasure to work with and are beyond accommodating, as they are attentive to the needs of patients and their families. We are looking forward to seeing everyone in Nashville for 2018 UMDF Symposium!

UMDF Staff says…
Every staff member looks forward to seeing Marko and George at our National Symposium each and every year. They have been long-time members of the UMDF family and we are forever grateful for their generous support, and also their friendliness and their dedication to our cause and our families.

UNITED MITOCHONDRIAL DISEASE FOUNDATION®

Adult Advisory Council Team

EST: 2006

AACT TEAM

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Sharon Shaw- Arizona
Jennifer Schwartzott- New York
Gregory Yellin - Maryland

Medical Advisors:
Bruce Cohen, MD
Amy Goldstein, MD

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.

AACT - ADULT CORNER PAGE

On behalf of the UMDF Adult Advisory Council Team (AACT), wishing you and your family a healthy, happy New Year. May peace and joy fill your hearts in 2018.

2018 will be another busy year. We hope to see you at upcoming UMDF meetings, events and especially at Mitochondrial Medicine 2018 in Nashville the end of June. Once again, a number of Adult and Young Adult Sessions, Workshops and Gatherings will be on the Family Program. We know it will be another informative, helpful and fun Symposium.

To view UMDF's Calendar, click here.
http://www.umdf.org/events

AACT is excited and happy to announce two new Council Members have joined the Team!

We are proud to now serve all of the UMDF U.S. Regions, including Canada. We welcome Nicole and Lillian and here are their stories:

"My name is Nicole DeJean and I live in Lafayette,Louisiana. I was diagnosed with Mitochondrial Disease in 2012 after it was determined that my daughter and I share a common mutation. I have been involved with UMDF since our diagnosis journey began in 2010."

"My name is Lillian DeJean, I am fifteen years old, and I live in Lafayette, Louisiana. In 2008, I was diagnosed with maternally-inherited Mitochondrial Disease. I have been involved in advocacy at the state and national level, and was recently selected to as the first ever youth member of the Louisiana Developmental Disabilities Council"

Lastly, be alert throughout the year for special AACT e-blast announcements. AACT is honored to represent, serve and support you. To 2018!

Joy & Gail
UMDF AACT Co-Chairs

Dr. Mark Tarnopolsky's Exercise Tip

"A combination of endurance exercise (walk, jog, bike, hike, swim) and resistance exercise (weights, Pilates, yoga, core exercise) on different days of the week is recommended. Remember to listen to your body and starting at low intensity and gradually increasing duration and/or intensity and try different activities until you find what you enjoy the most."

- Mark Tarnopolsky, MD, PhD, FRCP(C), Professor of Pediatrics and Medicine, CEO and CSO, Exerkine Corporation, Director of Neuromuscular and Neurometabolic Clinic, McMaster University Medical Center

Ask the Mito Doc℠

Living with mitochondrial disease presents many twists and turns, and a maze of questions. UMDF is pleased to offer answers to some of those questions as taken from Ask the Mito Doc℠ at www.umdf.org. Please note that information contained in Ask the Mito Doc℠ is for informational and educational purposes only. Such information is not intended to replace and should not be interpreted or relied upon as professional advice, whether medical or otherwise.

Q: My doctor with Functional Medicine has placed me on the Paleo diet. I have followed this diet closely for 2 weeks. I feel worse. I have already removed sugar and processed food from my diet 9 months ago. Is there any research in support of Paleo diet can help with mitochondrial disease?

A: There is no research on the paleo diet.The diet needs for mitochondrial patients vary depending on the type of disease - some do better with lots of carbs - while others with less. This needs to be individualized. All will benefit from reducing processed foods and simple sugars.

Sumit Parikh, MD

Sumit Parikh, MD

Q: Can an MRI of a patient with mitochondrial disease look similar to an MRI of a patient with MS (brain/Spine). In other words, can similar patterns on demyelination occur in both diseases?

A: In the strictest sense - the MRI in an MS patient would not look like an MRI seen in mitochondrial disease. Some features may overlap - but the patient's clinical course, exam and MRI findings should be distinct enough to allow for an expert to distinguish between the two. If that is not enough - the lab work and spinal fluid analysis usually helps.

