

# UMDF connect



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## Jack Black Makes Surprise Visit to Symposium



Jack Black talks with Caroline Cheung-Yiu and her children at the symposium. See page 9 for the story.

## UMDF Creates Vanguard Award



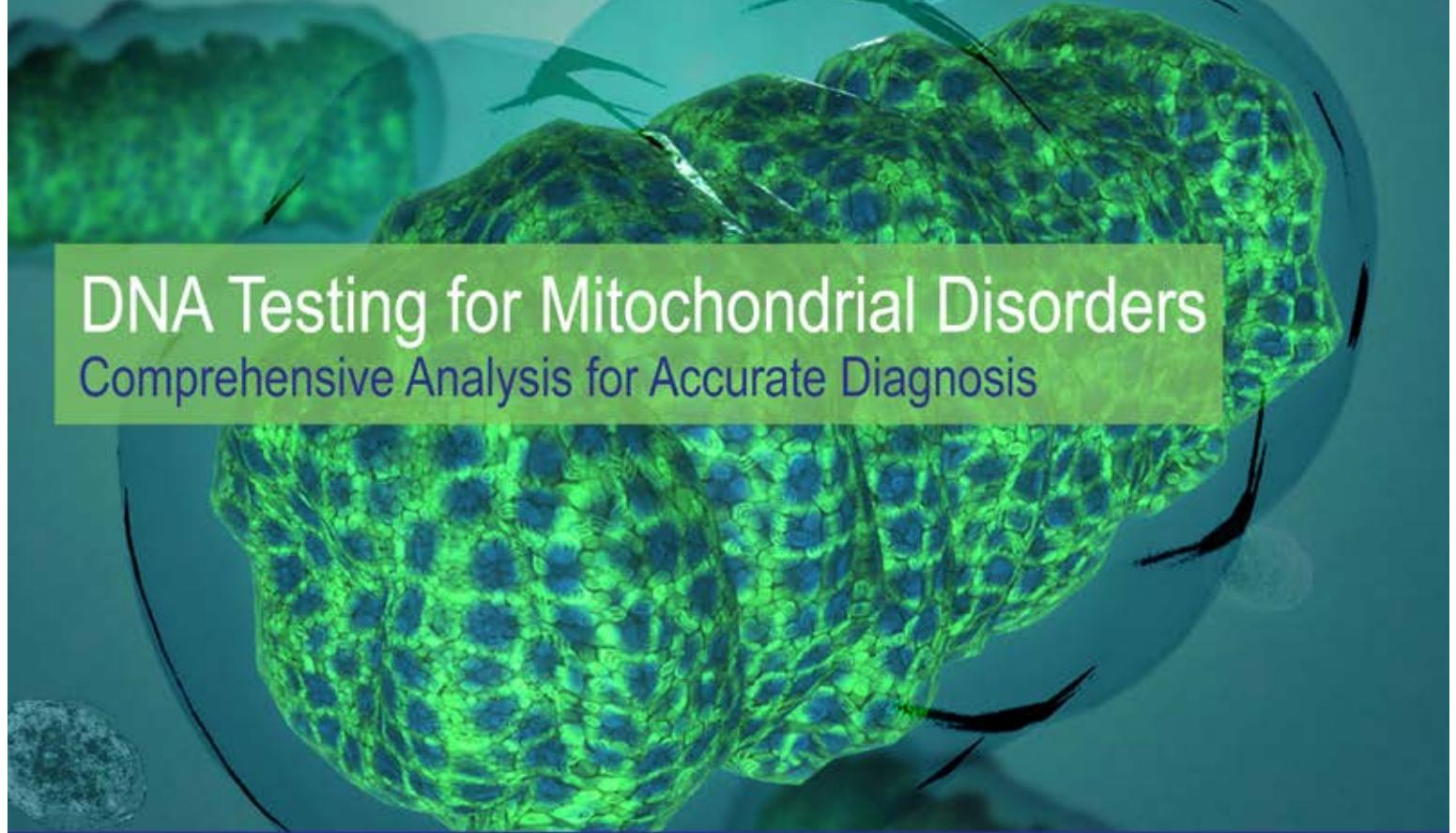
Chuck Mohan, UMDF CEO/ED; Dr. Salvatore DiMauro, recipient of inaugural Vanguard Award; Sharon Shaw Reeder, UMDF Vice Chairman of the Board of Trustees and W. Dan Wright, UMDF Chairman of the Board of Trustees. See page 6 for the story.



Stephen Holmes poses with Jack Black during a teen session at symposium.

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# From the Chairman

Zoom - Zoom! We are picking up the pace along our path towards treatments and cures for mitochondrial disease. After listening to the presentations of the scientists, clinicians and pharmacy representatives at the UMDF symposium in June, I am even more excited than ever about the progress we are making. I am excited because during the scientific and poster sessions, 22 different potential therapies were discussed. Some, of course, have better potential than others and many of you foodies will like this - one even uses a substance that occurs naturally in dark chocolate! Ten years ago, 22 different potential treatments were a dream. We are all incredibly excited now that this dream is starting to become a reality.

One of the compelling points of interest at the symposium this year was what we call a "Decade of Difference." Think back to 2003. At that time, there were only three clinical trials related to patients with mitochondrial disorders. Today, there are many more. A decade ago, researchers were only starting to connect mitochondrial dysfunction with other more common illnesses like Parkinson's, Alzheimer's, diabetes, and Autism Spectrum Disorders. Today, those links have clearly been established. In 2003, gene sequencing was just a dream. Today, gene sequencing is a reality and a must, identifying areas where mutations occur and targeting potential treatments. It has been an amazing decade. We should all be excited about these advances as we accelerate down the path towards a cure.



With the progress being made on the scientific front, UMDF is moving forward as well. We anticipate there will be a greater call for patients to participate in current and new clinical trials for many of the emerging potential therapies. Remember, successful clinical trials equals treatments. We see the need for patient registries that can identify those who

are interested in participating in clinical trials. And, we believe that there will be a stronger call for support for the grant requests for all of these exciting developments that are ahead down our path.

With these advances in mind, the UMDF, with funding from the Marriott Foundation and others, has been able to create the position of Science and Alliance Officer. Phil Yeske, Ph.D., joined the UMDF staff on July 1, 2013, to fulfill this roll. His main job is to facilitate the development and approval of treatments and cures within the mitochondrial community. Dr. Yeske will also help us understand the science behind all of these wonderful developments and prepare us for the next decade. Dr. Yeske will offer information in the newsletters on how all of these developments affect our community. So our goal is to pick up the pace until we are going so fast that we are RACING along this path. I predict it will not take another ten years to bring treatments, but with everyone's help, just like a good pit crew, we will see treatments and progress towards cures very shortly.

Zoom - Zoom,

W. Dan Wright, UMDF Chairman

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## Decade of Difference

**Read UMDF's  
Decade of Difference  
on the back page  
of this newsletter!**

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**UMDF**  
**symposium**

Providing education and networking.

## **UMDF Awards Nearly Half Million Dollars in Research Grants**

The United Mitochondrial Disease Foundation (UMDF) awarded more than \$490,000 dollars to six researchers whose projects may lead to a cure for mitochondrial disease or better treatments for those who battle it. The research grant awards were presented at the UMDF's annual symposium, "Mitochondrial Medicine 2013: Newport Beach" which was held at the Newport Beach Marriott in Newport Beach, California.

Charles A. Mohan, Jr., UMDF CEO/Executive Director, said, "We are funding projects that our Grants Review Committee believes will move us all closer to what we need now – a cure for mitochondrial disease. None of this would be possible without the work of the thousands of volunteers who participate in our Energy for Life Walkathons, golf tournaments, galas, car washes and bake sales. Every penny counts on the path towards a cure."

UMDF announced that Amel Karaa, M.D., with Harvard Medical School and Massachusetts General Hospital in Boston, Massachusetts, was the winner of the 2013 Clinical Fellowship Training Award. The clinical fellowship training award is designed to support the training of physician scientists who plan to practice clinical management of patients with mitochondrial disorders and to conduct patient-oriented research in the field of Mitochondrial Medicine. Karaa will receive \$70,000 for one year for the clinical fellowship and will focus on "Hypogonadotropic hypogonadism in mitochondrial disease: prevalence, phenotypic heterogeneity and hormonal spectrum variations in a tertiary hospital cohort."

A summary of the research grant recipients, a brief description of their projects and the award amounts follow.

### **Chairman's Award**

**James Stewart, Ph.D., Max Planck Institute for the Biology of Ageing, Cologne, Germany - \$90,000 for two years**

"Using mtDNA mutator mouse-derived lineages to generate mouse models of human mitochondrial diseases."

*By working with mice that are prone to mitochondrial mutations, Dr. Stewart will develop new genetic models of human disease. Once established, these mouse models can be used for the development of new drug therapies.*

**Alberto Sanz-Monterro, Ph.D., University of Tampere, Tampere, Finland - \$100,000 for two years**

"A Genome-wide RNAi Screening to Identify New Genes Involved in Mitochondrial Diseases."

*Dr. Sanz-Monterro will use a well-understood fruit-fly model to discover previously unknown genetic defects that can cause mitochondrial disease. Many mitochondrial disease patients have not had a specific genetic mutation linked with their disease, and this research will help to fill that gap.*



Amel Karaa, M.D.



James Stewart, Ph.D.



Alberto Sanz-Monterro, Ph.D.



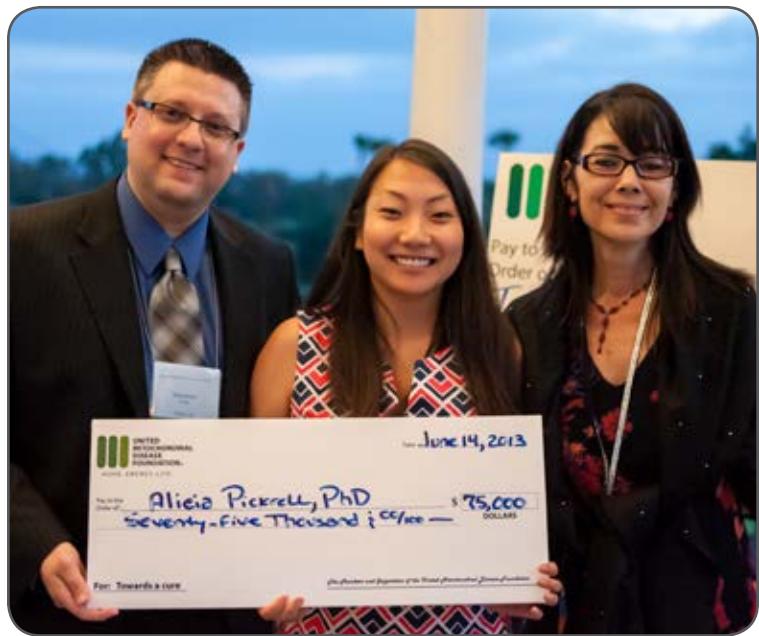
Stacey Mangni; Sharon Shaw Reeder, UMDF Vice Chairman of the Board of Trustees;  
Nancy Loberg and Rajesh Ambasudhan, Ph.D.

**Rajesh Ambasudhan, Ph.D., Sanford-Burnham Medical Research Institute, La Jolla, California - \$84,000 for two years**  
**"A Human Reprogrammed-Cell Model of MELAS."**

Dr. Ambasudhan will obtain skin cells from MELAS patients and reprogram them as nerve cells to be grown in culture. This "disease-in-a-dish" model will be used to gain insights into mitochondrial dysfunction in MELAS and other mitochondrial diseases.



Devin Shuman, UMDF Youth Ambassador; Natalie Niemi, Ph.D. and  
Sharon Shaw Reeder, UMDF Vice Chairman of the Board of Trustees.



Sebastien Cotte, UMDF Ambassador; Alicia Pickrell, Ph.D. and  
Sharon Shaw Reeder, UMDF Vice Chairman of the Board of Trustees.

**Natalie Niemi, Ph.D., University of Wisconsin, Madison, Wisconsin - \$75,000 for two years**

"Utilizing dynamically regulated phosphorylation as a means to modulate mitochondrial metabolism."

Dr. Niemi will study mechanisms that activate enzymes in the mitochondria, with the goal of understanding how this regulation is impaired in mitochondrial disease. This could lead to new therapeutic options for mitochondrial disease patients.

**Alicia Pickrell, Ph.D., National Institute of Neurological Disorders and Stroke, Bethesda, Maryland - \$75,000 for two years**

"Therapy for mitochondrial diseases: an investigation into the potential to stimulate Parkin-mediated mitophagy."

Dr. Pickrell is studying the effects of the drug Rapamycin on the removal of abnormal mitochondria from cells in mice. This FDA-approved drug has the potential to selectively eliminate dysfunctional mitochondria in humans, helping to restore normal energy metabolism in mitochondrial disease patients.

# **UMDF Announces Two Winners of the Stanley A. Davis Leadership Award**



Sharon Shaw Reeder, UMDF Vice Chairman of the Board of Trustees; Cherie Lawson and W. Dan Wright, UMDF Chairman of the Board of Trustees.

This year, the Stanley A. Davis Leadership Award was co-presented to Cherie Lawson of Chicago, Illinois, and Jennifer Schwartzott of North Tonawanda, New York. The Stanley A. Davis Leadership Award is the most prestigious UMDF National Award honoring a UMDF Leader who best represents the selfless dedication and commitment to the UMDF Mission as that of its namesake.

