

# UMDF connect



UNITED  
MITOCHONDRIAL  
DISEASE  
FOUNDATION.

HOPE. ENERGY. LIFE.

Quarterly publication

Volume 21, Issue 3, Summer 2017



*Bill Suzor and his daughter Lila walk in the St. Louis Energy for Life walkathon on May 20, 2017. It was Team Lila's second year participating and supporting the event.*

**UMDF**  
**education**  
Bringing the experts to your community.

With generous support from





**UNITED  
MITOCHONDRIAL  
DISEASE  
FOUNDATION®**

HOPE. ENERGY. LIFE.

**BOARD OF TRUSTEES**

- Brent Fields - Chairman
- John Kieffer - Vice-Chairman
- John A. DiCecco - Treasurer
- Bruce H. Cohen, MD- Trustee-at-Large
- Michael S. Frieberg, FACHE, CHAM
- Hooper Hardison
- Bill Kallaos, Jr., CRCP®
- Patrick Kelley
- Alicia Palladino, JD, PhD
- Sumit Parikh, MD - SMAB Chair
- Sharon Shaw - Secretary/Member Liaison
- Tyler Reimschisel, MD
- Charles A. Mohan, Jr. - Chair Emeritus

**SCIENTIFIC & MEDICAL ADVISORY BOARD**

- Sumit Parikh, MD - Chairman
- William Craigen, MD, PhD
- Marni Falk, MD
- Amy Goldstein, MD
- Andrea L. Gropman, MD, FAAP, FACMG
- Richard H. Haas, MB, BChir
- Marcia Haigis, PhD
- Amel Karaa, MD
- Carla Koehler, PhD
- Dwight Koerbel, MD, PhD
- Mark Korson, MD
- Robert McFarland, MBBS, PhD
- Gerard Vockley, MD, PhD
- Kendall Wallace, PhD
- Richard Youle, PhD

# From the Chairman

*by Brent Fields, UMDF Chairman*

**N**ext month, affected individuals and their families, researchers and clinicians will gather near Washington, DC, for the UMDF Symposium. This annual meeting is the single largest educational meeting for those impacted by mitochondrial disease and for those who care for them. It is also the meeting where new science is discussed and potential treatments and work towards cures are announced. None of this would happen without you.



Alongside those researchers, we honor those individuals who give of themselves in the areas of education, inspiration, time, talent and treasures.

It takes a lot of advance work to make our annual symposium happen. We've worked hard to provide new and exciting programming for all attendees. Whether this is going to be your first symposium or your 18th, I

hope you will come prepared to learn and find the answers that you need. I look forward to seeing you in Alexandria, Virginia at the end of June.

It is by your efforts that UMDF is able to raise the bar and dollars dedicated towards finding faster treatments and cures. One of the most important items on the agenda at Symposium is awarding grants to researchers whose projects fit the criteria required to advance all of us on the roadmap to a cure.

**UNITED MITOCHONDRIAL  
DISEASE FOUNDATION**  
8085 Saltsburg Road, Suite 201  
Pittsburgh, PA 15239  
P: 888-317-UMDF (toll-free)  
P: 412-793-8077 | F: 412-793-6477  
www.umdf.org | info@umdf.org



## 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 March 1 and June 1, 2017.

Genesis Noel Fierst  
Jennifer Rodabaugh Davis  
Laurie Austin  
Mackenzie Rae Thomas  
Noah Alan Shulman  
Deklyn Schulte

Tyler Lindstrom  
George Edward (Ted) Allen III  
Nick Dupree  
Amber Marie Hafer  
Caroline Demi Llarena-Neumann  
Anthony M. Varga

UMDF has created a brick in memory of each on UMDF's Path to a Cure. To visit each path, go to [www.umdopathtoacure.org](http://www.umdopathtoacure.org)

## Day on the Hill

Close to 200 people from 28 states have registered to participate in "Day on the Hill". Patients, families, friends and caregivers will travel to Capitol Hill on Thursday, June 29, 2017 for scheduled meetings with the offices of their Congressmen and Senators.

The meetings give patients and families and opportunity to discuss how mitochondrial disease impact them or their family. It will also give our community an opportunity to weigh in on the challenges new health care legislation may bring.

Check [www.umd.org/advocate](http://www.umd.org/advocate) in June, or watch for our eblast with details on how you can take part in Day on the Hill from your community.



## Congratulations, Christopher!

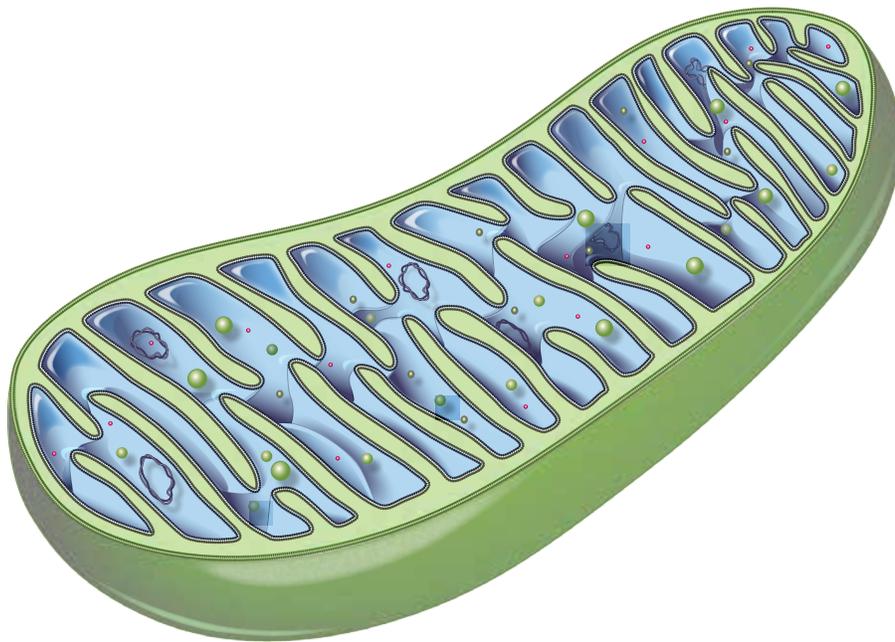
Christopher Adkins (*far right*), from Danville, IL, is a third degree black belt. He won the 2017 Illinois Special Abilities Triple Crown Championship in Taekwondo (form, sparring and combat weapons). Christopher is a multiple state champ, district champ and two-time former World Champ.

He will be competing for the District Championship in Linwood IL near Chicago.

Christopher's sister Katie will also be competing. She is 7 years old and a 1st degree black belt.

# 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](http://StealthBT.com) or follow us on social media:



@StealthBT



Stealth BioTherapeutics



Stealth  
BIOTHERAPEUTICS

Leading  
Mitochondrial  
Medicine

## UMDF Forms Advisory Council

The United Mitochondrial Disease Foundation (UMDF) has announced the formation of an Industry Advisory Council (IAC). The formation of the IAC allows the UMDF to collaborate with key members of the pharmaceutical industry to address priorities within the three key pillars of the Mitochondrial Disease Roadmap. Those priorities, announced at the UMDF International Symposium in 2016, have been identified as diagnosis, Therapeutic Development and Patient Care.

“The valuable insight and advice from members of industry will help guide us in developing faster non-invasive diagnostics and potential therapies for mitochondrial patients as well as develop a model for enhanced patient care,” said Charles A. Mohan, Jr., CEO and Executive Director of the UMDF. “We look forward to a productive and mutually beneficial working relationship.”

Mohan says the patient community will also play a key role in the IAC. “The formation of this council has the sole mission of sharing the expertise that is needed to bring about faster diagnosis, better treatments, and, ultimately a cure,” he added. “Patient participation, especially in the Mitochondrial Disease Community Registry (MDCR) is paramount to helping our community reach that goal.”

John DiCecco  
IAC Co-Chair  
UMDF Board of Trustees

Gene Kelly  
IAC Co-Chair  
Senior Director  
Commercial & Scientific Liaison  
Stealth Biotherapeutics

Marko Rosa  
Product Manager  
Tishcon Corporation

Mann Shoffner  
Pharmaceutical Relationships  
Courtagen Corporation

Matthew Klein, MD, MS, FACS  
Chief Medical Officer  
Edison Pharmaceuticals, Inc.

### IAC Committee Members

Edmund H. Doherty, Pharm D  
Associate Vice President, Product Strategy  
Reata Pharmaceuticals, Inc.

Andrew Sanford  
Operations Manager  
Solace Nutrition

Andrew T.J. Hope, Ph.D.  
Senior Director  
Business Development & Strategic Alliances  
GeneDx

Jodi Wolf, Ph.D.  
Director Patient Advocacy & Medical Science Liaison  
Santhera Pharmaceuticals

# The MOTOR Study

A study of omeveloxolone (RTA 408) in mitochondrial myopathies

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

## About the Study



Treatment: Omeveloxolone 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

### United States



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

### Europe



Pittsburgh, Pennsylvania: University of Pittsburgh  
Gerard Vockley, MD

Philadelphia, Pennsylvania: CHOP  
Marni Falk, MD

Boston, Massachusetts: Mass General  
Amel Karaa, MD

Copenhagen, Denmark: University of Copenhagen  
Karen Madsen, MD



Contact information for participating study centers can be found on the [clinicaltrials.gov](http://clinicaltrials.gov) listing



Go to [www.clinicaltrials.gov/ct2/show/NCT02255422](http://www.clinicaltrials.gov/ct2/show/NCT02255422) for more information

Version 1; September 2016



## Spotlight: Team Olivia & Liam's Crew

The 2017 Energy for Life Walkathon season is in full swing. Were you able to attend a spring walk in your area? Or, are you planning to attend a fall walk? 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 spring walk teams that is doing just that.