Sumit Parikh, MD

Q: The Handbook says avoid erythromycin. Does avoidance include erythromycin ophthalmic ointment?

A: Erythromycin is not an absolute contraindication; it is often used for mitochondrial patients with dysmotility. There is no contraindication to using it as an ointment either.

Sumit Parikh,MD

Q: My son has KSS and we are considering growth hormone treatment. What is your opinion on this? He has retinitis pigmentosa as a result of KSS.

A: Thank you for this great question. As a general rule, growth hormone deficiency is suspected by plotting out the linear growth (height or length), as well as weight, over time and determining that the linear growth is not sufficient. There are other symptoms that can be a sign as well including emotional or cognitive issues. Adults (and some children) with growth hormone deficiency will also have abnormal lipid and cholesterol levels. It is important to note there are many reasons why some people with mitochondrial diseases may not grow well or feel cognitively or emotionally "normal" – and these do need to be considered. For example, renal tubular acidosis, which is common in some forms of mitochondrial disease, will interfere with growth, and some forms of mitochondrial disease have short stature as a key feature, even without growth hormone deficiency.

Bruce Cohen,MD

Sometimes X-rays of the wrist are used to determine bone age, which is a helpful study in some cases. Finally, blood tests will be performed to include growth proteins known as IGF-I and IGFBP-3, other hormone levels in the blood and a growth hormone stimulation test. If a person with a mitochondrial disease, including Kearn Sayre Syndrome, has been diagnosed with a growth hormone deficiency, I generally recommend therapy with one of the commercial products used to treat the growth hormone deficiency. During the evaluation other levels of key hormones may be found to be abnormal, and would need to be treated.

Some of my patients treated with Growth Hormone have demonstrated clear cut improved growth and an improvement in the sense of well-being, in which case we continue the therapy. In other patients there is no benefit. I do not recall any patients that have had bad side effects, but this is obviously a concern when starting any new therapy in a patient with a mitochondrial illness.

Bruce Cohen,MD

Q: I am an adult with ox phos disorder and I developed a stiff shoulder and neck on same side. I am very sensitive to anyone moving the shoulder or to being startled or to loud sounds. My pmd who follows me now tested for GAD 65 antibodies which were absent. She says since the enzyme in this disorder is located in the mitochondria in the brain that probably it is related to my mito disease as I have so many other lab results that are abnormal from mito enzymes. When I get startled I get incapacitated with gasping and sweating and bounding heart. She is treating me with B6 and since I noticed sometimes that what I eat makes a difference she is trying a lower glutamate diet. But B6 has toxicity too. Since there is nothing we can find written about this except for the autoimmune kind, I wondered if you have ever seen this syndrome in mito patients."

A: Hard to answer but in general stiff shoulder and neck by itself are not a primary mitochondrial disease issue.
Sumit Parikh,MD

Q: Why is leukovorin prescribed in a mito cocktail?

A: Folate requires energy to get converted to a form that can easily get into the brain. Some mitochondrial disease patients have trouble getting enough folate into the brain in-part due to an energy deficiency. A brain folate deficiency can cause problems with brain health and proper brain growth. Leucovorin is prescribed as it is a form of folate that can get into the brain unlike over-the-counter folic acid. Some mitochondrial diseases - such as Pearsons or Kearns-Sayre syndrome - are more likely to develop this problem.

The only way to know if a patient has a brain folate deficiency is to measure levels via a spinal tap. This is not done routinely as it is an invasive procedure. So for many mitochondrial patients we give Leucovorin empirically in case they need it. Depin (methyl-folate) is another form of folate that can also be used if Leucovorin is not covered by insurance.
Sumit Parikh,MD

Q: Have you seen glutathione or idebenone work for people with mitochondrial disorder who have trouble swallowing? I know someone with lifetime history of this problem now getting worse with age

A: There is no clear evidence to say that glutathione or idebenone can help with trouble swallowing.
Sumit Parikh,MD