Lawson's determination comes from the progress and fight shown by her son, Andrew, now 20, who has battled mitochondrial disease since he was two months old. The Lawson family struggled with a variety of illnesses and medical ailments through Andrew's entire life until they finally received a diagnosis of Leigh's Disease in 2005, when they were able to treat his pain and nausea more effectively.

Lawson is a volunteer leader for the Chicago Chapter of the UMDF and the Co-chair for the Energy for Life Walk in Chicago. Ten years ago, the UMDF Chicago Chapter began offering regular support group meetings as well as helping with numerous fundraising and educational activities; it is because of volunteers like Lawson that these activities are still consistently offered in the greater Chicago area today. Lawson has served as a board officer, support group leader and has taken calls and e-mails from affected patients/families. Andrew has always been an inspiration for her dedication to the UMDF mission, but we also know her unwavering love and respect for all of our mitochondrial patients and families keeps her pushing forward.

Lawson works full-time and definitely has very little spare time beyond her duties as a "super mom" to Andrew. While her life can be hectic, she lets the UMDF know that her door is always open for input or help – and we do take her up on that offer. She does know her limitations, but she has never said no. Finally, this lovely woman brings a smile to the faces of many – she is warm, caring and a lot of fun to be around.

Schwartzott, who also received a Stanley A. Davis Leadership Award, was diagnosed with Mitochondrial Myopathy in 1990, but she does not let her disease hold her back from living life to the fullest. She is a volunteer leader for the UMDF New York Chapter, leads the support group meetings in the Western

New York area and helped start the Western New York Mito Group in 2010. She also led the Energy for Life Walkathon in Western New York for the past four years. She has found that being a part of a support group is good therapy for her, and she enjoys sharing her knowledge and experiences of what she has learned about living with mitochondrial disease. Schwartzott said, "I feel that maybe there is a purpose for me having this awful disease: helping others live with it."

Schwartzott joined the UMDF's Adult Advisory Council Team (AACT) in June of 2012. Sharon Shaw Reeder, Chairman of the AACT, invited Schwartzott to be a member of the AACT at the 2012 UMDF symposium. Reeder said, "I heard of her reputation from the UMDF staff and her dedication with her chapter in New York. Jennifer also mentioned that she was able to identify more than 300 adult mitochondrial patients through her networking in the New York area." In the fall of 2012, Schwartzott took awareness to a new level by getting UMDF bumper stickers made and placed on racecars. She has been an Auto Racing Official at professional and amateur road racing events for 23 years.

Mitochondrial disease has changed the vision of how Schwartzott thought her life would be. She has a Master's Degree in Multidisciplinary Studies that she worked very hard to obtain while she was sick, but now she cannot use it because the disease has impacted her ability to work. She has had to give up things she once enjoyed doing, such as skiing, dancing and reading. However, she doesn't dwell on the negative things that have happened in her life as a result of the disease. "I have just had to redefine what accomplishment is and find new hobbies and activities that make me happy. I have learned that you don't have to give up everything even though things might be difficult. There are days when I can't do anything, but I know that I can rest and eventually get back to doing things I love," she said. One person that can make her smile on the worst of days is her granddaughter, Amara.



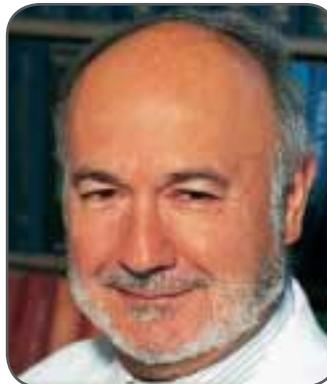
Sharon Shaw Reeder, UMDF Vice Chairman of the Board of Trustees; Jennifer Schwartzott and W. Dan Wright, UMDF Chairman of the Board of Trustees.

# UMDF Recognizes First Vanguard Awardee

The United Mitochondrial Disease Foundation (UMDF) is excited to announce the creation of a new Vanguard Award that will be presented annually to a scientist or physician in honor of his or her lifetime contributions to the field of mitochondrial medicine or research. The recipient will be one who is recognized for leadership and commitment towards a cure for mitochondrial diseases, whether through research or medical treatment, and whose inspiration has resulted in bringing new scientific or medical experts into the field.

Nominations were solicited from the UMDF's Scientific and Medical Advisory Board and the UMDF's Board of Trustees. Because they felt he exemplified the criteria for the award, they were unanimous in selecting Salvatore DiMauro, M.D., as the first recipient of the 2013 UMDF Vanguard Award.

Throughout his career, Dr. DiMauro has kept a focused interest on inborn errors of energy metabolism and recognized unusual cases through clinical observation using both biochemical and molecular approaches to define disease entities. He started as an "enzyme hunter" (and in 1973 discovered CPT deficiency, the first fatty acid oxidation defect in humans) and became interested in the molecular



Salvatore DiMauro, M.D.

basis of inborn errors of metabolism, especially mitochondrial encephalomyopathies.

Although Dr. DiMauro started as a myologist, his studies of mitochondrial diseases have taken him back to the brain. His interest in the relationship between mitochondrial disorders and mental retardation has been recognized by the National Institute for Child Health and Human Development (NICHD), which supports a program entitled, "Mitochondrial Encephalomyopathies and Mental Retardation." Dr. DiMauro was the Director of the H. Houston Merritt Clinical Research Center from 1999 to 2009. A well respected mitochondrial disease expert, Dr. DiMauro has written numerous textbooks, articles and research papers.

Dr. DiMauro was born in Verona, Italy, and he graduated in Medicine in 1963 from the University of Padua where he completed his residency in Neurology in 1966. In 1968, he obtained a postdoctoral fellowship to do clinical research in the Department of Neurology at the University of Pennsylvania. In 1974, he became Associate Professor at the Columbia University Medical Center (CUMC). In 1991, he was named Lucy G. Moses Professor of Neurology. He continues his work at Columbia University in New York, NY.

## UMDF Announces Recipient of First Energy Award

The United Mitochondrial Disease Foundation (UMDF) is proud to recognize Jennifer Schulz as the inaugural Energy Award recipient. The Energy Award was designed to recognize an individual who embodies the spirit of the UMDF and its Mission: to promote research and education for the diagnosis, treatment and cure of mitochondrial disorders and provide support to those affected. A tireless and selfless advocate for mitochondrial disease, Schulz is a main driving force for mitochondrial disease awareness and action in Charlotte, North Carolina. The mother of three became active in the UMDF shortly after her son Finn was diagnosed with a mitochondrial disease, pyruvate dehydrogenase complex deficiency, in 2009.

After struggling for years to find answers to Finn's myriad of medical conditions, the Schulz's realized how little awareness there was for mitochondrial disease. Since then, she has planned patient and family meetings, offering support to others with mitochondrial disease. Schulz works diligently to raise money and spread knowledge and was instrumental in bringing both Grand Rounds and a regional symposium to

Charlotte. She shares stories and events via an active Facebook presence. She looks for opportunities to share her positive experiences and best practices regarding mitochondrial treatment and educating physicians, nurses and therapists in Charlotte about the disease.



Jennifer Schulz is pictured above with her husband, Andrew and three children: Cadence, Finn and Liam.

For three years, Schulz served as Co-chair for the Energy for Life Walkathon in Charlotte, organizing logistical needs, such as registration, merchandise, route management and even ice delivery. Despite surgery shortly before the 2012 Energy for Life Walkathon, her great spirit was not diminished, as she managed the entire event while still in a cast. Thanks to her efforts, the Charlotte Energy for Life Walkathon has grown to be the largest in the nation.

Schulz has created a great sense of community and camaraderie in the Charlotte area, as evidenced by the overwhelming number of award nominations submitted in her name.

Being active with the UMDF gives her a sense of hope: "I am not only hopefully helping my Finny but raising awareness for lots of others with mitochondrial disease."

# **UMDF Announces the 2013 Heartstrings Award Winner**

Zerin Wolfgramm of Salt Lake City, Utah, is the recipient of this year's Heartstrings Award. The Heartstrings Award was created to recognize the efforts of an individual under the age of 18 who has donated or raised funds for the UMDF, enabling the UMDF to continue its mission. The winner is chosen based on the criteria of age, time invested, talents demonstrated, effectiveness and generosity.

Eleven-year-old Wolfgramm holds numerous Utah Boys swimming records and swims in honor of 10-year-old Lily Finnigan, who was diagnosed with Mitochondria Myopathy at 16 months. In April, Wolfgramm dedicated seven Utah swimming titles to Lily, who is the daughter of his swimming head coach, Matt Finnigan. "Every time I swam, I thought of Lily," Wolfgramm said. "Nothing seems to get her down, so why should I be?"

Wolfgramm's mother said that she and her husband have tried to teach their children to think about others who

have disabilities. The Wolfgramm family recently raised money for the UMDF by running around their neighborhood to raise awareness for mitochondrial disease. Wolfgramm, who is 11, ran 11 miles. Wolfgramm's father, age 50, ran 50 miles. Wolfgramm's siblings also ran the number of miles that are equal to their ages. Wolfgramm also participated in two Energy for Life Walkathons in Salt Lake City to raise funds for the UMDF in Lily's name.

Their outreach swim team, Race Swami, was honored in 2011 by the Utah Legislature while working to pass HJR3, designating September 5-11 as an official Mitochondrial Disease Awareness Week in Utah. The legislature "urges the citizens of Utah to observe the week with appropriate activities and programs."



Zerin Wolfgramm

# **UMDF Presents the 2013 LEAP Award Winner**

This year's LEAP award was presented to Cynthia Steward of Chicago, Illinois. The LEAP Award was designed to recognize an individual, 14 years or older, living positively with mitochondrial disease, highlighting the person's accomplishments and volunteer service. The individual should demonstrate a positive attitude, hope for a brighter future, and an enthusiasm that inspires others.

Steward is a mother, wife, grandmother and sister. She is also working a full-time job and studying to earn a Master's Degree in Accounting, despite fighting through the mitochondrial disease that has ravaged both her and her family. Her granddaughter, Bailey, passed from mitochondrial disease in 2011, and she began an annual fundraiser in honor of Bailey the following year.

Steward's daughter, Dawn, is also affected by Leber's Hereditary Optic Neuropathy (LHON) and Leigh's-like syndrome. Dawn suffers a visual impairment, so Steward

read Dawn's college textbooks to her, eventually inspiring her to return to school and work for her degree. They have since received their Bachelors in Business Administration in Accounting from Devry University and are currently in the Master's Program at Keller Graduate School of Management of Devry University. Both women have maintained a 4.0 grade point average.



Cynthia Steward

Steward is active in helping with the UMDF's Energy for Life Walkathon in Chicago, joining the Walk Committee for the 2012 and 2013 walkathons. She was pleasantly surprised at the generosity of people as she asked for donations and sponsors. She is also very active at Chicago Chapter Support Meetings, sharing coping skills and information on treatments and supplements.

She has also found comfort in being involved in a local church and is now a preschool teacher for the Sunday school program. A warm and loving person, her heart is open to help others and encourages all those around her to keep moving forward.

# Jack Black Attends Symposium

Those attending the UMDF symposium, Mitochondrial Medicine: 2013 in Newport Beach, California, were treated to a surprise. Actor/Musician Jack Black paid symposium goers a visit to show his support for the affected community and their families.

Black, who along with Kyle Gass make up Tenacious D, began his visit by spending time with those who attended the UMDF's teen sessions. During his visit, he was able to meet and talk with the teens. Each teen had the opportunity to interact one-on-one with Black. During that time, he autographed pictures, joked, sang and even made short video



Chuck Mohan, CEO/ED, gets his guitar autographed by Jack Black.

clips for them to share with their friends. Following the visit to a teen session, Black participated in a meet and greet opportunity with the rest of the family and scientific and medical attendees.

Black's involvement with the UMDF began in 2001. He continues to help raise awareness through the creation and production of public service announcements for the UMDF and for the Energy for Life Walkathons. Black has also generously provided more than \$300,000 in donations through appearances and fundraisers conducted by Tenacious D.

## UMDF Member Gains Knowledge, New Friends and Hope at Symposium

by Sara Brown, UMDF Member from Winnsboro, TX

I had the privilege of attending the UMDF symposium in Newport Beach, California, in June. It was an amazing experience. Before going to symposium, I thought I was pretty well educated on mitochondrial disease but after attending, I know so much more. There are so many advancements happening constantly in the field of medicine and mitochondrial disease.

I enjoyed a session about potential therapies and clinical trials. I was able to hear about research that is going on that may one day cure my daughter, which gave me hope. It was also wonderful to see that the money being raised for research is being used to its full extent and that made me want to do more to fundraise.

We have only been dealing with our mito diagnosis for two years, so I got a lot of information in the "What to Expect in the Beginning" session. I learned about the Mitochondrial Genome and how just a small change in that cell can cause such a devastating disease. The presenters

really helped me understand mitochondrial disease better than ever.

The most informational session of the symposium, in my opinion, was "The Doctor is In." It was so nice to be able to sit down one-on-one with some of the best mito docs in the U.S. and talk specifically about my daughter's history and not have to pay a co-pay. It doesn't get much better than that!



Sara Brown (far left) poses with friends at the symposium: Shawna McElveen, Luann Norris, Angela Winters, Tammy Reyna and Nicole DeJean.

and hope to be able to attend many more symposiums in my future.