The Kallaos family of Chesterfield, Missouri, has been a part of the St. Louis Energy for Life Walkathon since 2014. Their daughter, Olivia, was diagnosed with Leigh's Syndrome in 2013. At first, the diagnosis was a shock to Olivia's family. Her mom, Bonnie, says they quickly learned large lessons from their sweet Olivia. "Olivia is a true hero and has been the biggest inspiration to all of her family and friends," she said.

They knew they needed to mobilize, inspired by Olivia. The family formed "Olivia and Liam's Crew" and encouraged their family, friends and supporters to get involved in all aspects of the Energy for Life Walkathon event. Bonnie said a key was to talk about Olivia to their support network. "Don't be afraid to share your story and ask for support. Remember it's the only way to raise awareness and money to ultimately find a cure," Bonnie said.

And it has made a significant impact in St. Louis!

- Olivia and Liam's Crew team members FUNDRAISE! Since 2014, the team members have raised over \$33,000 for the St. Louis walk!
- Olivia and Liam's Crew bring CORPORATE PARTNERS! They have connected to 11 different corporate partners contributing \$32,000!
- Olivia and Liam's Crew raise AWARENESS! The Kallaos family and friends host multiple events throughout the year to promote the walk and encourage their supporters to get involved.
- Olivia and Liam's Crew are a part of the walk COMMITTEE! Bonnie is a sub-committee chair and several other friends and family members have been involved on the walk planning committee.
- Olivia and Liam's Crew VOLUNTEER! From the on-site EMS to the kid's area face-painting and crazy-hair crew, you can find Kallaos family volunteers involved in many day-of activities.

The Kallaos family and Olivia and Liam's Crew prove that there are so many different ways you can be involved in your local Energy for Life Walkathons. Bonnie shares, "We thank all of our family, friends, and supporters for always being there for us continuously through this life long journey. We couldn't do it without you."

Think about what YOU can give. Is it your time, your energy, your connections, your financial support? Make a commitment to get involved to make a difference for your loved one and others needing a cure.

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





## David's Story

by Tara Maziarz

**D**avid Porter, Jr., of Flowery Branch, GA, started out life pretty typical. He is one of five children in the Porter Family and went about life without interruption until 2009. When David was a senior at Gainesville State College, things started to change.

David began experiencing dizziness and blurred vision in mid-March of 2009. After two weeks, the symptoms had not changed and he began doctor visits - general doctor, eye doctor, ENT (ear, nose and throat). No one could figure out what was causing the extreme dizziness. He visited a neurologist who ordered an MRI. His condition continued to worsen and soon David was in a wheelchair, suffering severe muscle weakness. David struggled to focus and tried to finish out his last two weeks of college. Then the call from the neurologist came - the MRI revealed lesions on him brainstem. It was a mitochondrial disease: Leigh's Disease.

With help from local physicians, he started a "mito cocktail" of supplements and saw immediate improvement. He was able to step out of the wheelchair and walk again.

"It was a very dark time when I got sick," David said. "I felt helpless. I had to rely on others, which was new to me. Getting sick taught me to completely trust the people that I love and to be humble."



Tara Maziarz and David

## **Fast forward to 2017 ...**

David is doing quite well. He was able to begin a clinical trial in 2013 for the first drug developed to treat the symptoms of mitochondrial disease. He is still on the drug today and we are awaiting FDA approval. The drug has shown promising results. It seems to have slowed the progression of the disease. With the supplements and the Trial Drug David has seemed to stabilize.

He works a few days a week at Chick-fil-A. "I LOVE having a job," David said. "It gets me out among people near my age. I really enjoy the social aspect of my job. Also, the physical activity associated with my job helps my muscles feel better." David hopes to own his very own Chick-fil-A restaurant one day.

He has lots of hobbies, and loves all sports (his favorite teams are the Braves, the Falcons, the Steelers and the Penguins). He enjoys Legos, cleaning house and volunteering for the UMDF."

## **David and the UMDF**

David has been involved with the UMDF since 2011. He has attended Symposium, local educational events, and the Energy for Life Walkathon (in several cities!)

"I love the UMDF," David said. "My life would be completely different without them. I have been able to learn to fundraise for a cure, educate myself and others about this disease and most importantly, develop friendships with people who care."

"My favorite thing about the Energy for Life Walkathon is getting together with people that I don't see often. I help at the Walk by doing the heavy lifting. I enjoy meeting new families."



## **UMDF Staff says...**

"David is like family to me. He is enthusiastic, charming, motivated and a joy to be with. I can't even imagine the UMDF without him"

**Margaret Moore**

"Where do I even begin with David Porter?! Not only is he an amazing and positive role model for the teens in the Mito community, but he is also an outstanding friend. David is one of the most caring people I know. He shows so much empathy for those around him. He's funny, imaginative, and smart. I love when he comes to visit us in Pittsburgh, whether it's driving around site seeing, getting delicious food, or playing games our time is never short on laughs."

**Tara Maziarz**

"When you meet David you realize that your world has just become a little more special. His empathy and compassion for others added with his courage and bravery, all mixed together with his wit and wonderful sense of humor make up one very special guy."

**Julie Hughes**

"David is the most charming young man I have ever met. He is very sweet and lifts your spirits within seconds of seeing him. David and I have a secret 'date' at the UMDF symposium every June – and that's all I got to say about that - and it is the highlight of my week."

**Janet Owens**

"David Porter, where shall I begin? I do not think there are enough words to adequately describe him. First to mind comes Integrity, I do not know another young man who has such a strong sense of integrity as David. His love of people and spirit of the Lord is infectious and contagious. David has very strong convictions to which I so admire in him. I always look forward to the opportunity to talk to David. What stands out to me is the fact that when David may be having a bad day, he will give you that big smile, that only David can give and will always do his very best to work through it and continue to see what the Lord has done and will continue to do in his life. I am so blessed and honored to know and love him."

**Donna Nameth**



## Through Mito Eyes: Advocacy

by Liz Kennerley

Advocacy takes many forms; it can be done on and off the Hill. I like to

look at the big picture, while others may be more interested in advocating on the individual level. The way I see it, awareness brings attention to a problem, while advocacy is more assertive with the goal of trying to solve the problem made clear through awareness. They go hand in hand. I can't advocate for something without making others aware a problem is present. For example, telling a doctor you have insomnia is awareness. Turns out; it takes an exhaustive amount of advocacy to be treated. My doctors sometimes comment about how they have little chance of changing my stubborn thoughts since I talk with US Senators and Congressmen on a regular basis. I'm also the only child of parents who are the oldest of six. Enough said.

Let me first give you an example of Awareness. Last year, I helped organize, with support from UMDF, the very first disease-specific event held at BIO International Convention which is the world's largest gathering of the biotechnology industry, along with industry-leading investor and partnering meetings. I previously interned at BIO, and have connections going back over 20 years. Given this was the first event of its kind, we had to build the program from scratch. We had three panels.

The first panel was titled, "Understanding Mitochondria and Mitochondrial Disease: A Look at Challenges and Opportunities in Research." There were some familiar names in the immediate Mitochondrial Disease community, UMDF's own Phil Yeske, PhD, as well as researchers Marni Falk, MD; Mark Tarnopolsky, MD, PhD, FRCPC; and Michio Hirano, MD. One of my many comments acknowledged that I was the only panelist without a "D" after their name. I plowed into how dysfunction of our essential mitochondria plays an essential role in a multitude of neurodegenerative conditions affecting several million people.

What would you say to a room full of researchers from every angle about mitochondrial disease? I painted the picture of daily life using my favorite analogy and a famous line from Forrest Gump, "Life is like a box of chocolates. You never know what you're going to get. My final quoted comment was used in the BIO Convention's Daily Bio Buzz Newsletter. "This isn't just about me. This is about everyone I know with Mito. All of you give me hope, and that's the most important thing in the world when dealing with a disease you know is fatal." The patient perspective is crucial in many contexts from fostering

the patient-provider relationship to drug development. This cannot be understated. It is difficult to cure something without understanding.

The other panels were, "Drug Development for Mitochondrial Disease: Examining the Current Landscape and Scientific/Regulatory Gaps" and "Harnessing the Promise of Mitochondrial Research: The Potential to Unlock Answers in Other Diseases and Aging." Both panels had speakers that overlapped with others in the program. Dr. Marni Falk took a deeper dive into the multitude of reasons why drug development in our space is the antithesis of straight forward. Much of it was a review after participating in joint FDA and NIH meetings that combined to form the Mitochondrial Disease drug development CPIM (Critical Path Innovation Meeting). Sometimes this information can be hard to hear, not only technically speaking, but personal since there is nothing I can do to simplify the complexity of this disease.

After my first panel finished, my former co-worker from BIO leaned in and said to me and Phil Yeske, "There's so much interest that we need to bring in more chairs!" I didn't leave the space the entire time, but when I turned my head at one point, it was standing room only! I couldn't help but get teary eyed. I learned after the convention ended that there was an entire group of people banging on the door to get into the Mitochondrial Disease Forum, but were told, "They're in the middle of a session! You can wait!" It's a really powerful feeling realizing something you created may have planted the seed in Merck or Pfizer's head causing them to think about something that may not only treat this beast of a disease but cure it as well. It's a feeling that isn't easy to describe.

My positive successes on Capitol Hill have a different tone. Mike Fitzpatrick, my former Congressman, did not support the 21<sup>st</sup> Century Cures Act. He fought for more funding FDA for over a year, going as far as standing in front of the House Rules Committee in the wee hours before full House vote, mentioned me by name and all I've done for rare diseases. He had quite a few other reasons. Among them being concerns over expedited medical device approval, a prominent issue among other constituents. All the while acknowledging this Act would accelerate drug development.