Q: What relevance does elevated 3- methylglutaconic acid (or any organic aciduria) have as a marker for mitochondrial disorder? When my son first presented with a development regression and language stagnation, he also had symptoms of a mitochondrial disorder (constipation, dysmotility, fatigue, and pseudo-obstruction for starters). Early mito labwork did not show any mitochondrial disease but suggested mito dysfunction. He did not have a muscle biopsy. Whole Exome and mitochondrial sequencing also did not yield any findings. A couple of years later we discovered that my son had a rare childhood epilepsy (ESES). While searching for a root cause for the epilepsy, we began new metabolic investigations. Recent testing of blood, CSF, and urine have shown elevations of 3MGA. It does not appear that this is a case of an inborn error of metabolism, as it was not always elevated, nor was it detected at newborn screening or in Whole Exome sequencing. What might these elevations mean and what additional tests would rule in or out mito conditions?


Russ Saneto, DO

A: I am sorry to hear about your son. Biochemical abnormalities can be perturbations in metabolism that are often due to environmental changes such as diet, stress, exercise, medication, as well as disease. For example, eating lots of yogurt can produce abnormalities in the urine organic acid profile, that are clearly not a disease manifestations. Running a marathon can break down muscle and increase serum creatine kinase levels, again not a disease. In our workup to investigate possible mitochondrial disease, as well as other metabolic disease possibilities we do initial biochemical labs. Persistence in abnormalities and the conditions we see them can give clues to possible disease. One of the tests we do is look at the urine organic acid profile, and one component of the organic acids is 3-methylglucaconic acid. This acid can be elevated in multiple diseases, one of which is mitochondrial disease. But, occasionally this acid can be elevated in non-disease states. The context, meaning the other abnormalities would give some hints whether elevations of this specific urine acid is pathological. The job of the expert is to put all the pieces of the puzzle together to make an educated determination if the abnormalities on biochemical testing indicate a specific or group of diseases or not. Most of the time, this particular urine acid would not indicate disease, unless it is quite elevated on multiple occasions and when levels are taken during a more-or-less controlled state. Not knowing the complete work-up your son has had, I cannot give you the exact reason for the elevation, but does not sound like it was due to a disease.

Eliminating the persistent epileptiform discharges in ESES can be tough to do. I hope the right combination of anti-seizure medications are found for your son.
Russ Saneto, DO

UMDF

events

The energy providing education,support and research.

Past Fundraisers Benefitting the UMDF

November 18, 2017 The Lee Brothers Sidekick Foundation's Mad Dash benefitted the Aiden Lee Research Fund for a second year. The race featured three courses for adults, children & a walk that is filled with physical and mental challenges. This event, kind of like the Amazing Race, is filled with mito trivia! Over $16,000 was raised for the Aiden Lee Research Fund! Thank you, Lee Family and the Sidekick Foundation!

September 2017 The Upstate New York employees at M&T Bank held a Jeans Day event for the month of September. The employees donated over $1,370 to the UMDF in honor of Caroline Payne. Thank you to M&T Bank in Upstate New York for your support!

September 2017 The Hoekstra Family of Rock Valley, Iowa, hosted an event in honor of Autumn Hoekstra! The event raised over $700 for the UMDF! Thank you to the Hoekstra's for your support!

September 1, 2017 Friends and Family held their annual Nathan Mowrer Memorial Horseshoe Tournament in Pottstown, PA. Over $400 was raised in Nathan's Memory for the UMDF. Thank you Mowrer Family!

September 3, 2017 Next Yoga in Wheaton, Illinois held a donation class in memory of Luke Schiling to support EFL: Chicago. The event raised $200! Next Yoga has been a multi-year supporter of our mission – thank you!

September 6, 2017 Team Megan of EFL: Indianapolis held a bake sale fundraiser and raised $118. Thank you Team Megan!

September 8, 2017 The Stewart Family held their 5th annual Birdies for the Blind Golf Scramble at Gardner Golf Course in Massachusetts. This annual event is held in honor Shane Stewart. Way to go Stewart Family for raising $6,000!

September 9, 2017 The 2nd Annual Kennedy Burgess Memorial Ride was held in Manitowoc, Wisconsin. The Burgess family organizes the event to remember and celebrate their daughter Kennedy's birthday each year. Thank you for honoring the UMDF and Kennedy's memory through this event!