I will always remember my trip to Newport Beach

# ***Therapies Take Center Stage at Newport Beach Research Symposium***

**by Steven G. Bassett, Ph.D., Seton Hill University**

Scientists from around the world gathered in California for the 15th annual UMDF research symposium: "Mitochondrial Therapeutics – From Bench to Bedside." Featured topics included therapies, imaging and diagnosis and genetics of mitochondrial disease. Different experimental approaches employed by researchers included cell culture, animal models and direct treatment of mitochondrial disease patients. Dr. Gregory Enns of Stanford University Medical Center was the Course Chair and began the sessions with an overview of the current status of therapy. Other researchers shared their findings on a variety of agents that show promise in making significant inroads into the treatment of many mitochondrial diseases. At least 22 different therapies were referenced during both the platform presentations and the poster sessions. Not all of these treatments are new but some of the older ones remain in use as newer ones come on line. They ranged from drugs that can be purchased over the counter, such as idebenone, arginine, citrulline and CoQ10, to ones that are still available only in clinical trials, such as EPI-743 and Bendavia. Another agent, epicatechin, occurs naturally in dark chocolate, but may only be present in low levels in processed chocolate products.

The following summarizes some of the research presented at the symposium.

Dr. Iain Hargreaves of the Neurometabolic Unit, University London College Hospitals, discussed the status of Coenzyme Q10 (CoQ10) therapy. While this easily obtained supplement has been demonstrated to have some clinical benefit in patients, he focused on a subgroup of mitochondrial disease patients who respond more strongly and may have an underlying CoQ10 deficiency. Dr. Patrick Yu-Wai-Man from Newcastle University in the U.K. presented a poster summarizing work in Dr. Patrick Chinnery's lab on the treatment of Leber's Hereditary Optic Neuropathy (LHON) with idebenone, a drug chemically related to CoQ10. They established a beneficial effect of the drug in preserving vision if used early in the course of the disease. Patient follow-up an average of 30 months later revealed that no further visual deterioration had occurred.

Dr. Carla Giordano of the University of Rome was a 2012 UMDF research grant recipient. She is interested in the effects of phytoestrogens, naturally occurring plant compounds, on a cell model of LHON. At the symposium, she reported that cells treated with a combination of phytoestrogens displayed a significantly decreased rate of cell death. She hopes that this treatment will ultimately be useful in preventing the onset of LHON in unaffected carriers of the mutation that causes the disease.

Two other naturally occurring compounds, arginine and citrulline, were the subject of research presented by Dr. Fernando Scaglia of Baylor College of Medicine. His data demonstrated that patients with the severe mitochondrial disease MELAS had lower than normal plasma levels of these two amino acids and that when patients were supplemented with either one, their symptoms improved. Of the two, citrulline was the more effective.

And then there's dark chocolate, which has been in the news because regular consumption is associated with a decreased risk of heart disease and stroke. Flavanols are compounds occurring in chocolate that appear to be at least partially responsible for these beneficial effects. Dr. George Schreiner of Cardero Therapeutics in California presented data on the chocolate-derived flavanol known as epicatechin. This compound was shown to increase the number of mitochondria in muscle cells in elderly and improve their overall oxidative metabolism in elderly diabetic patients. The fact that

chocolate can have therapeutic effects is not surprising. In their 2012 book, "Chocolate As Medicine," Philip Wilson and Jeffrey Hurst document its use as a medicine in Central America for hundreds of years. What is interesting here is the finding that a component of chocolate has its beneficial effects by modulating the dynamics of mitochondria.

Dr. Matthew Klein of Edison Pharmaceuticals in California provided an update on the promising drug known as EPI-743. In a study of patients with the mitochondrial disease Leigh syndrome, the drug significantly improved neuromuscular function and quality of life. Studies are also underway to assess the efficacy of EPI-743 in patients with either Friedrich's Ataxia or Rett Syndrome.

Three researchers presented their data on the therapeutic effects of Bendavia, a product of the company Stealth Peptides. This drug was shown to:

- 1) restore mitochondrial function to impaired heart muscle (Dr. Hani Sabbah, Henry Ford Hospital)
- 2) improve visual function previously diminished by diabetes by directly enhancing mitochondrial activity (Dr. Peter Oates, Cornell Medical College)
- 3) limit the damage to heart muscle caused by heart attacks by preserving mitochondrial function (Dr. David A. Brown, East Carolina University)

In a presentation the last morning of the symposium, Travis Wilson, CEO of Stealth Peptides, asserted that Bendavia, because of its mechanism of action, has the potential for therapeutic efficacy across a broad spectrum of mitochondrial diseases.

Research with a number of other drugs was also presented at the symposium. These include bezafibrate, N-acetylcysteine and phenylbutyrate. Not content to study the effects of just one drug, Dr. Sandipan Datta of U.C.-Davis School of Veterinary Medicine reported on a study in which he used a high-throughput screening method to evaluate the efficacy of 1600 FDA-approved drugs to confirm their beneficial effects on a cell model of LHON. His lab identified multiple agents that will be investigated further to assess their clinical effects.

This was an impressive symposium in that it clearly documented the progress that has been made in recent years. The words of Patrick Chinnery and Eric Schon have certainly proved prophetic. They wrote in 2003, "There are currently no treatments for mitochondrial disease, but various avenues show promise, and are likely to enter clinical practice within the next decade." (J Neurol Neurosurg Psychiatry 2003;74:1188-1199). It is exciting to contemplate what the status of mitochondrial disease therapy will be when another 10 years have come and gone.

The complete list of the 22 potential therapies referenced at the 2013 research symposium include: AICAR, Arginine, Bendavia, Bezafibrate, Brain-derived neurotrophic factor, Citrulline, CoQ10, Creatine, Dichloroacetate, Epicatechin, EPI-743, Gene therapy, Idebenone, M1- Polyamide, Misc. drugs, N-Acetylcysteine, Nicotinic acid, Phenylbutyrate, Phytoestrogens, Photobiomodulation, Resveratrol, Small-molecule compounds

*Anyone reading this article should understand that the UMDF does not make drug recommendations. Patients should consult with their healthcare providers to learn if specific therapies are appropriate for them.*

# UMDF support

Connecting our community.

## UMDF Highlighted Support Events in April, May and June

**March 3, 2012.** The Central Florida Chapter of the UMDF held a Meet and Greet Picnic at Maitland Community Park in Maitland, FL. Despite a chilly and windy day, a great time was had by all! Thank you, Jennifer Sauter for setting up this fun event!

**March 12, 2013.** The South Carolina Chapter of the UMDF hosted a webinar titled, "Gastrointestinal Dysmotility Diet Guideline Overview." This informative educational session was presented by Jill Taliaferro, RD,CN,SC, of Thrive RX. Thank you, Jill Taliaferro and Charlene James of Gaffney, SC, for making this webinar a great success.

**March 21, 2013.** The Western New York Mito Group held a support group meeting at Williamsville United Methodist Church in Williamsville, NY. Dr. Georgirene Vladutiu made a surprise visit to the meeting. Linda Roesch, Co-leader of the Western New York Mitochondrial Support Group said, "Dr. Vladutiu talked about some of the different testings that are going on through NAMDC (North American Mitochondrial Disease Consortium). We had some great discussions with her."

**March 24, 2013.** UMDF Houston Family Day at the Zoo was a roaring good time for Houston area families! Over forty attendees enjoyed a day at the zoo with morning talks from the following doctors: Dr. Bruno Chumpitazi of Texas Children's Hospital, Dr. Mohammed Numan of the UT Dysautonomia Center and Dr. Mary Kay Koenig of the UT Mitochondrial Center of Excellence. A special thanks to the doctors for volunteering to speak on a Sunday morning! Special thanks also to Joy Roeh and Melissa Knight for organizing a zoo-tastic event and to Trisha, JC and Anastasia Kranz for contributing tasty muffins for mito at the entrance check-in.

**April 15, 2013.** A Patient and Family Meeting was held at East Tennessee Children's Hospital in Knoxville, TN featuring Bruce Cohen, M.D., of Akron Children's Hospital. Dr. Cohen provided an update on mitochondrial medicine and then answered any questions posed by the attendees. Fifteen families attended this event. A special thank you goes to Nancy Rubio and Meghan Shields for helping with the event!

**April 20, 2013.** The third annual UMDF Texas Mito Family Fun Day was held at Morgan's Wonderland Park in San Antonio, TX. Sixty-four UMDF members from across Texas gathered for a networking lunch and family fun in the ultra-accessible amusement park. UMDF received a very generous grant, which will fund this event for three years, from Palm Valley Lutheran Church in Round Rock, TX. A special thanks to Manuel and Nicole Castro for organizing this Texas-sized Fun Day.

**April 27, 2013.** The Chicago Area UMDF Chapter welcomed Dr. Dmitriy Niyazov, Section Head – Medical Genetics at the Ochsner Clinic, New Orleans, as a guest speaker. Dr. Niyazov spoke to approximately 50 attendees from the Chicago area regarding mitochondrial disease challenges of diagnosis and treatment. Special thanks to Tina Higens for organizing this educational event!

**May 3, 2013.** Las Vegas was the location for a family meeting featuring a presentation by Dr. Cohen. The audience was small, but the questions were powerful and helped raise awareness in the area. Thanks to

Paulette Milton, UMDF volunteer, for helping to make it a pleasant evening for all that attended.

**May 20, 2013.** A Patient and Family Meeting was held at Children's Hospital New Orleans in New Orleans, LA featuring speaker Dr. Bruce Cohen of Akron Children's Hospital and Dr. Dmitriy Niyazov of Oschner Children's Health Center. Each doctor presented and answered questions from the families and patients in attendance. Approximately 45 attendees came from across Louisiana and also from Mississippi and Alabama. A special thanks to local volunteers, Rhonda Mailhos and Chantel Wooley, for helping make this event and the pre-meeting meet and greet successful.

**May 21, 2013.** The Support Group in Detroit decided to mix it up in May from their usual support format and hold Spring Fling. Over 20 people gathered at a local restaurant to network, and enjoy refreshments with others affected by mitochondrial disease. A great time was had by all. Our appreciation goes to Missy Leone and Julie Scott for arranging this meeting as well all of the ongoing support that they offer.

**June 22, 2013.** Dallas-area Support group member families enjoyed a Mito Splash Morning at their local Hawaiian Falls waterpark in Garland, TX. Twenty-two members enjoyed Champions Day, a day that the park opens early and exclusively to families with special needs. A special thanks to local support group leader, Shawna McElveen, for helping to organize a little splash time for Dallas-area families!

**June 22, 2013.** Jacksonville, FL volunteers held their first ever Support Meeting/Meet and Greet! Many thanks to Andrea Lanier for organizing the event!

**June 26, 2013.** A family meeting with Dr. Bruce Cohen as the guest speaker was held in Wilmington, DE. More than 30 UMDF members and health care providers attended to increase their knowledge of mitochondrial disease. A special thanks to UMDF Volunteer, Carrie Mullen for her efforts in making this an enjoyable evening for all.

**June 29, 2013.** A UMDF Family Meet and Greet for those in Western Pennsylvania was held just outside of Pittsburgh at Boyce Park. Over 20 people enjoyed networking, crafts and food. Thanks to everyone who attended and especially to Amanda Butler for organizing the fun morning.

Support group meetings were also held in Huntsville, Chicago, Richmond, Williamsville, Milwaukee, Indianapolis, Detroit, Dallas and Memphis. Additional meetings dates and locations are being added on a regular basis, so be sure to check the UMDF calendar, [www.umdf.org/calendar](http://www.umdf.org/calendar), for a meeting in your area. All of our meetings are organized and run by dedicated local volunteers who have chosen to be a part of the cure. Not a meeting in your area? You can choose to become the part of the cure by planning a one-time Meet & Greet or organizing regular meetings with the assistance of the UMDF. If you are interested or need more information, call or e-mail the UMDF or go to [www.umdf.org/volunteerops](http://www.umdf.org/volunteerops) for more information.



Ian Castro explores a sensory butterfly wall at Morgan's Wonderland Park.

# Mitochondrial Disease and Disability Benefits

by Bryan D. Krumdiack, CPA and husband of Joy Krumdiack, a disabled mitochondrial patient and NW Washington Ambassador for UMDF. You can contact either of them by e-mail at [bryankr@msn.com](mailto:bryankr@msn.com).

So you just received the diagnosis that you or a loved one has a mitochondrial disease. There are probably a lot of thoughts spinning through your head. Maybe one of them regards money. How do I pay for all the bills? What if I can't work? Are there any resources available? One of the areas you should consider is Social Security Disability.

There are two different programs available under the Social Security Act that you might be eligible for. The first is the Social Security Disability Insurance also known as SSDI. It is a program for disabled workers and their dependents. This is a program for workers who have paid into the Social Security program. The second program is the Supplemental Security Income also known as SSI. It is a program for individuals who have limited income and assets and their dependents. When you apply for disability with the Social Security Administration, they will consider your eligibility under both programs.

To qualify under the SSDI program you must meet certain qualifications. You must be a disabled worker under age 65; or a family member of a disabled worker (these are referred to as auxiliary benefits). You do not have to be disabled to qualify for

auxiliary benefits. Under the auxiliary benefits, there are benefits for spouse and divorced spouse, child benefits and parent benefits.