One comment was, "I broke with tradition and voted against the bill – not for what it included, but for what it lacked." It took me awhile to wrap my head around this. As bummed as I was, there is a lot to be said that he knew 21<sup>st</sup> Century Cures Act

would pass, yet voted nay out of protest. As well as demonstrate his support of the rare disease community and our desperately needed funding for cures. Mike was asked several times by different people, including our own Chuck Mohan, to join our Caucus, but only did so after I asked, citing as I remember, "I'm joining the Mitochondrial Disease Caucus because Elizabeth Kennerley asked me to!" He also served on the Rare Disease Caucus. His brother, Representative Brian Fitzpatrick joined that Caucus after Mike's retirement from politics.

On April 5<sup>th</sup>, I participated in my third Legislative Fly-in with BIO. Among several meetings I had that day, three stand out the best. I met with Senators Casey (Pennsylvania) and Warren (Massachusetts), as well as Rep. Lloyd Smucker from Pennsylvania's 16<sup>th</sup> District and got him to co-sponsor the OPEN ACT within five minutes! Among other positive things, this piece of legislation will allow the repurposing of drugs by pharmaceutical companies for off-label use when proven effective for other indications. For example, Bezafibrate, is currently in trial overseas as a treatment possibility. This would be tremendous since the drug has already gone through the lengthy approval process and deemed safe for its primary indication to lower cholesterol. Opposed to, creating a product from scratch, costing more compared to something that's been on the market for longer. (Like Nexium vs. Zantac, both treat reflux, but one is more expensive because it's a newer drug.)

This bill was originally part of the 21<sup>st</sup> Century Cures Act, but unfortunately, Senator Elizabeth Warren pulled it at the last minute, and advocates must start from scratch. It was reintroduced in the House during Rare Disease Week, so hopefully, momentum can get it going again... In the meeting during the BIO Legislative Fly-in surrounded by pharmaceutical "high level" people, as the only patient in the room reliant on both Obamacare and Medicaid, I shared my stories (along with some others) as well as my dependency on both Obamacare and Medicaid.

I do this during every meeting but was especially crucial with Senator Warren after she recognized me since our last encounter two years ago. She is also very influential given her outspoken nature. In addition to talking about the above, I very carefully and tactfully expressed my disappointment and frustration regarding not only her opposition to both OPEN ACT and 21<sup>st</sup> Century Cures, but the importance regarding the timely passage of health policy decisions. As I spoke, she repositioned her chair and blinked maybe eight times. I hope I made an impact, but truly won't know until this unique piece of legislation is reintroduced in the Senate. One of my group members who is in the pharmaceutical industry commented, "We're so glad you're here. You're better than 100 of us." Among a few other points, our collective concern and "ask" was the reauthorization of PDUFA. The same I expressed during Rare Disease Lobby Day and Digestive Disease National Consortium's Lobby Day.

Drumroll to about three weeks ago...and Senate HELP Committee (Health Education Labor Pensions), which both Senators Casey and Warren serve on, took the very first steps towards reauthorizing PDUFA!! Back in 1992, the biopharmaceutical industry realized Congress wasn't going to appropriate enough money for the FDA to hire adequate

staff to review drug approval applications promptly. The growing industry went to Congress and said, "We are willing to pay significant user fees for each [new drug] application we submit for review. In exchange, the FDA will use that money to hire additional reviewers." Congress agreed and passed the Prescription Drug User Fee Act (PDUFA).

Every five years since, BIO and PhRMA (Pharmaceutical Research and Manufactures of America) negotiate with FDA how high these fees will be, as well as how many new reviewers need to be hired, and any procedure changes the FDA will undertake to accelerate drug approval process. Many of these changes will be the direct result of 21<sup>st</sup> Century Cures Act becoming law on December 13<sup>th</sup>, 2016, just ten days after my 30<sup>th</sup> Birthday. If PDUFA isn't reauthorized 60 days before this legislation expires on September 30<sup>th</sup>, 2017, FDA will be forced to send lay-off notices to more than 5,000 employees.

Each and every experience from the Hill to a podium raising awareness is advocacy because of the context. Many meetings and speaking events are successful in the long run. Yes, both my former Congressman Mike Fitzpatrick and "honorary Senator" Elizabeth Warren, didn't support 21<sup>st</sup> Century Cures, but they did so because they wanted MORE FUNDING and made that clear. Yes, she pulled OPEN ACT out, but this helps focus my efforts on getting it reintroduced and getting her aboard.

In the end, 21<sup>st</sup> Century Cures Act passed in both chambers with unprecedented margins, 392-26 in the House, and 94-5 in the Senate. I was up the gallery watching both with my own eyes. It's something I won't soon forget. We also have our champions in government. Senator Casey has been nothing but supportive of the mitochondrial disease community and drug development. I have a selfie with him!! He is a true light in this sometimes-dark tunnel. We are all more than ready for more drug approvals given the mitochondrial disease statistics we know as a community, and the larger rare disease space with 1 in 10 Americans living with a rare disease, yet just 5% have FDA approved treatment. The FDA could always use more funding. Even if it is "just a fig leaf" as Senator Elizabeth Warren said as her primary reason for also being unsupportive of the 21<sup>st</sup> Century Cures Act.

Living in Washington, DC, just four blocks from the Capitol and truly on the "front lines" of every decision health policy related in this political climate isn't easy. But I have a choice. I can either frown and mope along or take an active role by making sure our voices are considered. Not many things happen quickly in the rare disease space. On average, it takes seven years for an accurate diagnosis, even though our lives are impacted daily and can change instantaneously. Sometimes it may seem like everything is bad but trust me that there are many things going on "behind the scenes." These actions will not only serve to spur scientific discovery but in turn drug development. (The running joke in Washington is, "You know you're a 'DC insider' when you speak in acronyms." Why yes. I suppose, I am.) #path2cures



We had such a great response to the last issue's personal story that I thought I would continue to invite others to contribute about how they live with their mitochondrial disease. This story is from a friend I met while traveling in Florida, **Jordan Kalick**. Jordan is a very courageous and intelligent mitochondrial warrior and I am pleased to include his story in this issue – Thank you Jordan!

*Chuck Mohan*

---

## My Story by Jordan Kalick

**H**i, I'm Jordan Kalick. I'm a 10th grade student (16 years old) from south Florida with Mitochondrial Disease. I've been attending virtual school since mid-way through my 5th grade year. I was enrolled in a brick and mortar school before then. When I was in a brick and mortar school I missed many days of school and was tardy for even more. In my 5th grade year, my elementary school offered me to try a virtual school while in school experiment where I would be taking a few 6th grade classes online in my regular classrooms.

After winter break in my 5th grade year I was unable to go back to school. I missed the first two weeks after break and could feel the anxiety building up. I decided to finish up the semester by taking online classes, and, after a great experience, decided to complete middle and high school online.

Virtual school has been a perfect fit for me. I have been able to excel in my schoolwork without feeling guilt and anxiety over missing so much school. There is no more struggle to wake up for classes or feeling lousy in class because I didn't get enough sleep. With virtual school I am able to get my work done when I wake up or if I am not feeling well I can get my work done later on in the day or even later in the week.

Many of you probably don't know how virtual school works so let me explain my setup: Florida has its own virtual school network called FLVS (Florida Virtual School) and within that it offers a virtual school through my country called PBVS (Palm Beach Virtual School). Through my state accredited courses I am able to graduate with a regular high school degree just like everyone else. I am able to take six courses (English, Math, Social Studies and Science along with two electives) that have pace charts. The pace charts tell me what I need to get done each week in order to finish the semester by either winter

break or summer vacation.

Even while taking virtual school I am able to join clubs and sports from my zoned high school. I am currently on my high school varsity bowling team for the second year. FLVS also offers clubs of their own just like any high school; I am currently in the Newspaper club (online of course), Peer Tutoring club and I am the Vice-President of our National Honor Society chapter.

Many of you might be scared of the lack of socialization Virtual School provides but like any school your social experience depends on the choices you make. In virtual school you are not socializing with peers by default. You need to put in the effort to create your own experience by joining clubs or volunteering.

Virtual school has afforded me many opportunities I wouldn't have without it. Instead of wasting my energy in school walking to classes and spending 6 hours out of the house using energy I am able to finish my work in half that time and be able to socialize on my own.

After starting virtual school, I joined a recreational basketball league that I would never have been able to before virtual school. I remember coming home from school exhausted and just wanting to rest since my body couldn't handle the day. Before a wild weekend was resting Friday to save up energy for a short trip out of the house on Saturday and then resting up all Sunday just to only miss school Monday. Now I am able to pick up my friends and spend the whole day or weekend out because my body can rest the whole week while still getting my work done.

My average work week would be waking up and whenever I feel ready I would start my work. On Sundays I pre-plan my work week so I know what I need to get done each day. This averages out to about 3 assignments or every other subject

# members

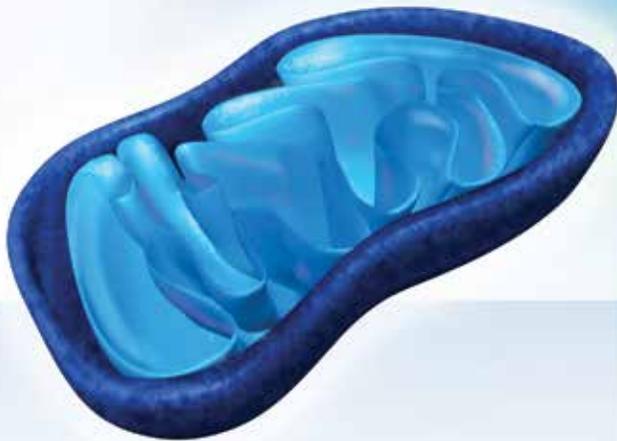
per day including a few assignments over the weekend ; some weeks may have more or less assignments. Once every 3-4 weeks, one of your assignments will make you call your teacher in and she will give you a verbal assessment before giving you a password for your chapter exam. These verbal assessments are called DBA's (discussion based assessment) and will test you on the information you learned in order to show you are ready to take the exam and to make sure you did your own work.