September 9, 2017 Our Indianapolis EFL team River Strong held bracelet and t-shirt sales to fundraise for their team. River Strong was the top fundraising team for the Indy EFL, raising over $3,193! Thank you River Strong!

September 10, 2017 A Cause an Effect Chipotle fundraiser was held in Edina, Minnesota, by EFL team Renewing Hope. The event raised $298. Thank you Nikki Abramson for organizing the fundraiser!

September 11, 2017 Kendra Scott Gives Back Fundraiser was held to benefit the Central Texas Energy for Life walkathon. The event raised $888. We are grateful to all who supported the event and our partner Kendra Scott!

September 16, 2017 The Annual Chris' Cakes Pancake Feed was held in conjunction with the Energy for Life Walkathon in Kansas City. Thank you to Amy Ray for helping to organize the event! We love Chris' Cakes!

September 16, 2017 The After Party at the Lodge was held after the Kansas City Energy for Life Walkathon. Thank you to Deidra Achtley, the Achtley family and supporters for organizing this fun post-walk event!

September 17, 2017 A Kendra Scott Gives Back fundraiser was held in Indianapolis, Indiana. Proceeds from the event supported the Energy for Life walk in Indianapolis. The event raised $345!

September 22, 2017 Saint Paul School in Louisville Kentucky held 2017 Mito Awareness Week events and fundraisers

to support local student Harper Johnson. The school raised over $571 during Awareness Week! Thank you Saint Paul School!

September 23, 2017 The Emma Boggs Research Fund held a CrossFit Workout Fundraiser to raise money for their research fund. We thank you for combining awareness, activating your mitochondria and fundraising into an event for the UMDF!

September 23, 2017 Want an idea for a unique fundraiser? Mom Carol and Uncle Bruce of The Somers 7 challenged each other in a fundraising contest for EFL: Southern Wisconsin. The Somers 7 raised over $1,318 – thank you!

September 23, 2017 The Jakubowski family of New Berlin, Wisconsin arranged an opportunity for our supporters of the EFL: Southern Wisconsin to win a chance to throw out the first pitch at a Brewer's game. Thank you Morgan's March for your support!

September 23, 2017 The 2017 Mito Awareness Walk held at the AnMed Health Women's & Children's Hospital was another great success! Over $1,000 was raised for the Samuel Cutliff Research Fund. Thank you so much for your support!

September 23, 2017 The Idaho Mito Group hosted their annual 'Mito Bowl' – this year fundraising over $200! Thank you to Jennifer Pfefferle for your continued support!

October 12, 2017 *A Candlelight Karma Yoga fundraiser was held at Soul Studio to benefit EFL: Minnesota. Thank you to Aiden's Red Wagon and Soul Studio for organizing this event!*

October 2017 The Ribeiro Family hosted a Pizza Fundraiser in honor of Colton Ribeiro – the event raised over $370 for the UMDF! Thank you for your support!

October 1, 2017 The 5th Annual Hope for Brionna 5K and 1 Mile Fun Run was held in Terre Haute, Indiana. The event's proceeds support the Brionna Myer's Research Fund and raised $13,500. Thank you Myers family and all of Brionna's supporters!

October 1, 2017 First United Methodist Church in West Allis, Wisconsin held a fundraiser to support Ken's Karetakers of the Energy for Life Walk in Southern Wisconsin. Thank you First United!

September 15, 2017 *The 4th Annual Carlos Alberto Memorial Golf Outing was held at the Pipestone Golf Course in Miamisburg, Ohio. Teams and sponsors came together in memory of Carlos Alberto to raise $12,000 for the Carlos Alberto Memorial Research Fund. Thank you, Cristina Rue & Family!*

October 1, 2017 Jason Steele held the annual Olivia Lauren Steele Golf Outing this October. The event raised awareness and almost $2,000 for the UMDF! Thank you Steele family for your support!

October 7, 2017 Chris and Nicole Florio held the 2nd Annual Luca's Legacy Golf Outing in memory of their son, Luca. This year's event was held at the Cannon Ridge Golf Club in Fredericksburg, Virginia. The event raised over $8,000 for the Luca Florio Research Fund. Thank you Florio Family!