To qualify under the SSI program you must also meet certain qualifications. Under the SSI program an adult or child who is disabled must meet certain income limits and certain resource limits in order to receive disability payments. SSI payments are normally only for U.S. citizens, although there are several exceptions.

The basis for these programs is a disability. To be disabled for the purposes of these programs, you must have a medically determinable disorder. You must also have no ability to engage in substantial gainful activity. This means that you aren't able to earn more than certain amounts set by the program. The disability must be severe enough that you are disabled for at least 12 months. If you think you may qualify for one of these programs, you should contact the Social Security Administration for additional information or application information. There are also several good self-help guides available that give more information on the programs and their requirements. If you qualify for benefits, it could be one less thing to worry about with your mitochondrial diagnosis.

## Mito S.P.A. – Support, Play and Acknowledgement Caregiver Workshop

September 14, 2013, 8:30 a.m. – 8:30 p.m. (lunch and dinner included)

September 15, 2013, 9:30 a.m. - 11:30 a.m. (breakfast is included)

Embassy Suites Houston - Energy Corridor, 11730 Katy Freeway, Houston, TX 77079

Join UMDF in Houston for the Mito S.P.A. weekend workshop where caregivers and adult patients will engage in activities of Support, Play and Acknowledgement. Our individual members struggle with different daily symptoms and diagnoses. The goal of this weekend is to promote a spirit of unity amongst all adults caring for patients with mitochondrial disease and/or living with mitochondrial disease. The cost of registration for the S.P.A. Caregiver Workshop is FREE. You must be 21 years or older to participate. Register online now at [www.umdf.org/MitoSPAReg](http://www.umdf.org/MitoSPAReg) or call UMDF at 1-888-317-8633. After registering, you will receive a follow-up e-mail from [umdf.houston.connection@gmail.com](mailto:umdf.houston.connection@gmail.com) with additional details and requesting specific information/materials for Mito S.P.A. programming.

## UMDF Joins RareConnect™

This winter, the United Mitochondrial Disease Foundation (UMDF) joined RareConnect™. What is RareConnect™? RareConnect™ is a website that was created by EURORDIS (European Rare Disease Organisation) and NORD (National Organization for Rare Disorders) to provide a safe space where individuals and families affected by rare diseases can connect with each other, share vital experiences, and find helpful information and resources.

The "Understand" section of RareConnect™ promotes awareness and understanding through daily life experiences. You can find patient testimonies and contact information for patient organizations under this section. Feel free to share your story. The "Meet" section of RareConnect™ provides a safe place to meet friends, ask questions, start or join conversations, and

interact with others experiencing the same challenges. Connect with others around the world to share support and encouragement. The "Learn" section of RareConnect™ links to FAQs, published articles, upcoming events, and other documents providing the latest information for each disease-specific community. Read publications from medical experts and patient organizations in this area.



Right now, there is a Leigh Syndrome community active where people from around the world can connect and share their stories. Another community may be developed in the future. Go to [www.rareconnect.org](http://www.rareconnect.org) to check it out!

For more information contact Alexandra Freitas, Online Communities Coordinator, at [afreitas@rarediseases.org](mailto:afreitas@rarediseases.org).

# **UMDF Chicago Support Group Celebrates 10 Years**

What a milestone! The UMDF Chicago Support Group recently celebrated 10 years of unbroken meetings - that's about 80 support and educational meetings! The group is still going strong with most of its original members (some in the photo on right), and it also continues to welcome new members.



*The UMDF Chicago Support Group celebrated 10 years on June 22, 2013.  
Some of the members at that meeting pose above.*

## **UMDF Central Virginia Support Group Helps Reinstate Hospital Program and Doctor**

UMDF Member Judi Bartle of Glen Allen, VA heard a rumor that the Virginia Commonwealth University Health System (VCUHS) was going to deny funding of their Pediatric Palliative Care Program. Dr. Jean Teasley has been serving in the unpaid position of medical director of the VCUHS Pediatric Palliative Care and Pain Program since January 1, 2013. Bartle said, "Her position is important to our membership since palliative care equals symptom management for our patients who have chronic complex medical illness."

After confirming that the rumor was true, Bartle asked UMDF Central Virginia Support Group members to write letters asking for support of the program and of Medical Director Dr. Jean Teasley. UMDF members, their families and friends wrote 19 letters to VCUHS. In addition, a member of the Coalition Against Pediatric Pain and a physician wrote letters.

Bartle's request to UMDF members went out on May 29, and she asked them to have their letters in the mail by May 31 since the VCUHS was going to make a quick decision about the program. Bartle said, "That was really short notice for busy, overwhelmed mito families!" On June 5, 2013, Bartle was informed that VCUHS

had decided to reinstate funding for the Pediatric Palliative Care Program. The letter writing campaign was a success!

Since the letter writing campaign, Dr. Teasley is receiving salary support in her role, and VCUHS now has future plans to grow the program. Dr. Teasley is the Central Virginia's local expert on mitochondrial disease. She treats most of the kids in the area, as well as mentors and teaches other physicians that mitochondrial disease patients see, including those who treat adults. She attends almost every one of the UMDF Central Virginia monthly support meetings. Bartle said, "None of us would be where we are without her unrelenting time and efforts on our behalf." Dr. Teasley feels that her position as medical director will further facilitate education of health care providers for patients with chronic complex medical illness as she works with pediatrics and neurology.

The Central Virginia Support Group meets the third Tuesday of the month at the Children's Hospital of Richmond at VCU Brook Road Campus, 2924 Brook Road, Richmond, VA 23220. For details, contact Judi Bartle at [umdfcentralva@gmail.com](mailto:umdfcentralva@gmail.com) or 804-346-0653.

### **SAVE THE DATE! MITOCHONDRIAL MEDICINE 2014: PITTSBURGH**

**SCIENTIFIC PROGRAM: JUNE 4 - JUNE 7, 2014**

**FAMILY PROGRAM: JUNE 6 - JUNE 7, 2014**

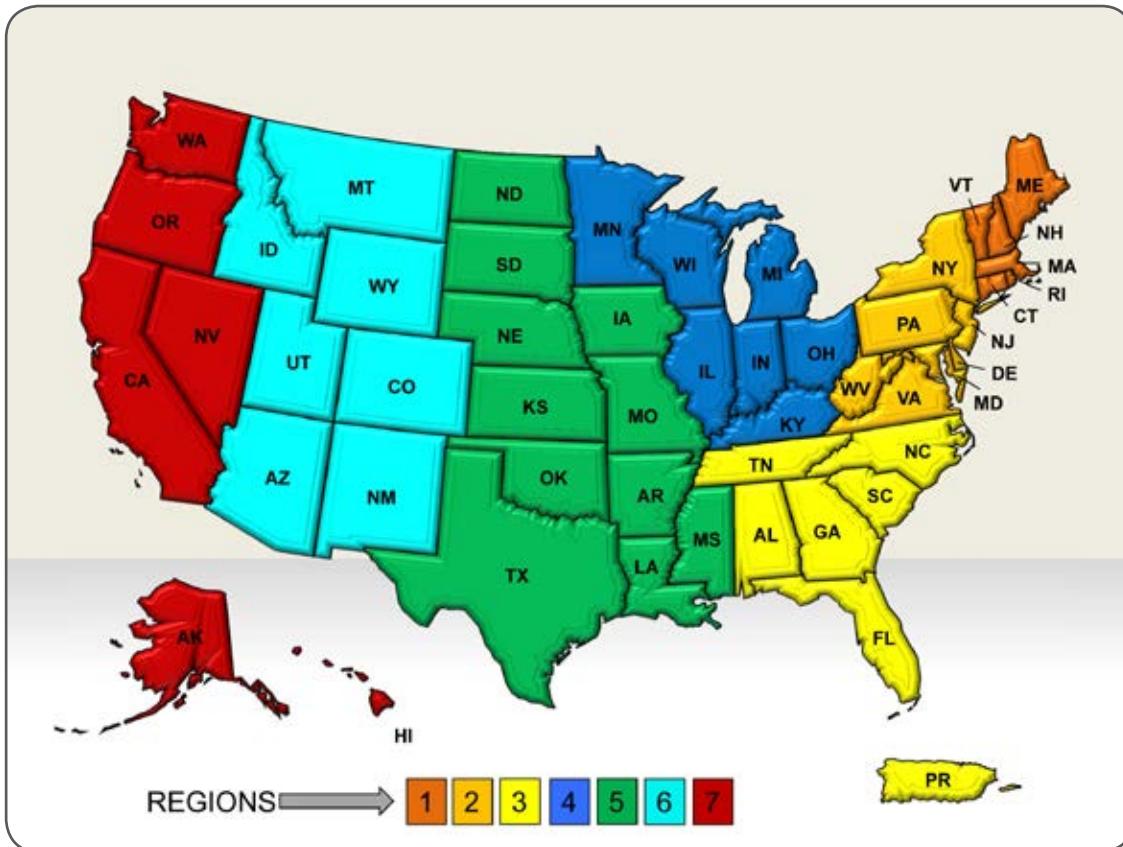
**SHERATON STATION SQUARE HOTEL**

**300 WEST STATION SQUARE DRIVE, PITTSBURGH, PA 15219**



In addition to the support staff at the UMDF National Office, help is available to you across the United States and around the world. To reach a state contact for support and/or if your state is not listed and you would still like to connect, simply e-mail to [connect@umdf.org](mailto:connect@umdf.org). When sending the e-mail, please include the leader's name or city/state in the subject line for us to better serve your needs. Interested in getting involved? Visit [www.umdf.org/volunteeropps](http://www.umdf.org/volunteeropps) or call 1-888-317-8633!

**Note:** The volunteer support leaders listed here have completed a Support Group Leader and/or Support Ambassador Agreement. If your name is not listed, please check with Member Services on the status of your agreement.



### New England Region (1)

#### **MAINE**

Amber Taylor, Bangor

#### **VERMONT**

MaryBeth LeFevre

#### **CONNECTICUT, NEW HAMPSHIRE, RHODE ISLAND**

Contact the National Office to Connect

#### **MASSACHUSETTS**

Julie Gortze, North Attleboro, MA

### Northeast Region (2)

#### **DELAWARE**

Kathleen Stapleford, Magnolia

Judy Weeks, Dover

#### **DC – See Virginia**

#### **MARYLAND**

Dawn Murphy, DC/Baltimore/Northern Virginia Chapter

#### **NEW JERSEY**

Laurel Smith, Delaware Valley Chapter

### Northeast Region (2)

#### **NEW YORK**

Kim Zuzzolo, NY Metro Chapter

Linda Roesch, Buffalo, Western NY Support Group

Jennifer Schwartzoff, Buffalo, Western NY Support Group

Erica Beyea, Buffalo, Western NY Parents Support Group

Sandy Sallaj, Buffalo, Western NY Parents Support Group

Jacqueline Perrotta, Albany

Lori Piccirilli, Binghamton

Kimberly Dedrick, Utica

#### **PENNSYLVANIA**

Daria Grabowski, Erie

Jessica Myers, Erie Mito Group

Kim Olenderski, Central Pennsylvania

Heather Pallas, Pittsburgh (children)

Karen Wilson, Pittsburgh (adults)

#### **VIRGINIA**

Heather Meyer, Lynchburg

Judi Bartle, Central Virginia Support Group

Sharon Hoffert, Central Virginia Support Group

Molly McCaffrey Adams, Richmond

## **Northeast Region (2)**

Sharon Goldin, DC/Baltimore/Northern Virginia Chapter  
Anne Tuccillo, DC/Baltimore/Northern Virginia Chapter

## **WEST VIRGINIA**

Contact the National Office to Connect

## **Southeast Region (3)**

*Margaret Moore, Regional Coordinator*

### **ALABAMA**

Margie Slemp, Huntsville, North AL  
Mito Group

### **FLORIDA**

Amber Ferrell, Gainesville, Central FL  
Mito Group  
Garry Krueger, North Central Florida  
Joan Morris, Titusville, FL  
Denise Richardson, Fort Lauderdale  
Holly Schneider, Coconut Creek  
Jennifer Sauter, Orlando, Central FL  
Mito Group  
Sophie Szilagy, North East Florida  
Marla Tobia, Tampa

### **GEORGIA**

Amy Blackwell, Kennesaw  
Hannah Bossie, Athens  
Sebastien Cotte, Atlanta  
Mary Beth Morris, Atlanta  
Cheryl Porter, Atlanta  
Gail LaFramboise, West Central Georgia  
Shelly Lorenzen, Sugar Hill  
Wendy Clegg, Loyd, Columbus  
Tiffany Tuggle, Stockbridge

### **NORTH CAROLINA**

Heather Baudet, Raleigh Durham  
Adriana Smith, Raleigh Durham  
Jenny Hobbs, Winston-Salem  
Terry Holeman, Fayetteville  
Christy Koury, Charlotte  
Kris Shields, Charlotte

### **SOUTH CAROLINA**

Christine Golden, Greenville/Spartanburg  
Hillary Miller, Charleston  
Karis Mott, Chapin

### **TENNESSEE**

Emily Culley, Memphis area Support Group  
Karrie LaCroix, Memphis area  
Courtney Fellers, Nashville area  
Nancy Garrison, Nashville  
Brandalyn Henderson, Nashville  
Nancy and Jeffrey Rubio, Knoxville