One of the great things about FLVS is that they really want you to excel and be the best possible you so they allow you to take an assignment up to three times (the original and two resubmissions excluding exams) to allow you to get your best possible score and go back to study more if you feel you are not prepared. Once per semester per class you will have to complete a collaboration assignment with another student; many teachers set up online sessions to complete this collaboration component which takes the stress of the students. My teachers are also in the office from 8am to 8pm and are open to phone calls or you can schedule an appointment to talk to them. You are able to email them questions by hitting an easy link on the announcement page just like you could raising your hand. My teachers also hold tutoring sessions throughout the week and your principle and guidance counselor are always there for you. Virtual School has also given me the ability to take dual-enrollment classes through my local college (FAU) which are also online without adding to much stress to myself.

I've realized now that I am socializing and excelling much more since starting virtual school. It was impossible to both attend every day of school and hang out with friends and I wasn't able to do either at that time. Now I am able to do both which really creates a sense of accomplishment that missing that much school never gave me. Instead of being a cause of anxiety and stress, school is fulfilling and virtual school allows me to do all those fun things I couldn't possibly dream about. Virtual School has also helped my health by not allowing my immune system to be compromised as much and staying in the stable environment of my home where I never need to worry about the air conditioning being broken or it being too hot to be outside. Virtual school has given me the opportunity to be my true self and enjoy my life rather than struggling through the week after a day of mundane tasks in a brick in mortar school.

I am interested in continuing to feature personal stories about living positively with mitochondrial disease. I want stories that show how you are part of the cure and not a victim of the disease. Please send your story to Chuck Mohan at [chuckm@umdf.org](mailto:chuckm@umdf.org).

**Baylor Miraca**  
Genetics Laboratories



The Baylor Miraca Genetics Laboratories (BMGL), is committed to providing quality genetic testing services relevant to patient care today. BMGL continues to develop comprehensive molecular testing and mitochondrial disease panels /testing, as well as testing for other disorders. We have specialists available to answer questions about billing and testing for your convenience.

For more information on our panels or to place an order, contact us at: [www.BMGL.com](http://www.BMGL.com) or call 1-800-411-GENE (4363) or 713-798-6555.

**solace**  
NUTRITION

*Nutritional Management  
for Mitochondrial Disease*



Visit us at [www.solacenutrition.com](http://www.solacenutrition.com)

# 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.

To find out more please  
visit our website:  
[www.santhera.com](http://www.santhera.com)



## Member Spotlight – Luca’s Legacy

**G**reek life in College is all about brother and sisterhood – students meeting new friends and forming a lifetime bond.

While some folks lose touch with one another others remain closer than ever. That feeling of camaraderie transcends into adulthood through marriage and kids. For two families from Lynchburg College’s Greek Family it has become so much more.

Kim Wilbur and her husband went to college with Nicole and Chris Florio. Nicole and Kim were in the Alpha Sigma Alpha sorority, while their husbands, Chris and Lacy, were in the Sigma Nu fraternity.

After college Nicole and Chris had a son named Luca, and he became very sick with mitochondrial disease, and tragically passed away. Kim and her husband Lacy have young children of their own and were closely following Luca’s journey. They were heartbroken watching their friends struggle. They talked with one another and decided they had to do something for the Florio family. Kim said her husband was actually the person who suggested a Golf Outing as Kim works at the Old Trail Golf Course in Crozet, VA and all of their mutual friend’s golf. It was the perfect venue and event to honor Luca.

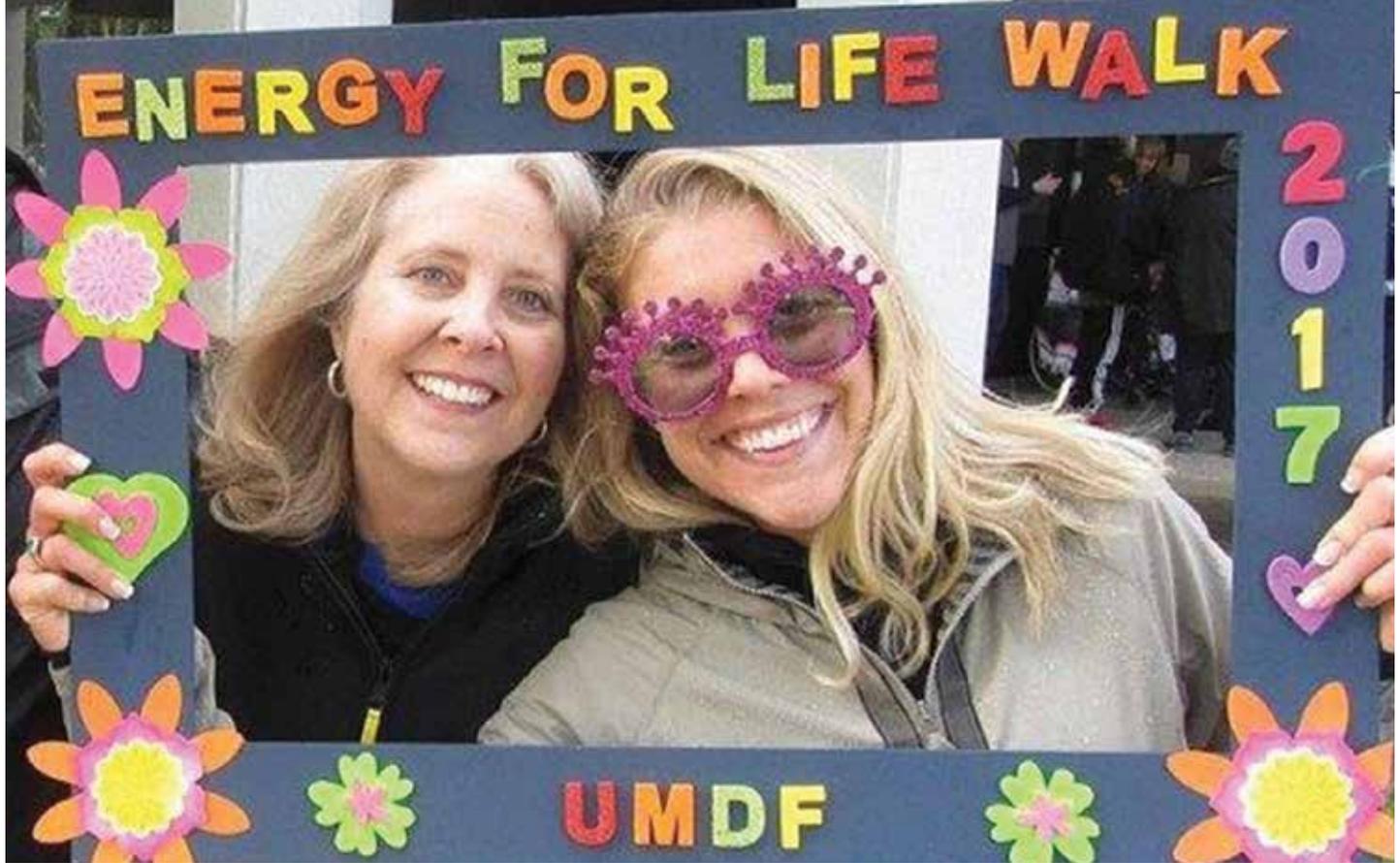
Originally Kim had planned to have the funds raised go directly to the family for medical bills. Nicole and Chris however turned them down, insisting that the funds go to the UMDF to start a research fund in Luca’s honor.



Kim went straight to working with UMDF. Their first golf outing was a huge success in September of 2016 raising over \$15,500. They were quick to call back and let the development team know that they were already planning next years’ outing... in June, less than a year away! Kim has been surprised at how many people are coming back again and also how easy finding sponsors has been. Chris and Nicole would ask many small business and more often than not they want to sponsor the event and even send volunteers!

When asked what the most difficult and the easiest parts of organizing the golf event were Kim responded, “I was shocked at how long it took golfers to sign up! Having a deadline and advertising closer to the event date seems to help with sign ups. Logistics for the event were definitely the easier part for me.” That however is no surprise there, Kim is the Director of Marketing for the Golf Course in Old Trail, which is a community of 1200-1400 homes within Crozet, VA. “A lot of charities don’t realize how great a golf tournament is for fundraising! For example, I am in charge of bringing new tournaments to the course and of those I would say about 90% are for a charity.

The most rewarding part of this golf tournament for Kim has been raising money in Luca’s honor and memory. “Seeing how much research happens, and being able to possibly change the future for other families has been a way to channel our grief into something positive to help others.”



## Raising Awareness & Hope Through Recurring Giving

Cindy Sickler and her nephew, Ryan O'Rourke, were diagnosed with Mitochondrial Disease in 2007, and it has been a life-changing event for their entire family. The family rallied in support—they wanted to get out and do what they could to be a part of the solution, knowing that every little bit counts. They have made it fun by putting together a great Energy for Life Walk team named the Future Flux Capacitors with its 1.21 gigawatts of power. Their maiden walk was in Cindy's hometown, Boston, in 2010 but has since moved to San Francisco. In just eight years, their walk team has grown to over 75 walkers. Over the years, family and friends have come to support the Walk team from all over the California Bay Area, and as far away as Boston, Florida, Colorado, Oregon, and Michigan.