October 7, 2017 The Dinner in the Dark event in Scottsdale, Arizona, was held in support of the LHON Project Fund with the UMDF. Jessica Loomer did another amazing job this year and was able to raise over $3,000 with this unique fundraising event! Thank you Jessica!

October 8, 2017 Isabel Taylor held a Kendra Gives Back Night in Nashville Tennessee for the Energy for Life Walk this spring that raised over $400. Isabel is a college student in Nashville and is looking forward to helping with the walk and our National Symposium for 2018, all in her brother Simon's memory. Thanks, Isabel!

October 8, 2017 Team Hope Energy Life participated in the Bank of America Chicago Marathon. The 17 members raised over $14,515 leading up to their

26.2 miles. Thank you to all of our runners and their supporters!

October 12, 2017 The 9th annual C.U.R.E. ride was held to raise funds for the LHON Project Fund with the UMDF. Riders rode from Santa Barbara to San Diego, California, and raised over $30,000!! A HUGE thank you to the Poincenot Family and Friends for such an amazing event!

October 14, 2017 Happy 40th Birthday, Matt Calhoun! Matt celebrated his 40th by requesting donations to the UMDF in his honor! Thank you, Matt!

October 14, 2017 The family of Chesnee Hooks participated in the Akins Fall Festival where food proceeds were donated to the UMDF! Nearly $1,000 was raised and donated in honor of Chesnee! Thank you to the Hooks Family!

October 25, 2017 The Lieberman, Thomas & Hagan Families hosted a Pumpkin Carving Contest this fall. Their event, which included silent auctions and a basket raffle raised $1,000 for the UMDF! Thank so much Jessica, Heather and Gina for all your support!

October 28, 2017 Sydney Breslow hosted 2 Kendra Scott Give Backs nights in Raleigh, North Caroina! Two stores in the Raleigh area participated and helped to raise over $550 for the Logan Sloane Memorial Research Fund in honor of Sydney Breslow. Thank you Breslow family for all your love and support!

October 28, 2017 Monica Manning of Ohio, held a Kendra Scott Gives Back Night in Columbus, Ohio, for her walk team 'Miles for Myles'. The Columbus community rallied together to raise over $700 in memory of Monica and Mark's son, Myles. Thank you Manning Family!

December 2, 2017 *Nebraska Lutheran High School in Waco, Nebraska held a Knights Shoot Out For Mito Fundraiser. The event was organized in honor of Wyatt Beiermann and raised $4,530. Thank you to the school and entire community of Nebraska Lutheran!*

November 5, 2017 A Kendra Scott Gives Back fundraiser was held in St. Louis to benefit the Energy for Life Walkathon. The event raised over $749! Thank you St. Louis supporters!

November 19, 2017 Corrie Racine and Colleen Robichaud held the 4th annual Mito Bowl in Wilmington, Delaware, at Pleasant Hill Lanes. This annual event which included two hours of bowling and a basket raffle, raised over $2,000! Thank you so much, Corrie, Colleen and the 4 East Staff for your support!

November 26, 2017 Privé Salon and Style Bar in Newtown Square, Pennsylvania, held a Style-a-thon event in memory of Riley Ingelsby. The event raised over $1,800 for the UMDF! Thank you so much for support the UMDF!

December 2, 2017 The 4th Annual Carter's Christmas was held at the Sayre Elk Club. The Christmas Vendor & Craft show included a Chinese auction, basket raffle, memory tree and gifts from Santa in memory of Carter James Lackey. Over $2,000 was raised for the Carter Lackey Family Research Fund. Thank you so much for all your support, Brittany and James!

December 2017 The Octorara Area High School Field Hockey Team in Atglen, Pennsylvania, held their annual event 'Stick it to Mito' in memory of Wyatt DeStephano and in honor of his sisters. The Varsity Field Hockey players helped kids through six different skill stations for kids in grades K-6. Wyatt's sister, goalie for the Varsity Team participated in the event. Thank you Octorara Field Hockey for your continued support!