## **Great Lakes and Midwest Region (4)**

*Anne Simonsen, Regional Coordinator*

### **ILLINOIS**

Cherie Lawson, Chicago Area Support Group  
Vicki Ternberg, Chicago Area  
Gail Wehling, Chicago Area Support Group  
Luke and Leslie Kirby, Philo  
Victoria Helms, Southwest Area/St. Louis Support Group

### **INDIANA**

Melissa Edmondson, Evansville Area Support Group  
Lori Meyer, Evansville Area Support Group  
Jackie Parrish, Indianapolis Area

### **KENTUCKY**

Krystena Richards, Lexington & Cincinnati Areas  
Mandy Salensky, Lexington & Cincinnati Areas

### **MICHIGAN**

Carrie Gervasone, East Michigan/Detroit Area  
Missy Leone, East Michigan/Detroit Area Support Group  
Julie Scott, East Michigan/Detroit Area Support Group  
Genevieve Angeloff, Upper Peninsula  
Holly Worden, West Michigan/Grand Rapids Area Support Group

### **MINNESOTA**

Stacey Pieper, Minneapolis/St. Paul Area  
Atom Wolff, Minneapolis/St. Paul Area

### **OHIO**

Ruth Gerke, Central Area/Columbus  
Jody Thompson, Central Area/Columbus  
Darcy Zehe, Northeast Area/Akron, Cleveland  
Chris & Alisa Rawski, Northwest Area/Toledo

### **WISCONSIN**

Mindy Welhouse, Central Area/Appleton, Green Bay  
Terilyn Musser, Central Area/Eau Claire  
Jaqueline Bohne, Northern WI/Rhinelander, Tomahawk, Minocqua Area  
Josh Bartz, Southeast Area/Madison  
Karen Loftus, Southeast Area/Milwaukee

## **Central Region (5)**

*Cassie Franklin, Regional Coordinator*

### **ARKANSAS**

Lacie Moore, Rogers

### **IOWA**

Ronda Eick, Northern Iowa  
Kim Novy, Des Moines, Iowa Support Group

### **KANSAS**

Anne Tramposh, Kansas City Area

### **LOUISIANA**

Nicole DeJean, Lafayette  
Rhonda Mailhos, New Orleans Support Group  
Mandy Poche, Baton Rouge  
Tammy Reyna, Bossier City  
Anna Stewart, Bossier City  
Chantel Wooley, Covington

### **MISSISSIPPI**

Tracy Shedd, Vicksburg

### **MISSOURI (see also Illinois)**

Matt Bishop, Kansas City Area  
Keli Stone, St. Louis Area Support Group

### **TEXAS**

Shamayn Kennedy, Wichita Falls  
Jamie Buryanek, Houston  
Melissa Knight, Houston  
Trisha Kranz, Houston  
Joy Roeh, Houston  
Crystal Lopez, Dallas/Fort Worth  
Shawna McElveen, Dallas/Fort Worth  
Heather McNair, Dallas/Fort Worth

## **NORTH DAKOTA, SOUTH DAKOTA, & MONTANTA**

Marty Campbell, Beach, ND

### **NEBRASKA**

Dana Ritterbush, Omaha

### **OKLAHOMA**

Contact the National Office to connect

## **West Region (6)**

### **ARIZONA**

Gina Blair, Peoria  
Marin Pelletier, Phoenix

### **IDAHO**

Jennifer Pfefferle, Boise, Idaho Support Group

### **NEW MEXICO**

Stephanie Cassady, Albuquerque

### **UTAH**

Laura McCluskey, Orem

### **COLORADO AND WYOMING**

Contact the National Office to Connect

## **Pacific and Northwest Region (7)**

### **CALIFORNIA**

Norma Gibson, California Chapter  
Cheryl Burge, Inland Empire  
Cory Greenlee, La Verne

### **HAWAII**

Kimo Phan, Honolulu

### **OREGON**

Kimberli Freilinger, Monmouth Support Group

### **WASHINGTON**

Mareesa Henderson, Spokane

Joy Krumdiack, NW Washington

### **ALASKA, MONTANA, NEVADA**

Contact the National Office to Connect

### **INTERNATIONAL**

Rob Ryan, Australia  
John Carreiro, British Columbia  
Atanga Emmanuel, Cameroon  
Andrew Alexander, Hungary  
Nilam Agrawal, India  
Sajjad Haider, Pakistan  
Anne Hansen, Norway  
Vidar Hunstad Vik, Norway  
Rowland Dicker, United Kingdom  
Keely Schellenberg, Winnipeg

### **YOUTH AMBASSADORS**

Joe Wise, California  
Emily Swinn, Georgia  
Briana Garrido, Hawaii  
Haley Wroth, Massachusetts/Connecticut  
Alexandra Simonian, New York  
Tyler Liebegott, Pennsylvania  
Colleen Powell, Pennsylvania  
Devin Shuman, Washington  
Jordan Schmeer, Virginia

# Adult Corner

**The Adult Advisory Council Team (AACT) -  
Representing, Serving, and Supporting Affected Adults**

## Attendees Share Benefits of Symposium

Once again, the Adult Advisory Council Team (AACT) had the pleasure of hosting the seventh annual Adult Gathering and the third annual Young Adult Informal Gathering and Roundtable with Doctors in Newport Beach at UMDF's Mitochondrial Medicine 2013. "There is always a magic that happens when fellow 'mito patients' get a chance to sit in person and share their stories, triumphs and resources. This was the vision when we started these gatherings," said Sharon Shaw Reeder, AACT Chair. At this year's gathering, attendees discussed their road to getting their diagnosis and focused on what they do on a daily basis in living with the challenges of juggling all of their symptoms.

Reeder continued, "For some folks, they have never met another individual with their exact diagnoses and specific challenges. Not only does this happen at our adult gatherings but at our symposium. In general...there are many 'ah ha' moments. The feeling of isolation goes away when one is surrounded by others who have gone through the same experiences. I was personally struck by how many significant others attended our adult gathering...supporting their loved one. They have a voice too, and living with mitochondrial disease affects them in many ways. Many people do not have that ready support system in place through family and friends, so it was great to witness new friendships being made. The main benefit, other than the invaluable medical information and special adult sessions, is the personal networking that goes on during our annual symposium."



Sharon Shaw Reeder, AACT Chair; Jennifer Schwartzott, AACT Member and Gail Wehling, AACT Co-chair at the symposium.

Some of the topics that were discussed at the Young Adult Informal Gathering and Roundtable with Doctors included: exercise and pacing yourself, the risks and considerations of pregnancy, genetics and the difficulty of taking so many supplements and how much/which supplements actually help. Devin Shuman, UMDF Youth Ambassador, said that

a young adult should consider attending the symposium, "because it is a life changing experience. Meeting a group of young adults who know what you're going through changes your perspective on the disease. Being part of a peer group is especially crucial at our age, and every year I've gone, I've made instant, life-long friends. Becoming part of a community makes you more than just a patient, but a person, not to mention the amazing wealth of information you learn every year from the top experts."



Young adults: Becca DeMuri; Elle Streitwieser; Devin Shuman, UMDF Youth Ambassador; Gina Wood and Tricia Melland pose for a photo at the symposium.

### **Adult Advisory Council Team (AACT)**

Sharon Shaw Reeder, AACT Chair, California

Gail Wehling, AACT Co-chair, Illinois

Bob Brief, New York

Linda Cooper, California

Whit Davis, Pennsylvania

Kirah Fasano, Pennsylvania

Rev. David Hamm, Maryland

Pam Johnson, M.D., Kansas

Joy Krumdiack, Washington

Debra Makowski, Arizona

David McNees, Ohio

Jennifer Schwartzott, New York

Gregory Yellen, Maryland

#### **Medical Advisors:**

Bruce H. Cohen, M.D.

Amy Goldstein, M.D.



#### **PURPOSE OF AACT**

*To represent and serve the unique needs of the affected adult community and to ensure that those needs are adequately represented to UMDF resulting in enhanced services to the affected adult population.*

*AACT is a liaison to the UMDF Board of Trustees and will assess and evaluate, provide advice and guidance, and make recommendations to UMDF on adult-related issues.*

# 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.

Responders for this issue include:

William Copeland, Ph.D., National Institute of Environmental Health Sciences, Research Triangle Park, NC; Russell Saneto, D.O., Ph.D., Seattle Children's Hospital, Seattle, WA and Mark Tarnopolsky, M.D., Ph.D., FRCP(C), McMaster University, Hamilton, Ontario, Canada.

**The Question is...** I have connected with many patients who are strongly suspected of having a mito disorder but remain undiagnosed despite extensive biochemical and genetic testing (including comprehensive mtDNA and nDNA sequencing). Is there a theory about why this is? Is it possible for several partial defects to cumulatively result in disease? If someone is heterozygous for a missense mutation in TWO nuclear genes associated with recessively inherited mitochondrial disorders (i.e. has a mutation in SUCL2 and COX10), is it possible that together disease results? Is it possible that an mtDNA (mitochondrial DNA) mutation of unclear pathogenicity could result in disease if the patient is also a carrier of an nDNA (nuclear DNA) mitochondrial disorder?



**William Copeland, Ph.D.**

of genetic mutations in two or more genes (digenic and multigenic defects), and these mutations could reside in both the mitochondrial and nuclear genomes. This is one of the biggest challenges in the field to discern pathological mutations (especially recessive heterozygous ones) from benign single nucleotide polymorphisms. One way to think about this is in regard to energetic thresholds of organ function. A (one) particular recessive heterozygous mutation in gene X may result in a subclinical effect by itself. But, this mutation in combination with another mutation in gene Y (also subclinical in nature) may result in a drop in ATP production, especially when combined with an environmental stressor, resulting in the presentation of symptoms. Unfortunately, we have very little (if any) model systems to address digenic and multigenic defects in mitochondrial disease.

**The Question is...** I have a relatively mild mitochondrial disorder and am currently pregnant. Since my child is at an increased risk for mitochondrial dysfunction, would there be a different vaccine schedule recommended for her since many concurrent vaccines could cause metabolic stress?



## Response from Russell Saneto, D.O., Ph.D.

One of the first questions will be concerning whether your new youngster will have mitochondrial disease and that will depend on what type (mtDNA or nuclear encoded) you have. The American Academy of Pediatrics has a set of guidelines for immunizations that has been developed for normal healthy infants and children. If there is a thought that your newborn may have a mitochondrial disease risk, the first thing you should do is talk with your pediatrician. (In fact, it is always nice to have a pre-visit before the birth just to get acquainted with him/her.)

Then you can talk about working out a vaccination schedule that will keep your youngster up-to-date as best possible, without giving too many shots at one time. Some vaccinations will be deferred for a time while others will likely need to be given on time. For instance, here in Washington State, we have a high rate of pertussis with a population that is sparsely vaccinated (so there is no "herd-effect"-meaning that the percentage of vaccinated people is high enough to give passive immunization to the rest of the population). So, here in my state, I would suggest getting this vaccination on time, as well as you and the family making sure you are vaccinated. Your pediatrician should have this information for your state/living area. The live virus vaccinations may present a little more of a problem, but these can be given with a little forethought to what other vaccinations to delay. Again, your pediatrician will have a good feel for this. In the end, you will have to be adaptable and in good communication with your pediatrician (or family physician).



**Russell Saneto, D.O., Ph.D.**

**The Question is...** My 16 year old son was diagnosed with mito five years ago and has been on the mito cocktail with marked improvement in his cognition. (He was diagnosed with severe/profound retardation, epilepsy and distal arthrogryposis prior to the mito diagnosis.) However, his balance issues, hand contractures and constipation have seen a marked increase. The mito has primarily affected his cerebellum. Does this mean the mito cocktail is not working or that his disease will continue progression despite the cocktail?

## Response from Mark Tarnopolsky, M.D., Ph.D., FRCP(C)

The mitochondrial cocktail is not a "cure", and people can still decline even when on it. In general, it may slow things down and, in some, improve aspects of function. Before ascribing a decline to the disease per se, we always look at other potential mitigating factors, i.e., Infections, renal or gall stones, vitamin deficiencies, etc. I would also say that unless a definitive diagnosis is made, one must keep open to the possibility that there is an alternative diagnosis with secondary mitochondrial dysfunction. In this case, a comparative genomic hybridization test (microarray) would be warranted, if not done already.



**Mark Tarnopolsky, M.D., Ph.D., FRCP(C)**

## **Family Meetings and Grand Rounds for Health Care Providers**

The United Mitochondrial Disease Foundation (UMDF) has made Grand Rounds a top priority to reach out to physicians through introducing and/or broadening the knowledge base on mitochondrial diseases. The primary purpose of our Grand Rounds program is to provide continuing education to health care providers on topics specific to mitochondrial disorders, which also furthers the 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.

With the generous support from Transgenomic Labs, the UMDF hosts approximately 12-15 Grand Rounds per fiscal year throughout the United States. These meetings have received nothing but positive feedback from the medical community.

In addition to Grand Rounds, the UMDF also schedules a special family meeting with our members during this time to not only help educate patients/families about mitochondrial disease but to allow them to speak one-on-one with a mitochondrial specialist. These family meetings are also suitable for outreach to nurses, therapists, educators and various allied health professionals.

Please see the schedule below for family meetings and Grand Rounds. Invite your health care provider to attend or you may want to go to a family meeting.