Additionally, Cindy has supported UMDF through Liberty Mutual's "Give with Liberty" employee giving program for almost four years. Each year, Liberty Mutual encourages employees to sign up for the program through a recurring payroll deduction or to make a one-time

gift. Liberty Mutual, in turn, matches all donations at a rate of 50%. Cindy believes that everyone who has the opportunity to participate in employee giving should find a cause that means something to them—whether they want to give as little as \$5-10 per year or a percentage per paycheck. She is so happy to be able to support UMDF throughout the year.

Cindy gives through employee giving and of her time and energy to walk with the Future Flux Capacitors because she wants to do what she can to help find a cure. She has two daughters and knows that they both want families. Cindy wants to help find a way to leave Mitochondrial Disease behind and believes that, if we all try hard enough, it can happen. In the last five years, Cindy has given over \$5,000 through recurring and matched gifts from "Give with Liberty." Her level of giving has grown each year, and she plans to increase every area of her support toward finding a cure. Cindy gives of her time and treasures for HOPE, and we appreciate her so much!





## Family Research Funds

**J**ohn Kieffer has been a member of the UMDF Board of Trustees since May 2014. He and his wife Andrea have two grown children, Matt and Melissa, both in their 20's. Melissa was diagnosed with MELAS. John, Andrea, and Melissa were active in the Minnesota Chapter and are now active fundraisers for the Austin, Texas Energy for Life Walkathon. They decided to establish a Research Fund in honor of their daughter, Melissa in March 2016. Since that time, they have managed to raise over \$22,000 to support the mission of UMDF. Read on to learn more about the Kieffer family:

**Q: Tell us a little bit about your family.**

**A:** The Kieffers live in Austin, Texas! Melissa is 26 years old and lives with her Mom and Dad (Andrea and John). Melissa's brother Matt, and his new bride Hilary, live in Houston Texas. The entire Kieffer family have lived all around the world in exciting places like Singapore, Budapest Hungary, and many US states.

**Q: When did you learn that Melissa had mitochondrial disease?**

**A:** It was spring of 2011 when Melissa was diagnosed with MELAS at the age of 21 years old.

**Q: When did you decide to start your Family Research Fund?**

**A:** Melissa started her Family Research fund in 2015.

**Q: What made you decide the Research Fund was the right option for your family?**

**A:** Since Melissa has moved a lot, she had participated in several EFL around the country (walks in Minnesota, North Carolina, and Central Texas). As she has built a committed group of followers in many states and countries around the world and a family

research fund seemed like a great option for Melissa to engage friends and family that could not necessarily participate in her local EFL walks.

**Q: What has been the most rewarding part of the entire process?**

**A:** Melissa has built a great legacy of friends who support her in her mission to create awareness and raise money to find the cure for Mitochondrial Disease. Melissa is an incredible artist who creates unique holiday and greeting cards, sells them, and donates all of her earnings to her research page at UMDF. Many of her friends and family around the world have joined the mission to support her research fund.

**Q: What advice can you give to other families who are thinking about starting a Family Research Fund?**

**A:** Find your purpose! Find your passion! Put them to work to Find the Cure!



## Corporate Partnership Spotlight



### **Who are they?**

Stealth BioTherapeutics (Stealth) is an innovative biopharmaceutical company developing therapies to treat mitochondrial dysfunction associated with certain genetic mitochondrial diseases and common diseases of aging. Stealth's team works with patients and advocacy organizations to better understand their journey and raise awareness of the unmet needs the Company's programs seek to address. They have relationships with highly regarded academic and medical institutions, scientific thought leaders, and clinical key opinion leaders in seeking to develop the first generation of targeted therapies focusing on mitochondrial dysfunction as it presents in these rare genetic diseases and common diseases of aging.

Stealth is committed to positively impacting the lives of patients by seeking to develop novel treatment options and by offering healthcare professionals the opportunity to transform clinical practice and patient outcomes. Their compounds are being developed for mitochondrial diseases where there are no FDA-approved therapies, and where current approaches are often only palliative. Stealth's lead product candidate, elamipretide, is being investigated in three

primary mitochondrial diseases – primary mitochondrial myopathy (PMM), Barth syndrome and Leber's hereditary optic neuropathy (LHON) – as well as in heart failure, Fuchs' corneal dystrophy and dry age-related macular degeneration. Elamipretide has the potential to modify disease through mitochondrial protection – the ability to preserve and restore normal energy production in mitochondria while decreasing oxidative stress. Stealth received Fast Track designation for elamipretide for the treatment of PMM from the FDA in December 2015.

### **What do they do for UMDF?**

Stealth BioTherapeutics is one of our long-standing and most committed corporate partners, directing their support to events and areas that have the greatest positive impact on our families and individuals. This includes educational programs like the National and Regional Symposia, Grand Rounds and Family Meetings, as well as our Scientific Investments such as the Mitochondrial Disease Community Registry and Clinical Trials. Stealth most recently became a founding member of our newly formed Industry Advisory Council.



### **Stealth says...**

*UMDF's relentless commitment to serving the mitochondrial disease community has made them an incredible partner as we continue to work to develop treatments. Their spearheading of the annual UMDF national symposium provides a unique opportunity for the community to come together, share progress in science and make meaningful connections. We are proud to support their many programs that have the potential to make such a difference in the lives of patients and families.*

### **UMDF Staff says...**

*The entire team at Stealth is wonderful and easy to partner with. Their commitment to keeping patients at the center of therapeutic development is a model for other pharmaceutical companies to follow. Our patients and families are extremely excited and hopeful with the encouraging early results from studies evaluating elamipretide. UMDF is honored to have Stealth BioTherapeutics as a partner in UMDF's quest for a cure.*

July 1, 2016 – March 31, 2017

# Donor Spotlight

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

## ***Visionary Investors*** **\$100,000 and above**

William Wright Family Foundation (13)  
The J. Willard & Alice S. Marriott Foundation (5)

## ***Power Investors*** **\$50,000 - \$99,999**

Edith L Trees Charitable Trust (13)  
Reata Pharmaceuticals, Inc. (2)  
Stealth BioTherapeutics (4)

## ***Life Investors*** **\$25,000 - \$49,999**

William S. Kallaos Family Foundation (3)

## ***Energy Investors*** **\$10,000 - \$24,999**

CB&T Bank \*  
Charlotte Pipe & Foundry Company (6)  
Robert J. Bauer Family Foundation (3)  
Lee Brothers Sidekick Foundation (1)  
Mr. and Ms. Thomas Hefferon (19)  
Tishcon Corp (9)  
Hilton Worldwide, Inc. \*  
PNC Charitable Trust \*  
Mr. and Mrs. David Dobke (8)  
The Breslow Family (7)  
Mr. and Mrs. Chris Florio \*  
Mr. and Mrs. Pat Geary \*  
Mr. David Heikkinen and Dr. Ann Heikkinen (3)

Mr. Peter Kelley (11)  
Mr. Sebastiano Lopresti & Family (5)  
Mr. W. Thomas York Jr (6)  
Kelley Management Consulting (8)  
Rachael's Gift Inc. (6)  
FedEx Ground & Package Handlers (6)  
Summit Health Pharmacy (3)  
The George W. Bauer Family Foundation (2)  
RA Kirby Foundation (2)  
Gensight Biologics \*  
GVM Associates Inc. \*

## ***Hope Investors*** **\$5,000 - \$9,999**

Mr. Mark Braverman (4)  
Mrs. Molly Auth Manning (13)  
Pearce Family Foundation \*  
Solace Nutrition (9)  
Insurance Industry Charitable Foundation (IICF) \*  
Barth Syndrome Foundation Inc. \*  
Kendra Scott Design (2)  
JDM Fund (8)  
RacesOnline (9)  
Mr. and Mrs. Brent Staples (7)  
Austin Canvas & Awning (6)  
BioElectron Technology Corporation (4)  
Jay Roberts Jewelers \*  
Mr. Brett Cohen and Dr. Gwen Cohen \*  
Mr. and Mrs. Josh Albertson (7)  
Mr. Joseph Auth (14)  
Mr. Conor Davis \*

Mr. John Duffey \*  
Mr. and Mrs. Peter Geisler (4)  
Mr. Bruno Granville \*  
Mr. Walter Hawrylak \*  
Mr. and Mrs. Gordon Kidd (16)  
Mr. and Mrs. David Langer (19)  
Mr. and Mrs. Matthew Richardson (5)  
Dr. Annette St. Pierre-MacKoul MD (3)  
Dr. and Mrs. Harry Weinrauch (11)  
March of Dimes (12)  
Nikos S. Kefalidis Foundation, Inc. (11)  
The Hunt Michael Hollis Fund (5)  
Raptor Pharmaceuticals (4)  
NIEHS (4)  
VOYA (3)  
Henry Lea Hillman, Jr. Foundation \*  
The WaWa Foundation \*