December 21, 2017 The friends & family of Cameron Genie hosted a fundraising event with the Springfield Thunderbirds raising nearly $900 for the UMDF! Thank you to the Genie Family for your continued support year after year!

January 14, 2018 Neil's Quest of the Chicagoland area held a gives back event at Barnaby's Family Inn in Arlington Heights. The event raised $350! Thank you to the Mowery family and all of Neil's supporters!

December 3, 2017 *The Kavan family of Overland Park, Kansas participated in a Mitzvah Marketplace fundraiser to benefit Team Zoe and the Energy for Life Walk in Kansas City. The event raised $260 – thank you Kavan family!*

EFL: Southern Wisconsin

Upcoming Events

January 2018 St. Bernadette Catholic School in Monroeville, Pennsylvania, will be participating in their 17th Annual Coins for a Cure Campaign in memory of Gina Mohan, who attended St. Bernadette's school. The winning classroom will receive a movie and pizza party!

February 10, 2018 UMDF will be partnering with Kendra Scott in Las Vegas, NV for a 'Kendra Gives Back Party' at the Fashion Show Mall location! Stop in and enjoy sips, sweets and some beautiful jewels – plus, 20% of the proceeds will benefit the UMDF!

March 24, 2018 The 9th Annual Jackson Culley Mito-What? 5k will be held in Millington, Tennessee. Registration is open until March 19. Learn more at *https://racesonline.com/events/mitowhat5k*.

March 31, 2018 Fitness 1440's Fools 5K in Seymour, Indiana, will benefit the UMDF. Learn more and download a race brochure here: *http://indianatiming.com/Entry%20Forms/2018%20-%2003-31%20Fools%205K.pdf*

April 7, 2018 Bet on Baylee is back again for 2018 – its 14th year! You can join in the silent auction and basket raffle at the Roseville Community Center in Roseville, Ohio. Reach out to *nicole@umdf.org* for more information!

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 Marathon. For more information, please visit *www.crowdrise.com/umdfpitt2018*.

June 23, 2018 The Nicholas J. Torpey Butterfly Golf Classic will be held Sycamore Hills Golf Club in Macomb, Michigan. Thank you to Jennifer Ruhana-Smerek for organizing this annual event.

July 2018 Rachael Howard will be participating in the 500km Last Annual Vol State Road Race and using this as an opportunity to fundraise in honor of her nephew, Theo. Rachael will be Activating her Mitochondria for sure on this race! Help Rachael meet her goals at *www.umdf.org/activemito* and search for Rachael!

October 8, 2018 Team Hope Energy Life will be represented at the Bank of America Chicago Marathon. The team currently has 25 members. If you would like to learn more about our runners or support their efforts visit *https://www.crowdrise.com/o/en/team/bank-of-america-chicago-marathon-2018*

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. 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.

REGISTER NOW!

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.

EFL Walkathons

August 19, 2017 The Energy for Life Walkathon in Minnesota was held for the 8th year! It was a beautiful, sun-filled day at Normandale Lake Bandshell with 27 teams raising $53,850. Thank you to all of the volunteers and families that make this day so special for all!

September 9, 2017 The Energy for Life Walkathon in Indianapolis was held at Hummel Park in Plainfield, Indiana. This year's Superhero theme was enhanced by a visit from Courage and Kindness princesses, who stopped by to take photos with our families and walkers. The event raised $14,854. Thank you Indianapolis!

September 16, 2017 The Energy for Life Walkathon in Detroit moved to a new location this year, James C Nelson Park in Sterling Heights, Michigan. The event was attended by over 215 walkers and raised $27,978. Thank you to the 13 teams and amazing committee and volunteers in Detroit!

September 16, 2017 The Energy for Life Walkathon in Kansas City was held at T-Bones CommunityAmerica Ballpark. 14 teams gathered to support the UMDF mission and connect with each other. The event raised $48,788 exceeding their goal. Thank you Kansas City!

September 16, 2017 The Energy for Life Walkathon in Western New York was a success! The day started off with great weather and plenty of smiles! The walk in Cheektowaga, New York had a great year with meeting their goal of $20,000. Way to go, Teams, Volunteers and Supporters!