Contact Janet Owens, UMDF Executive Administrative Assistant, for more information. She can be reached at the National Office, toll-free, at 1-888-317-8633, ext. 107 or at [janet@umdf.org](mailto:janet@umdf.org).

### **Upcoming Family Meetings and Grand Rounds with Bruce Cohen, M.D.**

<u>Date</u>	<u>Type of Meeting - Presenter</u>	<u>Location</u>
7/25/2013	Grand Rounds	Ronald McDonald House, Austin, TX
7/25/2013	Family Meeting	Embassy Suites Conference Center, San Marcos, TX
7/26/2013	Grand Rounds	Dell Children's Hospital, Austin, TX
8/1/2013	Family Meeting	Oak Brook Library, Oak Brook, IL
8/2/2013	Grand Rounds	Lurie Children's Hospital, Chicago, IL
8/7/2013	Grand Rounds	Mayo Clinic Children's Center, Rochester, MN
8/7/2013	Family Meeting	Gillette Children's Hospital, St. Paul, MN
8/8/2013	Family Meeting	Saint Mary's Hospital, Rochester, MN
8/8/2013	Grand Rounds	Gillette Children's Hospital, St. Paul, MN
8/9/2013	Grand Rounds	Saint Mary's Hospital, Rochester, MN
10/8/2013	Family Meeting	Nemours Children's Hospital, Orlando, FL
10/9/2013	Grand Rounds	Nemours Children's Hospital, Orlando, FL

*For complete meeting details, visit the UMDF calendar on the UMDF website: [www.umdf.org](http://www.umdf.org).*

### **Special Patient Meetings**

#### **Patient Meeting – Saturday, October 12, 2013, 10:00 a.m. - 3:00 p.m.**

A Patient Meeting with Drs. Douglas Wallace, Amy Goldstein, and Marni Falk will be scheduled at the 3rd Annual Translational Research in Mitochondria, Aging, and Disease Symposium in Philadelphia, PA. Visit [www.umdf.org/calendar](http://www.umdf.org/calendar) in mid August for more details.

#### **Patient Meeting – Wednesday, October 30, 2013, 6:00 p.m. – 9:30 p.m. (tentative)**

A Patient Meeting with Drs. Michio Hirano, Mark Tarnopolsky, and Sumit Parikh will be scheduled at the Child Neurology Society Annual Meeting in Austin, TX. Visit [www.umdf.org/calendar](http://www.umdf.org/calendar) in mid August for more details.

# In the Clinic

In this edition of UMDF Connect, we take you to The Molecular Diagnostics Laboratory at Cincinnati Children's Hospital. Dr. Huang is board-certified in Pediatrics, Clinical Genetics and Clinical Molecular Genetics. The primary interest of his lab is in translation research, such as the genetic basis of optic atrophy and other mitochondrial diseases. Dr. Huang recently moved to Cincinnati Children's Hospital Medical Center to direct the program of mitochondrial medicine. The goal of the program is to integrate the research, molecular testing and clinical service to improve the care of patients with mitochondrial disease.

## 1. Tell us how you became interested in mitochondrial disease patients?

I became interested in patients with mitochondrial disorders while I was a pediatric resident at Georgetown University Medical Center. I had seen patients with diagnoses of mitochondrial diseases, and they had extremely complicated medical histories. At that point, I wished I knew more about mitochondrial diseases. After my residency at Georgetown, I moved to Boston Children's Hospital, Harvard Medical School, where I did my Clinical Molecular Genetic Fellowship. There I worked with Dr. Mark Korson and Dr. Frances Dougherty in the Mitochondrial Disease Clinic. The clinic served almost the entire New England area. I still remember patients traveling from far away to see us in our clinic. I then realized how much of a burden and inconvenience traveling was for many families and felt that a local clinic would serve many people much better. After working at Boston Children's Hospital, I moved to California to work at the University of California, Irvine. Two years into my work, Dr. Douglas Wallace joined the University. He set up the Center for Molecular and Mitochondrial Medicine and Genetics (MAMMAG). For the next 8 years, I served as the Director and Co-director for the MitoMed Diagnosis Lab. As a member of the medical center team, I spent time taking care of patients and further developing my expertise in mitochondrial diseases. When the opportunity opened at Cincinnati Children's Hospital Medical Center, I decided to move here. My goal is to establish a local center for mitochondrial medicine and provide excellent care for patients with mitochondrial diseases.

## 2. Does your clinic accommodate children and adults?

Our clinic serves both pediatric and adult patients.

## 3. Does your clinic diagnose patients and provide follow up care?

Mitochondrial diseases are notoriously difficult to diagnose. Our expert team works to establish (or rule out) a diagnosis of mitochondrial disease for our patients. A physical examination is performed and further testing may be ordered to help us reach a diagnosis. Follow-up appointments and appropriate medical management will be provided.

## 4. Do patients need referrals?

Patients do not need a referral to make an appointment with our clinic, but their insurance companies may require a referral for reimbursement. Patients should contact their insurance companies before making an appointment.

## 5. What types of research are you doing right now? And have you made any discoveries in research?

We are currently developing cutting edge research in molecular genetic testing. Several years ago, we used "next-gen" (next generation) genetic testing technology to detect heteroplasmic mutations of the mitochondrial genome. We found that with this new DNA sequencing technique, we could detect very low levels of heteroplasmy. This testing is very accurate; for example, we can detect mutations even when they



Taosheng Huang, M.D., Ph.D.

are present in only 1-2% of the mitochondrial DNA. For mutations that are present in at least 5% of mtDNA, the test was 100% sensitive and specific, which means that the test is very good. We are hoping to develop a cost-effective way to diagnose mitochondrial diseases using new sequencing technologies.

Our research lab has been studying the pathogenesis of mitochondrial genome mutations using iPS cells (induced pluripotent stem cells) and cells that have combined the nuclear genome from one source with the mitochondrial genome from another source (cybrid cells). This technology enables us to study cellular function of mtDNA mutations in an identical nuclear background. With this research, we have found that the nutritional environment has a significant impact on mitochondrial function.

Clinically, I have seen a large number of patients with optic atrophy, and analyzing the OPA1 gene is a research interest of mine. OPA1 is encoded by the nuclear genome but the protein functions in mitochondria. It is very important for the mitochondrial morphology and function, such as ATP production. In our Drosophila knockout model, we found that antioxidant activity could partially rescue the optic atrophy phenotype. Since many patients with optic atrophy have loss of the retinal ganglion cells, we decided to develop iPS cell therapy to help with the retinal damage. We are working to be able to take a piece of skin and convert it to an embryo-like cell (iPS cell). This will hopefully help with the retinal ganglion cell damage. Pre-clinical trials are now on-going with animals.

The following link provides more information about the research currently done in our lab: <http://www.cincinnatichildrens.org/research/divisions/h/genetics/labs/huang/default/>.

## 6. How many researchers and clinicians are involved at your clinic?

Our clinic team consists of a geneticist and a genetic counselor. Additional faculty members at Cincinnati Children's Hospital Medical Center are interested in researching mitochondrial diseases.

## 7. How many mitochondrial disease patients do you see?

Our clinic meets weekly and we see approximately five patients per week. If the need for appointments increases, we will work to expand the number of patients we see.

## 8. Are there openings for new patients? Yes.

## 9. What are your plans and goals for the future at your clinic?

Currently, we see patients in clinic and then make necessary referrals to other specialties inside the hospital or to the patient's preferred providers. However, our goal for the future of our clinic is to expand into a multidisciplinary and comprehensive care center for patients with mitochondrial diseases. In the future, we hope to include multiple medical specialties (such as a cardiology, neurology, and genetics), and provide all of these services at one patient appointment. We currently also strive to integrate basic and translational research with cutting edge molecular testing and clinical care.

## 10. Tell us who works on your team?

Our clinical team includes Taosheng Huang, M.D., Ph.D.; Ashley Brazil, M.S., Genetic Counselor and a large group of physicians in different subspecialties. The Molecular Diagnostics Laboratory at Cincinnati Children's Hospital is using cutting-edge technology for clinical genetic testing, and our mitochondrial research program is rapidly expanding. For more information, please visit: <http://www.cincinnatichildrens.org/service/m/mitochondrial-disorders/tests/>.

## Fundraisers Benefiting the UMDF

**March 23, 2013.** The fourth annual Jackson Cully Mito – What? 5K was held at USA Stadium in Millington, TN. The event raised over \$26,000 for the UMDF in honor of Jackson Culley. A special thanks to Angie Hayes and Cindy Kraft for organizing this event!



Cookies at the fourth annual Jackson Cully Mito – What? 5K

CT. The event raised over \$2,000 for Lizzy's Research Fund with the UMDF! Great work!

**March 24, 2013.** Bruster's of Ingomar, PA hosted an Easter egg hunt, held a silent auction and donated 10% of all food sales to the UMDF. Over \$300 was raised for the UMDF. We are so "hoppy" Bruster's has chosen to support the UMDF!

**April 12, 2013.** The Elizabeth Piro Research Fund held a Zumba for a Cause event in honor of Lizzy Piro of Norwalk,

**April 15, 2013.** Dawgs for Mito, the country's first collegiate chapter of the United Mitochondrial Disease Foundation, helped organize the second annual Carson's Classic golf outing. The golf outing was held at the University of Georgia Golf Course in Athens, GA in honor of Carson Coburn. Over \$22,000 was raised for the UMDF!



UMDF Member David Porter, Jr., attended Carson's Classic golf outing. He is pictured above on the right. Also pictured are Thomas Farrell, Nick Biggee and Bob Garret.



This picture represents businesses and individuals who have donated for 8 years or more to the Bet on Baylee fundraiser. The UMDF staff has been told that these volunteers will hang their plaques with great pride. Jody Thompson, Baylee's mom and Bet on Baylee coordinator, is in the pink t-shirt and has a huge grin.

**April 13, 2013.** Baylee's family and friends were proud to put on the 10th Bet on Baylee in Roseville, OH. Baylee was not able to attend due to having strep throat, but her supporters kept the festivities going for 13 hours that day and night. The Thompson family has committed themselves to proudly giving funds to research and raising awareness for mitochondrial disease.

**May 4, 2013.** Stick it to Mito Fundraising Expo was held in Lakeview, MI in memory of Breylon Senn. The expo brought together over 10 vendors who raised funds for the Breylon Senn Research Fund with the UMDF. Thank you to Breylon's Aunt, Amanda Kinsey, for organizing this event!

**May 4, 2013.** The second annual UMDF Zumbathon was held in Sheffield Lake, OH in honor of Nicholas Zehe. Zumba enthusiasts enjoyed the activity outside of a local school with proceeds benefitting the UMDF!

Thank you Darcy Zehe for organizing the event again this year!

**May 5, 2013.** Josh Clark of Cincinnati, OH organized a fundraising event surrounding his participation in the Flying Pig Marathon in honor of Leslie Whitt-Williams. Josh, along with several family members and friends of Leslie, collected donations towards their running of the marathon to benefit the Leslie Whitt-Williams Research Fund.

**May 18, 2013.** The fourth annual UMDF Breylon Senn 5K Run/Walk/Stroll was held in Howard City, MI to benefit the Breylon Senn Research Fund. The event also featured a kid's fun run and balloon release in honor of what would have been Breylon's 5th birthday. Thank you to the over 350 people who attended the event in support of the Senn family and the UMDF. Also, special thanks to the family and friends who volunteer their time as a part of the Breylon Senn Research Fund Committee!

**May 30, 2013.** The UMDF was selected as the benefiting non-profit for this year's Chicago Turkish Festival and opening Black Tie Gala. The UMDF was featured in all promotional and press release items from the Turkish Council prior to the event and had an awareness booth during the gala and two-day festival. Thank you to the local Chicago volunteers who helped during the event and to the Taser and Goerges families who were instrumental in organizing this opportunity for the UMDF.



Nathaniel White and Marshal Andres at the Breylon Senn 5K.

**June 8, 2013.** The Turner Family in South Carolina held a HUGE Nealie's Hope Yard Sale at their home! The yard sale brought in over \$1,300 for the UMDF and a priceless amount of awareness! Thank you to all who helped make this sale possible!

**June 14-17, 2013.** The Zombspawn Gaming group held their annual Cartridge Champions Gaming Marathon which consisted of four days, 24 hours a day, of video game playing. The group was able to raise over \$600 for the UMDF with the worldwide gaming marathon! Thank you Jake and Tad for organizing this amazing awareness and fundraising event.

**June 15, 2013.** The inaugural Nicholas James Torpey Memorial Golf Outing dubbed the "Butterfly Classic" was held at Sycamore Hills Golf Course in Macomb, MI. The annual event was held in memory of Nicholas James Torpey and raised over \$22,000 for the Nicholas James Torpey Research Fund with the UMDF. Thank you to the supporters of the Nicholas James Torpey Research Fund for your hard work on this event!



*Promoting research and education  
for the diagnosis, treatment, and cure  
of mitochondrial disorders  
and providing support to affected  
individuals and families.*

RoseMary and Raymond Wasielewski at the Chicago Turkish Festival and Black Tie Gala.

**June 8, 2013.** The seventh annual Greater Mito Open was held at Broadlands Golf Course in North Prairie, WI. Approximately \$12,000 was raised for the UMDF. Thank you to all the families who made this outing a success!

and cash prizes to the top three teams. A special thank you to Jason Schmid for organizing this wonderful event that raised over \$23,000.