**July 1, 2016 – March 31, 2017**

## **Friends**

### **\$1,000 - \$4,999**

Santhera Pharmaceuticals (3)	Mr. and Mrs. Michael Hall (4)	Mr. and Mrs. Jim Mowrer (11)
GeneDx (6)	Cincinnati Children's Hospital (13)	St. Bernadette Catholic School (15)
Bill & Melinda Gates Foundation *	Lalilab, Inc. (7)	Baxter Pharmaceutical Solutions LLC
Mr. and Mrs. Charles A. Mohan Jr. (21)	Showalter Construction Co (6)	Mechanical, Inc. (8)
Mr. and Mrs. Tim Babiarz (3)	UPMC (5)	Quten Research Institute *
Mr. John Zetterower (7)	A Tow Atlanta, Inc. (5)	Chemistry RX *
Ball Corporation (7)	Bradley Arant Boult Cummings (3)	Mr. and Mrs. Francis Mroz (8)
Mr. Hooper Hardison (8)	Footprints Case Management (1)	American Junior Golf Foundation
Mr. and Ms. Tom Hodge (5)	Horwitz (1)	Mr. William Daniel (2)
David & Paula Kirsch Family Fund (5)	Dooley Gasket and Seal, Inc. (1)	Mr. Brian Greene *
Texas Chili Company (3)	Broadway Dental Inc. (1)	Mr. and Mrs. Joel Hasfjord (9)
Dr. David Charney (9)	Landmark Builders *	Mr. William McMenemy Jr (9)
Dr. and Mrs. Andy Geer *	Dr. Jerry Butler *	Mr. and Mrs. Brett Schoenecker (14)
Mr. and Mrs. Bryan Manley (11)	Courtagen Life Sciences, Inc. (5)	Mr. and Mrs. Jerry Shuck (16)
Ruth Camp Campbell Foundation (2)	Illinois Tool Works Foundation (ITW) (11)	Mr. and Mrs. W. Wright (20)
GraceWorks Unlimited (2)	Mr. and Mrs. Neal Palmer (16)	Babiarz Law Firm P.A. (10)
The Whitehouse Family (2)	Baxter International Foundation (4)	Kohl's (8)
Ryan Companies Us Inc. (5)	Mr. Eric Dobke (9)	Grace Community Church (7)
MNG (Medical Neurogenetics) *	Mr. John Cohen (5)	PriceWaterhouse Coopers (3)
Akron Children's Hospital (5)	Mr. Lawrence Bellis *	TJ Maxx - District 4101 (5)
The Joshua & Luke Welch Charitable Fund *	Mr. and Mrs. Michael Foglia (3)	United Heritage Charity Foundation *
Mr. and Mrs. Nicholas Koch (2)	Nathalie Garner *	CBRE Charlottesville *
Pennsylvania Moose Association *	Mr. and Mrs. David Gray (14)	Rev. Mark Mast (2)
Mr. and Mrs. Glenn Jordan (3)	Prof. William Highsmith Jr. (6)	Mr. and Mrs. Eric Mullin (5)
Mr. Charles Myers *	Ms. Jodi Johnson *	Dr. Samantha Vergano *
Autarch LLC *	Mr. and Mrs. Edgar Levy (8)	TD Bank (5)
Dr. and Mrs. Bruce Cohen (17)	Mr. David Russell *	Ms. Angelina Foglia (3)
Mr. and Mrs. James Dooley (3)	Mr. and Mrs. Don Ryan (5)	Mrs. Norma Gibson (21)
Mr. and Mrs. Justin Ebert (3)	Keystone Pharmacy *	Mr. John Lore (2)
Ms. Patricia Gordon and Mr. Alan Koreneff (6)	Haggerty Family Foundation *	Judge and Mrs. McMaster (8)
	BP America Production Company *	St. Paul Episcopal School (2)
	H. Kirsch Family Foundation Fund *	Independent Health *

Mr. and Mrs. Ryan Hessler (2)	Mr. Louis Hazel (10)	Mr. and Mrs. Chris Swinn (14)
Mr. Robert Bromm (14)	Mr. and Mrs. William Heidebreder (2)	Mr. and Mrs. Douglas Szopo *
Mr. Sigmund Gjesdal *	Mr. David Hess *	Mr. Robert Tranbaugh (11)
Mr. Nick Schneider *	Mr. and Mrs. Darren Jackson *	Mr. and Mrs. Li Kan Wang (4)
Mrs. Tiffany Grove (4)	Mr. David Jacobson *	Mr. Brent Warner (2)
Mr. and Mrs. Mark Kaminsky (5)	Dr. and Mrs. Glenn Kalick DVM (16)	Dr. Terry Yochum (15)
Amazon Smile Foundation (3)	Mr. Patrick Kelley (12)	S&T Bank Charitable Foundation (13)
Ms. Holly Collins (3)	Ms. Janice Kezirian (5)	Roofing Consultants Ltd (12)
Mr. and Mrs. John Kieffer (4)	Mr. Ben Koenig (10)	Star Fuel Centers, Inc. (13)
Henrico Fraternal Order of Police - Lodge #4 *	Dr. John Linn (2)	The TJX Companies, Inc. (10)
Ms. Gail McNutt *	Mr. Joseph Maressa (6)	Gloria & Frank Pipp Family Foundation (15)
Appalachian Disc Golf Association *	Mr. Robert Marks *	Corporate Office Properties Trust (9)
Lou Kryzer *	Mrs. Lori Mason (6)	Children of Light (6)
Mr. and Mrs. Gregory Ray (2)	Mrs. Karen McAtee (2)	Enterprise Holdings Foundation (5)
Mr. Fabian Holguin *	The Mouk Family (5)	Puget Sound Kidney Centers (4)
Ms. Cynthia Sickler (5)	Mr. Brad Mountz (5)	Columbus Regional Healthcare System, Inc. (4)
Mr. and Mrs. William Kallaos Sr. (2)	Mr. David Neill (6)	Electrical Associates (4)
Ms. Paula Barnard (7)	Mr. Jason Nemeth *	Allegis Group Foundation (4)
Mr. Jack Zenthoefter *	Mr. and Mrs. Frank O'Connor (19)	Anthony Baldi & Associates (4)
Mr. and Mrs. William Diaz (2)	Mr. Martin Packouz (14)	Scharff Charitable Foundation (3)
Mr. and Mrs. Randy Adams *	Mr. and Mrs. Andrew Reardon (3)	Revenue Optimization Solutions LLC (4)
Mr. and Mrs. Tommy Baker (11)	Drs. Fred and JoAnn Reckling (3)	Auction Masters (2)
Mr. Anthony Baldi (6)	Mr. Stephen Red *	Arena Sports, Inc. (2)
Ms. Rosemarie Bastone (2)	Mr. Taylor Reid *	Coconut Point Ford *
Mr. and Mrs. Mike Bech (11)	Ms. Megan Ritchey *	Northern Virginia Plant People *
Mr. John Belk (6)	Mr. James Rodabaugh *	Rothlisberger Family Charitable Fund *
The Bell Family *	Mr. and Mrs. Jim Ruhana (5)	Living Saviour Lutheran Church *
Mr. and Mrs. Andrew Benney *	Dr. and Mrs. Mark Schnitzler PhD (10)	KPB Corporation *
Mr. Lyle Caddell *	Mr. Lyle Schutte (18)	Bernard's Salon & Day Spa *
Mr. and Mrs. Ronald Christenson (6)	Mr. Larry Sidwell *	Pediatric Dentistry of Ft. Myers *
Mr. James Collins (6)	Mr. Matthew Smith *	The Dean Team *
Mr. and Mrs. Scott Connell (12)	Mr. David Smith (3)	Ying Family Foundation *
Mr. and Mrs. Willard Dickerson (19)	Mr. Robert Snell (7)	ELCO Chevrolet *
Dr. Cynthia Dorroh and Mr. Craig Topham (3)	Ms. Patty Stageman (6)	Transwestern *
Mr. and Mrs. Joseph Ernst (6)	Mr. Peter Stathakis (4)	
Mr. and Mrs. Edward Hardison (7)	Mr. Robert Stevick (10)	



AACT members hope to see affected adults and young adults as we gather in Washington, DC for UMDF's Mitochondrial Medicine 2017! Please see below for the outline of important meetings specific to the adult and young adult community! We hope to see you there!

## Thursday, June 28, 2017

6:00-8:00 pm

11th Annual Adult Gathering

Moderators: AACT Leadership

## Friday, June 30, 2017

8:00-9:30 am

Welcome, UMDF Updates (MDCR), and Managing your Symposium Experience

Speakers: Brent Fields, Chuck Mohan, Philip Yeske, PhD and Kara Strittmatter

11:00 am-Noon

Networking Room Session: Adult

Patient Mentoring Workshop

4-6 tables with topics - each table will have a moderator/leader for the specific topic and patients will engage in informal round table discussions. (Topics TBD)

11-12 pm

Teen/Young Adult: Informal Roundtable with Mitochondrial Specialists Amy Goldstein, MD and Shana McCormack, MD.

1:30-2:30

ADULT – Ask the Mito Doc Panel

Moderator: UMDF Staff

Panel Speakers: Greg Enns, MD, Andrea Gropman, MD, Austin Larson, MD, Zarazuela Zolkipli Cunningham, MBChB, MRCP, and Sirisak Chanprasert, MD

1:30-2:30

ALL – Transition to Adulthood: What Happens after High School for Students with Disabilities? When Should Families Start Thinking about and Planning for Adult Life?

Diane Monnig, ARC of Northern VA with Anne Tucillo, Parent Perspective

2:45-3:30 pm

Transition for Young Adults – Going to School or Entering the Job Force

3:00-4:00 pm

ADULT – CPEO...Exploring Signs, Symptoms, and Treatments

Zarazuela Zolkipli Cunningham, MBChB MRCP

## Saturday, July 1, 2017

8:30 am-12:00 pm

UMDF National Updates, Roadmap to a Cure, and Clinical Trial Review

Moderator: Philip Yeske, PhD, UMDF

Science and Alliance Officer; Speakers: Reps of Open Trials

10:30-11:30 am

Adult - Palliative Care and You: Be Not Afraid

Sarah Frieber, MD

1:30-2:30 pm

ADULT - Endocrine Involvement in Mitochondrial Disease

Shana McCormack, MD

3:00-4:30 pm

Closing Session with Scientific

Summary: What's on the Horizon!