September 17, 2017 The Energy for Life Walkathon in Chicago was a great celebration this year. We honored two long-term volunteers with tributes, a slide-show and cake! It was a beautiful day at Katherine Legge Memorial Park and the 20 walk teams exceeded their goal raising $69,988. Thank you Chicago!

September 23, 2017 The Energy for Life Walkathon in Southern Wisconsin had a great honor this year with UMDF CEO Chuck Mohan attending! It was fun-filled day of face-painting and mito-rock painting. 14 teams raised $23,701 exceeding their walk goal. Way to go Southern Wisconsin!

September 23, 2017 The Energy for Life Walkathon in Delaware Valley had its biggest year yet! Blowing away our fundraising goal, the walk in Cherry Hill, NJ raised over $108,000! Congratulations to all our volunteers, teams and supporters for a tremendous day!

September 30, 2017 The Energy for Life Walkathon in Central Texas was held at Northeast Metropolitan Park in Austin, Texas. 14 teams gathered to raise $37,239 to support the UMDF mission. Thank you Central Texas!

October 1, 2017 The weather in Seattle held out for our 3rd Energy for Life Walkathon at Warren G. Magnuson Park! We gathered for a great morning of walking and supporting each other – plus were able to fundraise over $54,000! We thank all of our teams, walkers, volunteers and donors!

October 14, 2017 What another great year for Energy for Life Charlotte! Freedom Park was never more hopping than this year with amazing teams & music! Charlotte crushed their goal and raised $186,000! A huge thank you to all our teams, supporters and volunteers, we can't do it with out you!

November 18, 2017 The Energy for Life Walk in Fort Myers, Florida, was changed to a virtual walk due to Hurricane Irma. Our walk site, along with many of our families homes were severely impacted by damages left behind. If you have donations that you didn't get to bring to the walk, you can still make those at www.energyforlifewalk.org/southwestflorida.

EFL: Indianapolis

Upcoming EFL Walkathons

Our Spring 2018 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:

Minnesota
www.energyforlifewalk.org/minnesota

Indianapolis
www.energyforlifewalk.org/indianapolis

Western New York
www.energyforlifewalk.org/westernnewyork

Kansas City
www.energyforlifewalk.org/kansascity

Detroit
www.energyforlifewalk.org/detroit

Chicago
www.energyforlifewalk.org/chicago

Delaware Valley
www.energyforlifewalk.org/delval

Southern Wisconsin
www.energyforlifewalk.org/southerwisconsin

Central Texas
www.energyforlifewalk.org/centraltexas

Seattle
www.energyforlifewalk.org/seattle

Charlotte
www.energyforlifewalk.org/charlotte

Southwest Florida
www.energyforlifewalk.org/southwestflorida

COMING SOON:

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

April 7, 2018 - Houston
www.energyforlifewalk.org/houston

April 14, 2018 - Nashville
www.energyforlifewalk.org/nashville

April 21, 2018 - Tampa Bay
www.energyforlifewalk.org/tampabay

April 21, 2018 - San Francisco Bay Area
www.energyforlifewalk.org/sanfrancisco

April 28, 2018 - Dallas/Fort Worth
www.energyforlifewalk.org/dallasforthworth

April 29, 2018 - Atlanta
www.energyforlifewalk.org/atlanta

May 6, 2018 - New England
www.energyforlifewalk.org/newengland

May 20, 2018 - Columbus
www.energyforlifewalk.org/columbus

June 9, 2018 - St. Louis
www.energyforlifewalk.org/stlouis

June 9, 2018 - Pittsburgh
www.energyforlifewalk.org/pittsburgh

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!

UMDF advocacy

ORPHAN DRUG TAX CREDIT

UMDF joined the National Organization for Rare Diseases and 35 other patient advocacy groups in opposition to the proposed weakening of the Orphan Drug Tax Credit. The reduction of this credit could financially hurt biomedical research. With the Senate and House versions of the Tax Cuts and Jobs Act in conference committee, 125 people visited the UMDF's Advocacy Action Center an sent a letter to their federal officials opposing the weakening of this legislation.