# Energy for Life Walkathons



UMDF Trustees Carl Daniels and Dr. Tyler Reimschisel proudly hold a check for the UMDF with Ellen, Hunt and Mike Hollis. Team Hunt raised \$10,000 for the Energy for Life Walkathon in Nashville, TN.



Team MegaWatts smile for a photo at the Energy for Life Walkathon in Atlanta, GA.

**April 6, 2013.** The third annual Energy for Life Walkathon: Nashville was held at Centennial Park in Nashville, TN. Twelve teams gathered together for a beautiful morning at the park and raised over \$40,000 for the UMDF! Thank you to all of our teams and walk committee for your hard work this year!

**April 13, 2013.** The second annual Energy for Life Walkathon: San Francisco was held at Golden Gate Park in San Francisco, CA. The second year brought 28 teams from across California and raised over \$72,000 hitting their walk goal! Congratulations to the teams and walk committee for all of your hard work! We will see you in 2014!

**April 20, 2013.** The second annual Energy for Life Walkathon: Atlanta was held at Centennial Olympic Park in Atlanta, GA. With over 650 walkers, they raised more than \$79,000. A big thank you to the entire walk committee for all your hard work!



Little Angel's Team gather together for a group photo at the Energy for Life Walkathon in San Francisco, CA.



Team Brittney's Bunch are above at the Energy for Life Walkathon in St. Louis.

**April 20, 2013.** The St. Louis Metro Area Mito Group held an Energy for Life Walkathon at Tower Grove Park in St. Louis, MO. With about 225 walkers, they raised over \$24,000. Thank you Victoria Helms, Keli Stone and Mike Stone and the entire committee for your hard work!

**May 18, 2013.** The fourth annual Energy for Life walkathon was held in Indianapolis Indiana. Almost 300 participants raised over \$28,000! Thank you to Co-chairs Thomasina Alexander, Kristen Senetar, all committee members and volunteers who braved another rainy Indiana Energy for Life Walkathon to show their support for affected families and individuals in the greater Indianapolis area.



Team BU Bioblasts pose for a photo at the Energy for Life Walkathon in Binghamton, NY.



The Pitt M&M's Team hold up test tubes of fruit flies at the Energy for Life Walkathon in Pittsburgh, PA. Team Captain Dr. Amy Goldstein and members of the Palladino Laboratory at the University of Pittsburgh comprised the team.

**April 27, 2012.** The second annual Energy for Life Walkathon: Binghamton was held at Otsiningo Park in Binghamton, NY. With over 220 walkers, they raised more than \$13,500. Congratulations to the Binghamton committee on such a successful walk!

**May 4, 2013.** The second annual Energy for Life Walkathon was held in Evansville, Indiana. Almost 500 participants raised close to \$33,000! Thank you to Co-chairs Melissa Edmonson, Lori Meyer, all committee members and volunteers who made a rainy day an amazing and successful day!



Wyatt Winternheimer and Team Walk N Roll "say cheese" at the Energy for Life Walkathon in Evansville, IN.

**June 1, 2013.** The second annual Energy for Life Walkathon: Pittsburgh was held at Heinz Field Lawn in Pittsburgh, PA. The event brought 28 vibrant teams to the Pittsburgh riverfront on a beautiful Saturday morning! Those teams were able to raise nearly \$49,000 for the UMDF! Thank you to all of the teams and this year's walk committee!! We look forward to seeing you in 2014!

**June 8, 2013.** The Energy for Life Walkathon: Iowa was held at the Zsavooz in Cedar Falls, IA. Over 80 participants joined together in the small Iowa town and raised over \$1,500! Thank you to our Co-chair, Ronda Eick who has been chairing the walk for the past seven years!

# Gifts from the Heart

**December 2012.** Brookside Animal Hospital in Coral Springs, FL hosted a fundraiser this past holiday season. Their clients were able to schedule appointments with a photographer to have pictures of their pets taken. Owners, Stacy and Glenn Kalick, matched donations for the UMDF. Thank you Stacy & Glenn for a great event!

**December 2012.** Pat Shelly of Harleysville, PA sent in donations that he collected this year as "Santa." Local schools, house parties, tree lightings, etc., that he attended made donations to the UMDF in his honor. Thank you so much Pat for donating your "jolly" time!

**February 2013.** A birthday party was held in memory of Samuel Robert Anderson from Mechanicsville, VA and \$125 was donated to the UMDF! Thank you to the Anderson family and guests for the generous donations!

**February 2013.** Shelly Hingsbergen of Cincinnati, OH sent in donations for the research fund for her children, Ayden and Faith. The Southern Ohio Bikers (SOB) held a Valentine's dinner and dance, as well as a benefit ride during awareness week for the UMDF. Thank you to the SOB for your continued support to the foundation!

**March 2013.** The Indiana Ice held their 19th annual Pack the House for Charity on March 2, 2013, at Bankers Life Fieldhouse in Indianapolis, IN. Over \$250 was raised for the UMDF! Thank you Indiana Ice!

**March 2013.** Great River Medical Center in West Burlington, IA chose the UMDF as their charity for Casual Day. During the month of March, the employees can purchase a \$2.00 sticker to dress down. Great River Medical Center raised almost \$600 for the UMDF! Thank you employees for such a great contribution in honor of Kristen McGuire!

**March 2013.** The employees at Ackerman, Link & Sartory in West Palm Beach, FL chose the UMDF for the month of March as recipients of their Dress Down for Charity. During the weekly Dress Down Day, over \$100 was raised in honor of Christopher and Michael Link of Palm Beach Gardens, FL. For the past three years, the firm has raised over \$1,000! Thank you for your ongoing support of the UMDF.

**March 2013.** Aidan Pasmore of Orion, IL held a fundraiser in honor of his brother, Jake, who has a mitochondrial disease. Aidan and his classmates raised almost \$500 for the UMDF! Thank you, Aidan and the students at CR Hanna School in Orion, IL!

**April 2013.** Pat Blankenship of Bronx, NY sent in the latest contributions, over \$300, from the Coins for a Cure boxes that she has placed in the cafeteria at work. Pat donates all collections to the UMDF in honor of her niece, Michelle Mohan, who has a mitochondrial disease. Thank you, Pat, for your continued support!

**April 2013.** The second annual Crop for a Cure was held in Sandy Springs, GA in honor of Braeden Vaughter. This all day scrapbooking festival complete with door prizes, classes and fun raised \$550 for the UMDF. A special thank you to Leyna Vaughter for organizing this exciting event!



Jake Pasmore with Principal Lowe at the CR Hanna School Fundraiser.

**April 2013.** The National Honor Society at Minnechaug Regional High School, in Wilbraham, MA raised money for the UMDF by holding their 10th annual basket raffle. Donations were received from the NHS members, as well as individuals and businesses in their community. They had 27 gift baskets, which raised over \$5,000! Thank you NHS Members at Minnechaug Regional High School!

**April 2013.** A special thank you to Gia Visto in Pittsburgh, PA for hosting a Business after Hours event and donating proceeds from the basket raffle to the UMDF. Thank you Gia Visto for supporting your community nonprofit, UMDF.

**May 2013.** Kennedy Polhamus of Midland Middle School in Midland, GA donated her birthday money in honor of Ava Wilson. Ava is the daughter of Kennedy's teacher, Jill Wilson. Kennedy also went door-to-door asking for donations and helped spread awareness about mitochondrial disease. We are very grateful for Kennedy's generosity!

**May 2013.** Mike Belcher of Blue Bell, PA held the fourth annual Brackets for Mito NCAA Tournament fundraiser in honor of his daughter Juliet Belcher. The event raised \$800! A big thank you to all who participated, and a special shout out to David and Denise Garvin from Maryland for contributing \$150 towards a cure!

**May 2013.** Mike Wilson of Wooster, OH organized a UMDF awareness table at a local lacrosse tournament and collected donations which totaled \$500.

**May 2013.** Faith, Hope, Inspire Jewelry hosted a jewelry party in Tampa, FL in honor of Megan Garrett and donated \$250 to the UMDF.

**May 2013.** As part of her senior project, Victoria Hayes of Johnson City, NY organized a penny wars competition at her school and raised \$220 for the UMDF. The sophomore class was the winning team and received an ice cream treat at lunch. Thank you Victoria for initiating this awesome fundraiser!

**May 2013.** The E.A. Harrold Elementary School in Millington, TN held a talent show in honor of Jackson Culley. Members of the student council voted to donate all profits from the show to the UMDF. Thank you to the kind students who worked so hard to make the show a success that raised over \$700!

**May 2013.** Kimberly Svagdis of Cumming, GA hosted an awareness night at Hardee's where 20% of sales were donated to the UMDF. Not only did Kimberly enlist her local Hardee's in Georgia, she also organized a fundraiser at the Hardee's in Macclenny, FL. Thank you Kimberly for raising awareness and funds for the UMDF!

**May 2013.** Each month, the Pittsburgh Airport Marriott donates to a different nonprofit through their dress-down fundraiser, and they selected the UMDF to be the recipient in May! They raised nearly \$300! Thank you to everyone who participated, and a special thanks to Sarah Nemanic, the Pittsburgh Airport Marriott's Human Resources Coordinator!

**June 2013.** The Friends of the Lavallette 8K Run & Fitness Walk donated to the UMDF in honor of Lauryn and Owen Boyle. The event, which would have been held on June 22, 2013, was cancelled due to the damage from Hurricane Sandy. They are looking forward to a fresh start in 2014, with a 5K run and fitness walk in Lavallette, NJ. Thank you Lavallette Race Committee for your ongoing support!

# Upcoming Events to Benefit the UMDF

**August 3, 2013.** The annual Run4Raley event will be held at the Philo Ball Park in Philo, IL. The annual event is held in honor of Raley Kirby and features a 5K run, 1 mile walk and a kids run! For more information, please visit [www.umdf.org/run4raleyl](http://www.umdf.org/run4raleyl).

**August 3, 2013.** Heidemarie Chernushin will be participating in the Cleveland Triathlon and Moving for Mito in honor of her dear friends Alexander and Lauren Lindemann. To learn more about Moving for Mito or to make a donation, visit [www.umdf.org/movingformito](http://www.umdf.org/movingformito)!

**August 10, 2013.** Please join our team for the Cincinnati Mudathlon. The Mudathlon will donate \$5 to the UMDF for each person that registers as part of our team. When you go register, select, "Joining A Team" and select our team name, "Slippery When Wet." This must be done in order for UMDF to receive the donation. Also, make sure you enter the word SLIPPERY in the promotional code box when you check out. This is required for the donation to go to the UMDF. Other members of our team have selected the 11:30 am start time, but you can choose whenever is convenient. Participants must be 14 years old to participate. Go to <http://www.mudathlon.com/> to sign-up!

**August 16 - 18, 2013.** Baylee's Ball Bash, an annual softball tournament, will be held at Cannelville Softball Field in Cannelville, OH in honor of Baylee Thompson and to benefit the UMDF. For details, contact Jody Thompson at 740-704-2994.

**August 17, 2013.** The fourth annual Energy for Life Walkathon: Minnesota will be held at Normandale Lake Bandshell in Bloomington, MN. For more details, please visit the following website [www.energyforlifewalk.org/minnesota](http://www.energyforlifewalk.org/minnesota).

**August 17, 2013.** The second annual Swing for a Cure for Alex Golf Outing will be held in Tiffin, OH in honor of Alex Sendelbach. The outing will feature door prizes and a prize for the best dressed golfer! To register for the outing, please visit [www.swingforacureforalex.org](http://www.swingforacureforalex.org). Questions? E-mail [laura1973lol@gmail.com](mailto:laura1973lol@gmail.com).

**August 24, 2013.** The second annual Energy for Life Walkathon: Akron will be held at Lock 3 in Akron, OH. We are hoping for better weather this year with an earlier walk date! Please visit [www.energyforlifewalk.org/akron](http://www.energyforlifewalk.org/akron) for more information and to form your team today!

**August 24, 2013.** Support the UMDF by purchasing a \$5.00 coupon to Macy's Shop for a Cause. By purchasing a shopping pass, you support the UMDF while enjoying a day of spectacular discounts. Shopping passes will be available for sale starting July 24, 2013, at this website [www.umdf.org/macysshopforacause](http://www.umdf.org/macysshopforacause).

**September 7, 2013.** The Energy for Life Walkathon: Detroit will be held at Dodge Park in Sterling Heights, MI. For more details on the new location, please visit the following website [www.energyforlifewalk.org/detroit](http://www.energyforlifewalk.org/detroit).

**September 7, 2013.** A Bowling for Mito event will be held at the Pleasant Hills Bowling Lanes in Wilmington, DE in honor of the children under the care of the 3F Nursing Staff at Nemours AI DuPont Hospital for Children. Please contact Corrie Scarberry at [cs3178@msn.com](mailto:cs3178@msn.com) for more information!

**September 14, 2013.** The third annual Energy for Life Walkathon: Salt Lake City will be held at Liberty Park in Salt Lake City, UT. Please join us for another great year by going to [www.energyforlifewalk.org/saltlakecity](http://www.energyforlifewalk.org/saltlakecity) and forming your team today!

**September 14, 2013.** The Energy for Life Walkathon: Kansas City will be held at the CommunityAmerica Ballpark (Kansas City T-Bones Stadium) in Kansas City, KS. For details, visit [www.energyforlifewalk.org/kansascity](http://www.energyforlifewalk.org/kansascity).