Moderator: Chuck Mohan, UMDF CEO

Day One: Amy Goldstein, MD

Day Two: Russ Saneto, DO, PhD

Day Three: Amel Karaa, MD

4:30 pm Adjourn

## AACT TEAM

Joy Krumdiack

Co-Chair - Washington

Gail Wehling

Co-Chair - Illinois

Devin Shuman

YA Coordinator

Washington

Kailey Danks - Toronto

Whit Davis - Pennsylvania

Rev. David Hamm - Maryland

Deb Fox - Arizona

Christy Koury - N. Carolina

Joy Krumdiack - Washington

Terry Livingston - Florida

David McNees - Ohio

Linda Ramsey - New York

Sharon S. Reeder - 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.*

**Miss the May 9th AACT Webinar on Clinical Trials and Studies?  
Watch the webinar anytime online at [www.umdf.org/AACT!](http://www.umdf.org/AACT!)**



# Mitochondrial Medicine 2017: Washington DC

**REGISTER NOW!**

**It's not too late!**  
**Complete the form**  
**or register online at**  
**[www.umdf.org/symposium](http://www.umdf.org/symposium)**

**Scientific Program:**  
**June 28 - July 1, 2017**

**Family Program:**  
**June 30 - July 1, 2017**

**UMDF "Day on the Hill":**  
**Thursday, June 29, 2017**

**Hilton Alexandria**  
**Mark Center**  
**5000 Seminary Road,**  
**Alexandria, VA 22311**

#### REGISTRATION FEES

- \$225 Individual Registration
- \$450 Family Registration (2 adults/same household)
- \$85 LHON Program Only
- \$65 Additional Friday Night Banquet tickets (per ticket)

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

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

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

Special Assistance  Scooter  Other \_\_\_\_\_

Special Dietary Requirements  Vegetarian  Gluten-Free  Other \_\_\_\_\_

(Contact hotel directly prior to arrival to confirm)

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

*Online registration closes Wednesday, June 21, 2017*

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

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

Card Number \_\_\_\_\_ Expiration Date \_\_\_\_\_

Name as listed on card (please print) \_\_\_\_\_

Signature \_\_\_\_\_ (invalid without signature)

#### PLEASE PRINT CLEARLY

Last Name \_\_\_\_\_ First Name \_\_\_\_\_ MI \_\_\_\_\_

Degree/Suffix \_\_\_\_\_ Specialty \_\_\_\_\_

Address \_\_\_\_\_

City \_\_\_\_\_ State/Province \_\_\_\_\_

Country \_\_\_\_\_ Zip/Postal \_\_\_\_\_

Email: \_\_\_\_\_

Phone: \_\_\_\_\_ Fax \_\_\_\_\_

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

# UMDF national

News from the national office.

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

## WHAT UMDF REGION DO YOU LIVE IN?

### **Northeast Region**

**Nicole McCaslin**

Nicole@umdf.org

[www.umdf.org/regions/northeast](http://www.umdf.org/regions/northeast)

### **Southeast Region**

**Margaret Moore**

Margaret.Moore@umdf.org

[www.umdf.org/regions/southeast](http://www.umdf.org/regions/southeast)

### **Great Lakes Region**

**Anne Simonsen**

anne.simonsen@umdf.org

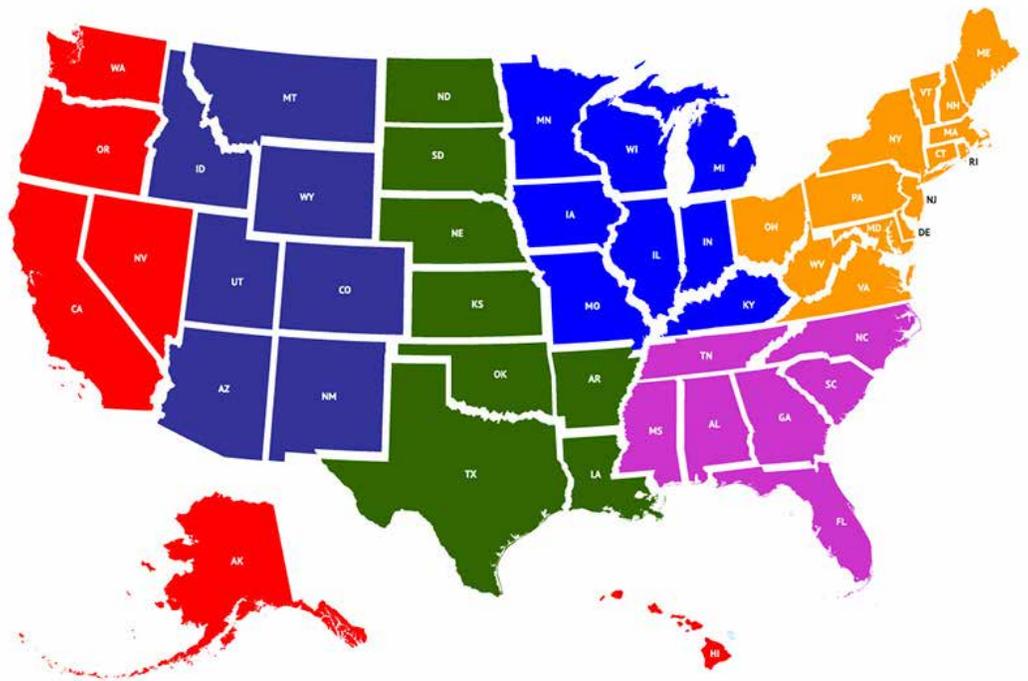
[www.umdf.org/regions/greatlakes](http://www.umdf.org/regions/greatlakes)

### **Great Lakes Region 5**

**Jessica Rios**

jessica.rios@umdf.org

[www.umdf.org/regions/central](http://www.umdf.org/regions/central)



## UNITED MITOCHONDRIAL DISEASE FOUNDATION STAFF

### Executive Staff

**Charles A. Mohan Jr.**  
CEO/Executive Director

**Janet Owens**  
Executive Administrative Asst.

**Philip Yeske, PhD**  
Science and Alliance Officer

### UMDF Symposia

**Kara Strittmatter**  
Meeting Event Director

### Finance

**Mark Campbell**  
Chief Financial Officer

**Donna Nameth**  
Data Entry Manager

**Barbara Cullaj**  
Administrative Assistant

### Communications

**Clifford Gorski**  
Director of Communications

**Jeff Gamza**  
Multimedia Coordinator

### Development & Member Services

**Beth Whitehouse**  
Director of Development

**Tania Hanscom**  
National Walk Manager

**Cassie Franklin**  
Donor Relations Manager

**Julie Hughes**  
Development Associate/  
Grants

**Tara Maziarz**  
Development Associate/  
Social Media

**Nicole McCaslin**

Regional Coordinator -  
Northeast

**Margaret Moore**

Regional Coordinator -  
Southeast

**Anne Simonsen**

Regional Coordinator -  
Great Lakes

**Jessica Rios**

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.

## Ask the Mito Doc<sup>SM</sup>

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<sup>SM</sup> at [www.umdf.org](http://www.umdf.org). Please note that information contained in Ask the Mito Doc<sup>SM</sup> 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 Great Plains Labs report shows that I have Mitochondria dysfunction. What do you think about that blood test?

**A:** The Great Plains Laboratory is not a traditional/commercial lab. Their interpretations do not align with how mitochondrial and metabolic physicians interpret metabolic test results. We do not know what to do with these results. For many patients with these abnormalities on the Great Plains testing - nothing concrete is seen on traditional tests.

**Sumit Parikh, MD**



Sumit Parikh, MD

**Q:** Inheritance question: My mother in law has Chronic Progressive External Ophthalmoplegia(CPEO). No genetic testing was ever done for her two boys (one being my husband). Could my husband be a carrier and be asymptomatic? And -my main question -could he therefore pass it on to his offspring?

**A:** CPEO is often a sporadic disorder caused by mtDNA deletions. However, there are also autosomal dominant forms of the disease. You need to determine the genetic etiology for your mother-in-law and once that is known, you can address the possible recurrence risk for your husband and subsequently your children.

**Fran D. Kendall, MD**



Fran D. Kendall, MD

**Q:** My daughter has Complex IV / Leigh's. Is it normal for her to start losing her hair? She recently had blood work and nothing showed out of her norm so doctors are stunned because they are not familiar with the illness.

**A:** No. But, hair loss can be reflective of multiple other issues to include poor nutrition and general poor health.

**Fran D. Kendall, MD**

**Q:** My currently 2.3 year old has hypotonia. We did genetic testing and received a result of: Variant of unknown significance in the MT-TV M.1647 T<C Homoplasmic. Apart from the hypotonia and balance trouble he has never had any other issue. He eats well, hasn't had any seizures etc... Could this be a new pathogenic finding? Thanks.

**A:** I am sorry to hear about your 2-year old. I can tell you my thoughts but as you could guess, there is no hard and concrete answer yes or no- seems never to be that way in mitochondrial disease. The MT-TV gene is the tRNA for valine. Valine is an amino acid that forms part of proteins made by the mitochondria. What is used to place the valine into the protein is partly due to the tRNA molecule. So, it is important. There are various regions of the tRNA molecule, some more important for functioning than others- these regions are usually called "conserved regions" meaning they are important so they stay the same between subjects and across animals. There have been only three pathological (disease causing mutations) in this molecule reported, m.1606; m.1624; m.1644. All mutations are found in heteroplasmy, meaning that there is some wild type and some mutated molecules. The m.1647 has never been reported as pathological (yet). So, could the m.1647 mutation be pathological? Since it was found to be homoplasmic, that means all the tRNA for valine molecules are identical (having the same change). Since all of our mtDNA is inherited from mother- likely you are homoplasmic for this change as well, as well as your mother and any other children you might have. Since you are likely healthy, the probability is that this change in the tRNA is neutral- meaning does not cause disease. I am not sure how conserved the m.1647 region in the molecule might be so I cannot speak to that aspect, but since it is so close to the m.1644 region likely not too conserved. So, my thought is that the change is not pathological and there is something else at work in your son. Certainly, this is only my opinion. I am sorry that it does not give you closure about your son with a diagnosis."