BIOMEDICAL RESEARCH ACT

UMDF was asked by Senator Elizabeth Warren (D-MA) to be one of the Patient Advocacy Groups supporting her reintroduction of the Biomedical Research Act. We agreed to support. This important legislation would increase funding for targeted biomedical research initiatives through a new, reliable funding stream supporting the NIH and the Food and Drug Administration (FDA). The purchasing power of the NIH has been cut by Congress for more than a decade. This funding boost would help restore the NIH budget roughly to its 2006 levels, adjusted for biomedical inflation. The National Biomedical Research Act would create the Biomedical Innovation Fund, a new fund designed specifically to provide predictable investments in life-saving biomedical research conducted by leading scientists at the nation's top research institutions. The funding would provide $5 billion annually in supplemental funding for grants for young emerging scientists and breakthrough research that will help accelerate the development and approval of new medicines, improve prevention, and increase understanding of life-threatening diseases.

MEDICAL NUTRITION EQUALITY ACT OF 2017

Senator Casey (D-PA) asked UMDF to gather support for this important legislation co-sponsored by Sen. Chuck Grassley (R-IA). The legislation will, if enacted, cover medically necessary food and vitamins for digestive and inherited metabolic disorders under federal health programs and private insurance.

CONGRESSIONALLY DIRECTED MEDICAL RESEARCH PROGRAM (CDMRP)

We learned that Mitochondrial Disease has been included for another year in the Congressionally Directed Medical Research Program and the Department of Defense. This will mean millions in Mitochondrial Disease Research. In late December, we received an email asking us for help in recruiting patients, clinicians and scientists to serve as reviewers.

Want to learn more and lend your voice? Visit the UMDF Advocacy Action Center and TAKE ACTION on the issues impacting our community.

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.

WHAT UMDF REGION DO YOU LIVE IN?

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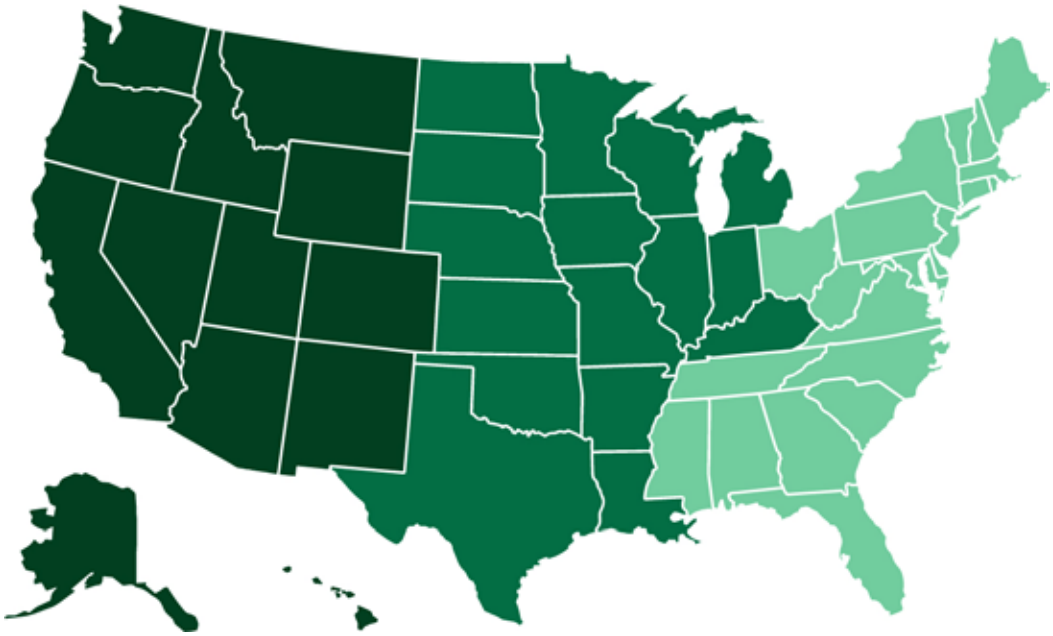

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Cassie Franklin
Donor Relations Manager

Nicole McCaslin
Regional Coordinator - East

Anne Simonsen
Regional Coordinator - Central

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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