**September 14, 2013.** The inaugural Energy for Life Walkathon: Columbus, GA will be held the Golden Park Baseball Field/Riverwalk in Columbus, GA. For more information, visit [www.energyforlifewalk.org/columbusga](http://www.energyforlifewalk.org/columbusga).

**September 14, 2013.** The third annual Lizzy's Walk for KSS has moved to the fall! Please join us on the new date, but same location, at Calf Pasture Beach in Norwalk, CT. For more information, please visit [www.umdf.org/lizzyswalk](http://www.umdf.org/lizzyswalk).

**September 15, 2013.** The fourth annual Energy for Life Walkathon: Chicago will be held at Katherine Legge Memorial Park in Hinsdale, IL. The Mix 101.9 will be joining us again this year! For more details, please visit [www.energyforlifewalk.org/chicago](http://www.energyforlifewalk.org/chicago).

**September 20, 2013.** The Pittsburgh Group will be hosting a Mitochondrial Disease Awareness Night at the Pittsburgh Pirates game against the Cincinnati Reds! For more information, please visit [www.umdf.org/pirates](http://www.umdf.org/pirates). Be sure to get your tickets early as the club is expecting a sellout game!

**September 21, 2013.** The sixth annual Savannah's Hope Mito Walk will be held at Adventure Life Reformed Church in Altoona, IA. The event will include family friendly activities and much more! For information, visit [www.savannahshope.com](http://www.savannahshope.com).

**September 21, 2013.** The second annual Energy for Life Walkathon: Central Texas will be held at Old Settlers Park in Round Rock, TX. For more details, please visit [www.energyforlifewalk.org/centraltexas](http://www.energyforlifewalk.org/centraltexas).

**September 21, 2013.** The New York Chapter of the UMDF has planned its fourth annual Energy for Life Walkathon in Cheektowaga, NY. Help the committee celebrate by joining them at their new location, Cheekotaga Town Park. A picnic is scheduled after the walk. For more information, please visit [www.energyforlifewalk.org/westernnewyork](http://www.energyforlifewalk.org/westernnewyork).

**September 23, 2013.** The third annual "Life for Lila" Golf Classic, supporting the UMDF and presented by UNUM, will be held at The Golf Club at Ballantyne in Charlotte, NC. For more information about the event, to become a sponsor or register to participate, please visit [www.lifeforlila.org](http://www.lifeforlila.org) or contact Dan Richardson at [drichardson@hfcb.com](mailto:drichardson@hfcb.com).

**September 28, 2013.** The fourth annual Energy for Life Walkathon: Delaware Valley will be held along the beautiful waterfront at Campbell's Field in Camden, NJ. Please join us for a morning of fun and exercise as we spread awareness of mitochondrial disease. For details, go to [www.energyforlifewalk.org/delval](http://www.energyforlifewalk.org/delval).

**September 28, 2013.** The second annual Energy for Life Walkathon: Milwaukee will be held at Greenfield Park Site 3A in Milwaukee, WI. We look forward to you joining us again this year as we come together to show support of our teams! To form your team, go to [www.energyforlifewalk.org/milwaukee](http://www.energyforlifewalk.org/milwaukee).

**October 12, 2013.** The Charlotte Chapter has planned an Energy for Life Walkathon at Freedom Park in Charlotte, NC. The day will include an opening ceremony and post-walk program. For more information, go to [www.energyforlifewalk.org/charlotte](http://www.energyforlifewalk.org/charlotte).

**October 12, 2013.** The Glow Run 5K in Lawrence, KS has chosen the UMDF as their charity partner this year! If you are interested in participating, please visit [www.glowrun5k.com/ku.html](http://www.glowrun5k.com/ku.html).

**Ongoing.** Dr. Jaime Buryanek, who is not only a doctor but has mitochondrial disease, has created a special fundraiser to help the UMDF. It is called the Mito Challenge. For more information, go to [www.umdf.org/takethemitochallenge](http://www.umdf.org/takethemitochallenge).

## **UMDF Welcomes Four New Employees**

The UMDF has recently hired four new employees: Liz Weiss, Jeff Gamza, Fred Prefling and Philip Yeske, Ph.D.



Liz Weiss

On March 11th, Liz Weiss joined the UMDF as Special Events and Member Services Associate. She has a Bachelor of Arts degree in Media and Professional Communications and two certificates from the University of Pittsburgh: one in digital media and one in corporate/community relations. Weiss was an intern at the UMDF last summer in the Member Services Department. Prior to joining the UMDF, she worked at the Holiday Inn for more than two years as a Front Desk Agent.

Jeffrey Gamza was hired on April 1st as Multimedia Coordinator. Gamza is an award-winning multimedia communications manager, with over 15 years of experience in multimedia design, corporate communications and marketing in a variety of professional environments. Gamza began his career as Media Relations Coordinator at Alliance International, Ltd. in downtown Pittsburgh. He later served as Director of Media and Communications for USA Field Hockey, covering the 2008 Olympic Games in Beijing for the United States Olympic Committee. Gamza comes to UMDF after most recently serving as Marketing Specialist for Conco Systems in Verona, where his article, "Optimizing Condenser Tube Life with Non-Destructive Testing," was featured on the cover of the December 2012 edition of "Energy-Tech" magazine. He holds a Bachelor of Arts degree in Journalism from the University of Pittsburgh and a Masters in Business Administration in Marketing from Robert Morris University.



Jeff Gamza

## **UMDF Member Receives Community Award**

UMDF Member Christy Koury of Matthews, North Carolina, was recently named one of Charlotte's 50 Most Influential Women by the *Mecklenburg Times*. She has been an active member of the UMDF since 2008. Koury led the past four Energy for Life Walkathons in Charlotte as Co-chair, and she was instrumental in bringing UMDF's Physician Grand Rounds to Charlotte's two largest hospitals in 2010. She is a previous winner of the



Fred Prefling

Fred Prefling recently joined UMDF as Associate Director of Corporate Development. Prefling will be working on a part-time basis, and his role at UMDF will be pursuing donations from corporate donors. He has a Bachelors Degree in Marketing from Myers University in Cleveland Ohio.

Prefling's background in consumer product sales and sales management has prepared him well for this position. He has worked for two Fortune 500 companies, Kraft and Pillsbury, along with two privately held food companies, Kozy Shack Pudding and Dora's Naturals. He had progressive careers at these companies, having held the title of Sales Director with regional and national responsibility.

Philip Yeske, Ph.D., former UMDF Board of Trustee, was recently hired as the Science and Alliance Officer. Prior to joining the UMDF, Dr. Yeske served as President and Chief Executive Officer of Fluorous Technologies, Inc., a Pittsburgh-based early stage life sciences company, while also sitting on the company's Board of Directors. He also worked at the Bayer Corporation for 10 years, where he held positions progressing from basic research to global account management, including a four year marketing assignment at Bayer's corporate headquarters in Leverkusen, Germany.

Dr. Yeske holds a Bachelor of Science degree in Chemistry from Allegheny College and a doctorate in organic chemistry from Emory University. He completed a Postdoctoral Fellowship at the University of Pittsburgh.



Philip Yeske, Ph.D.

UMDF's Stanley A. Davis Leadership Award, which is the most prestigious UMDF National Award honoring a UMDF Leader who best represents the selfless dedication and commitment to the UMDF Mission as that of its namesake. An inspiration to many through her good works and tireless advocacy, Koury has helped to increase awareness of mitochondrial disease and to improve medical care in the Charlotte community.

# Mitochondrial Disease Awareness Week 2013

How will you raise awareness for mitochondrial disease in September? Awareness Week is scheduled this year for September 15-21, 2013. If you are looking for some ideas, the UMDF has a number of suggestions on our Awareness Week web page at [www.umdf.org/2013awareness](http://www.umdf.org/2013awareness). If you are looking for specific ideas, help is just a click away. Visit [www.umdf.org/awarenessideas](http://www.umdf.org/awarenessideas). We also offer you the availability to download materials for any presentations or meetings that you are planning.

If you are planning to conduct an outreach activity, please visit [www.umdf.org/2013materials](http://www.umdf.org/2013materials) to place your

order for materials that you may need for your distribution. All orders for materials must be received by 5:00 p.m. EST on September 6, 2013. If you have any questions about these materials and their appropriate uses, please contact our Member Services Department, toll-free at 1-888-317-UMDF.

If you are planning an Awareness Week activity, please tell us about it and send us pictures so that we can include them online and in an upcoming newsletter. Submit your Awareness Week activity photos and stories to [news@umdf.org](mailto:news@umdf.org).

## UMDF Announces Changes to Board and SMAB

In June, the UMDF Board of Trustees said goodbye and thank you to three members whose terms had ended: Sharon Shaw Reeder, Vice Chair; Gerald A. Cook, Esq., and Joseph Rice. All three served on the UMDF Board of Trustees for nine years.

Patrick Kelley will now serve as the Vice Chairman of the Board of Trustees. Carl Daniels has taken on the role of Secretary and Linda Cooper will be the Trustee At Large.

There have also been changes to the UMDF Scientific and Medical Advisory Board (SMAB). The terms of three members

ended: David Chan, M.D., Ph.D.; Patrick Chinnery, Ph.D., MRCP and Mark Tarnopolsky, M.D., Ph.D., FRCP(C). Each of them served for five years.

The three new members of the SMAB include Carla Koehler, Ph.D., UCLA, Los Angeles, CA; Mark Korson, M.D., Tufts Medical Center, Boston, MA and Gerard Vockley, M.D., Ph.D., University of Pittsburgh Medical Center, Pittsburgh, PA.

The UMDF is extremely grateful for the contributions each person has made to the organization, and it welcomes the newly appointed members.

## UMDF Grieving Program

The UMDF reaches out to those who recently lost a loved one affected with a mitochondrial disease. If you know of someone who passed away, and you would like to receive information from the UMDF, please call the UMDF National Office, toll-free, at 1-888-317-8633.

## UNITED MITOCHONDRIAL DISEASE FOUNDATION STAFF

### EXECUTIVE STAFF

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CEO/EXECUTIVE DIRECTOR

**JANET OWENS**

EXECUTIVE ADMINISTRATIVE ASSISTANT

**PHILIP YESKE, PH.D.**

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**TANIA HANSCOM**

SPECIAL EVENTS COORDINATOR

**RACHEL MAZUR**

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**NICOLE SHANTER**

SPECIAL EVENTS AND  
DEVELOPMENT ASSOCIATE

**FRED PREFLING**

ASSOCIATE DIRECTOR OF  
CORPORATE DEVELOPMENT



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**ALISON COOLEY**

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**JEFF GAMZA**

MULTIMEDIA COORDINATOR

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**MELINDA O'TOOLE**

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**JEAN BASSETT**

RESEARCH GRANTS COORDINATOR

**Liz Weiss**

SPECIAL EVENTS AND  
MEMBER SERVICES ASSOCIATE

### REGIONAL COORDINATORS

**CASSIE FRANKLIN**

REGIONAL COORDINATOR - CENTRAL

**MARGARET MOORE**

REGIONAL COORDINATOR - SOUTHEAST

**ANNE SIMONSEN**

REGIONAL COORDINATOR - GREAT LAKES



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**Donate today!**  
Every dollar counts.  
Visit [www.umdf.org](http://www.umdf.org).

## **United Mitochondrial Disease Foundation** **~ Decade of Difference ~**

- In 2003, our cumulative dollars spent on mitochondrial research had not yet reached a million. Today, UMDF has contributed more than \$10,000,000 leading to new clinical trials and potential treatments.
- In 2003, there were three clinical trials related to mitochondrial disorders. Today, there are 305 mitochondrial-related clinical trials ongoing.
- In 2003, researchers were just beginning to understand the link between mitochondrial dysfunction and other more common diseases. Today, a clear link has been established between dysfunctional mitochondria and Alzheimer's, Parkinson's, diabetes, certain cancers and even the aging process itself.
- In 2003, UMDF had six chapters and 20 support groups. Today, UMDF has representation in every state and in 152 countries.
- In 2003, only two members of Congress knew about mitochondrial disease. Today, 372 members of the House and Senate have been informed. When they make decisions about how to spend significant federal money on health-related issues, mitochondrial disease will be on their minds.
- In 2003, there were 124 participants at the UMDF symposium, including only three from other nations. Today, more than 500 scientists, clinicians, allied health professionals, and family members attend each year, representing 16 countries.
- In 2003, gene sequencing was just a dream. Today, gene sequencing is a reality, identifying areas where mutations occur and targeting potential treatments.
- In 2003, 36 researchers applied to UMDF with research proposals. Today, over 200 new research proposals are received annually.
- In 2003, there were no UMDF "Grand Rounds" (programs where mitochondrial specialists travel to different hospitals to brief medical personnel on mitochondrial disease). Today, over 80 grand rounds have taken place, serving approximately 50 people each time, leading to 4,000 medical professionals being informed about the diagnosis and treatment of mitochondrial disorders.
- In 2003, there were only 21 identified "mito docs" worldwide. Today, there are nearly 400 identified mito docs worldwide.

### **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.**

UMDF's intent is to keep you informed. We ask that you always discuss any diagnoses, treatments or medications with your personal physician. UMDF assumes no liability for any information in UMDF Connect.