**Russell Saneto, DO, PhD**



Russell Saneto, DO, PhD

**Q:** 3 months ago, my sweet 2 1/2 year old daughter was diagnosed with Complex 1 Disorder, m.3688G>A (otherwise known as Leigh's). While she has lost a lot of her mobility, she is stable. She is currently on a mito cocktail of Levocarnitine, Leucovrin, Ubiquinol, Biotin and a 7.5 mg of Liquid B-Complex by Natures Answer. I am concerned that the B-Complex I am using contains PABA. I am considering switching her to a more natural product such as Buried Treasure Brand Liquid B Complete or the Tropical Oasis Liquid B Complex, but neither have been reviewed by the FDA and I don't know if these supplements are legitimate. I would appreciate your advice on a suitable solution and dosage for my little one. After reading many of the posts I have also seen creatine, alpha- lipoic acts, and magnesium 400- 800 mg and I'd like to know your thoughts about adding these supplements to the current cocktail. Thank you.

**A:** In regards to supplement or vitamin therapies for mitochondrial disease - we do not have evidence to show that any of these are effective. However, they are generally low risk and can sometimes make a difference in our patients - and so they are tried - sometimes one-at-a-time or sometimes as a combination or "cocktail." Depending on the physician you see - each one may have their own preferences. Alpha lipoic acid, creatine, CoQ10 as ubiquinol, carnitine, leucovorin and B-complex vitamins are some of the most popularly used supplements. L-Arginine is tried if there is a history or risk of strokes. The list of things that could be tried is long. The truth is that these may not help so it may not be worth continuing them endlessly. If there are no benefits noted after 3-6 months - it may be worth streamlining the vitamins given to a crucial 2 or 3. We hope that future research provides us with better guidance on which supplements may work better for specific patients.  
**Sumit Parikh, MD**

**Q:** My 6 year old daughter is diagnosed with PDHD E1. Her caregiver was just told she has mononucleosis. I know this can last for up to six weeks. What precautions should I take with her caring for my daughter? Do I need to suspend her until it has completely cleared, or for a shorter period of time? Is there extra risk for someone with mitochondrial disease when contracting mono? What should I watch out for?

**A:** Mononucleosis is spread from person to person by direct contact through saliva. As such, coming into close contact with somebody with the disease makes you at risk for contracting the infection. While good handwashing and use of gloves and mask would reduce the transmission possibility, to be completely safe, you may wish to find another caregiver until the infection has resolved.

Although some patients with a host of mitochondrial diseases do have low immunoglobulin levels making them more prone to infection, the primary risk for infection contraction in mitochondrial disease patients is the metabolic instability that occurs when they get sick.

I would recommend that you speak to your caregiver, and your local providers to determine your best option for managing the situation.

**Fran D. Kendall, MD**

**Q:** I have epilepsy, cardiac, endocrine, muscular and gastric issues. My daughter has asthma gastric issues cvs severe abdominal and regular migraines we are both the second child the other people in our family have been fairly healthy we know she has a mito disease just not sure which one. Is it possible that we can be a swinging door so to speak where I got some symptoms and she have separate symptoms but basically have the same disease?

**A:** Yes, clinical symptoms can vary between mother and daughter who have the same mutation. This is most often seen in mtDNA related diseases. There is something that is called heteroplasmy where the percentage of mutation can vary from one child to the next or from mother to her offsprings. This is less so in disease caused by nuclear DNA mutations, but still symptoms can vary in generations depending on the which chromosomes the defect lies in, e.g. X-chromosome. Other nuclear mutations are most often not seen in one generation and then the next generation unless the other parent has the same mutation- pretty rare.

I would suggest getting testing, as there are some pretty interesting medications being used in clinical trials for certain disease.

**Russell Saneto, DO, PhD**

# UMDF events

The energy providing education,  
support and research.

## Fundraisers Benefitting the UMDF

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

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

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

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

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



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

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

**March 24, 2017** BYS Yoga in Pittsburgh hosted a charity yoga night. The event raised \$275.

**April 1, 2017** The 6th annual Jackson Culley Mito What?! 5k and fun run was once again held in Millington, TN and raised over \$18,000!

**April 8, 2017** The Mito Dawgs club at the University of Georgia hosted a Spring Festival filled with games, a bounce house, and delicious food! They raised over \$1,000!

**April 22, 2017** The second Brady's Bunch 5k and Fun Run was held in Vincennes, IN.

## Upcoming Events

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

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

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

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

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

<https://www.crowdrise.com/>

*UnitedMitochondrialDiseaseFoundation1*



EFL: St. Louis

## Upcoming EFL Walkathons

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](http://www.energyforlifewalk.org/sanfrancisco)

Houston

[www.energyforlifewalk.org/houston](http://www.energyforlifewalk.org/houston)

Tampa Bay

[www.energyforlifewalk.org/tampabay](http://www.energyforlifewalk.org/tampabay)

Dallas/Fort Worth

[www.energyforlifewalk.org/dallasforthworth](http://www.energyforlifewalk.org/dallasforthworth)

Nashville

[www.energyforlifewalk.org/nashville](http://www.energyforlifewalk.org/nashville)

Atlanta

[www.energyforlifewalk.org/atlanta](http://www.energyforlifewalk.org/atlanta)

New England

[www.energyforlifewalk.org/newengland](http://www.energyforlifewalk.org/newengland)

St. Louis

[www.energyforlifewalk.org/stlouis](http://www.energyforlifewalk.org/stlouis)

Cincinnati

[www.energyforlifewalk.org/cincinnati](http://www.energyforlifewalk.org/cincinnati)

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, June 10 – Pittsburgh

[www.energyforlifewalk.org/pittsburgh](http://www.energyforlifewalk.org/pittsburgh)

Saturday, August 19 - Minnesota

[www.energyforlifewalk.org/minnesota](http://www.energyforlifewalk.org/minnesota)

Saturday, September 9 - Indianapolis

[www.energyforlifewalk.org/indianapolis](http://www.energyforlifewalk.org/indianapolis)

Saturday, September 16 - West. New York

[www.energyforlifewalk.org/westernnewyork](http://www.energyforlifewalk.org/westernnewyork)

Saturday, September 16 - Kansas City

[www.energyforlifewalk.org/kansascity](http://www.energyforlifewalk.org/kansascity)

Saturday, September 16 - Detroit

[www.energyforlifewalk.org/detroit](http://www.energyforlifewalk.org/detroit)

Sunday, September 17 - Chicago

[www.energyforlifewalk.org/chicago](http://www.energyforlifewalk.org/chicago)

Saturday, September 23 - Delaware Valley

[www.energyforlifewalk.org/delval](http://www.energyforlifewalk.org/delval)

Saturday, September 23 - S Wisconsin

[www.energyforlifewalk.org/southernwisconsin](http://www.energyforlifewalk.org/southernwisconsin)

Saturday, September 30 - Central Texas

[www.energyforlifewalk.org/centraltexas](http://www.energyforlifewalk.org/centraltexas)

Sunday, October 1 - Seattle

[www.energyforlifewalk.org/seattle](http://www.energyforlifewalk.org/seattle)

Saturday, October 14 - Charlotte

[www.energyforlifewalk.org/charlotte](http://www.energyforlifewalk.org/charlotte)

Saturday, November 18 - Southwest Florida

[www.energyforlifewalk.org/southwestflorida](http://www.energyforlifewalk.org/southwestflorida)

## Upcoming Symposia

The UMDF is proud to enhance our educational programming through regional symposia. Below is a list of the locations for 2017 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**

### UMDF Mitochondrial Medicine Central Regional Symposium

University of Texas Health Science Center  
San Antonio, TX

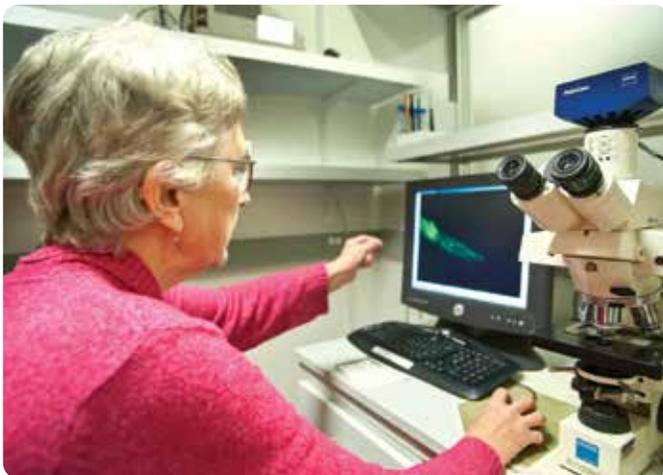
Course Chair: Sidney W. Atkinson, MD

**Friday, November 3 &  
Saturday, November 4, 2017**

For details on all UMDF Symposia visit  
[www.umdff.org/symposium](http://www.umdff.org/symposium).



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



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



**Seattle Children's**<sup>®</sup>  
HOSPITAL • RESEARCH • FOUNDATION

Research Institute