

## UMDF Prepares for 15th Annual Symposium



*Newport Beach Marriott Hotel & Spa*

The United Mitochondrial Disease Foundation (UMDF) is holding its 15th annual symposium this year - Mitochondrial Medicine 2013: Newport Beach. The symposium will be held at the Newport Beach Marriott Hotel & Spa in Newport Beach, California. The Scientific Program will be from June 12 - 15, 2013 and the Family Program will be held on June 14 - 15, 2013.

Every year, the UMDF holds a symposium, which is organized by staff with input from volunteers. Researchers and clinicians from all over the world, representing many scientific and medical disciplines, attend research and clinical sessions. At the same time, patients and families attend family sessions and meet some of the top clinical mitochondrial specialists. There are many opportunities for networking and the exchange of information among physicians, researchers, patients and families. The symposium includes a four-day scientific/clinical program and a two-day patient/family program.

Donors' gifts not only help UMDF offer this one-of-a-kind conference, but they also help people like Sonya Murray go, who would not otherwise be able to attend. She was among 25 people who received a scholarship to attend the 2012 symposium. Murray of Smyrna, Tennessee, said, "What was so special to me about attending the symposium is that there are famous doctors right in front of you, and you can talk to them. It's not like seeing a superstar in Nashville and not being able to talk to them because you want to respect their privacy. These doctors are superstars to us, and yet they are there to talk to us."

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*Every dollar counts! To contribute to research, go to [www.umdf.org](http://www.umdf.org) and click on "Give Today."*

### White House Meeting Starts Dialogue with Administration

In an effort to begin a conversation with the Obama Administration about the importance of understanding mitochondrial dysfunction and disease and its impact on a host of other diseases, the United Mitochondrial Disease Foundation (UMDF) was able to secure a high level meeting at the White House to discuss the topic. The meeting was hosted by Carole Johnson, Senior Advisor for Healthcare for the White House Domestic Policy Council (DPC). The DPC coordinates the domestic policy-making process in the White House and offers advice to the President.

Participating in the meeting with Johnson on behalf of the UMDF was Charles A. Mohan, Jr., CEO/Executive Director; Richard A. Leach, UMDF Trustee; Phil Yeske, Ph.D., UMDF Trustee; Douglas Wallace, Ph.D., Director of the Center for Mitochondrial and Epigenomic Medicine at Children's Hospital of Philadelphia; Vamsi Mootha, M.D., Professor in the Department of Systems Biology at Harvard and Department of Medicine at Massachusetts General Hospital; and Bruce Cohen, M.D., Director, Pediatric Neurology,

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# From the Chairman

It's not too often that someone gets an invitation to the White House, but that's exactly what happened in February, when the UMDF was invited to meet with Carole Johnson, Senior Advisor for Healthcare for the White House Domestic Policy Council (DPC).



dollars. While one meeting doesn't solve all of these problems, we are confident the conversation with the Administration will continue.

I also want to personally invite you to Mitochondrial Medicine 2013: Newport Beach. The UMDF symposium has become internationally recognized as the place for

scientists, clinicians, and allied health members to gather with affected individuals and families. "Mitochondrial Therapeutics - From Bench to Bedside" is the topic for the scientific sessions, which will begin on June 12. While there are many intriguing topics for members of the scientific and medical community, we are especially interested in the sessions that deal with bringing new therapies to the market.

Details about the meeting are in this newsletter. As you know, UMDF and our members have been very active in trying to educate our elected officials about mitochondrial disease and mitochondrial dysfunction and the promise that research holds for more common diseases like Parkinson's, Alzheimer's, diabetes, and such. Thanks to all of you, we have been successful in keeping the National Institutes of Health (NIH) focused on this issue, and now we have begun the conversation with the Obama Administration.

Meeting with Ms. Johnson was important because it allowed us to clearly explain the current research, the links to other diseases, and diagnostic and treatment costs associated with mitochondrial disease. In addition, we know that many of the diseases of aging have a component of mitochondrial dysfunction in their progression – which are very similar to primary mitochondrial diseases, which are caused by mitochondrial dysfunction. These types of diseases have the biggest impact on the cost of health care. Our message was clear – research into mitochondrial dysfunction and disease could chart the course for treatments and potential cures for these other illnesses and could save our health care system billions of

Our family sessions start June 14th. Our popular, "Ask the Mito Doc" sessions will be held, along with special teen sessions. For the early birds on June 13th, we have a special half-day program on Leber's Hereditary Optic Neuropathy (LHON). As you might know, LHON has the highest occurrence of all of the primary mitochondrial diseases. In addition, we are offering a number of patient and family centered talks, such as dealing with chronic illness, insurance issues, genetics, and help for children with mitochondrial disease. We will also provide updates on all of the mitochondrial clinical trials that are in process and how you might participate. It will be an amazing week, and we hope to see you in Newport Beach, California, in June!

W. Dan Wright, UMDF Chairman



HOPE. ENERGY. LIFE.

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For details call  
Nicole Shanter  
1-888-317-UMDF

**White House Meeting Starts Dialogue with Administration**

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*UMDF representatives pose before they brief President Obama's Senior Domestic Policy Advisor Carole Johnson on mitochondrial disease at the White House.*

Akron Children's Hospital. Each was able to discuss with Johnson their role in research and in the clinical care of affected children and adults. "This is the next big thing," said Dr. Mootha, in telling Johnson about the impact this research could have on human health and for an Administration already tackling health care and economic issues.

Statistics indicate that within the next decade or so, there will be more people over the age of 65 as opposed to those under the age of 5. The group stressed to Johnson that as our population ages, more and more health care dollars will be needed to address the medical needs of this aging population. If we continue down this path, not only will it have a financial impact on health care costs, but on the nation's budget. "For many years, we have been telling members of Congress and

the U.S. Senate that investing in research into mitochondrial dysfunction not only may lead to potential treatments and cures for our patient community, but for other diseases like Parkinson's, Alzheimer's, diabetes, and some cancers," said Charles A. Mohan Jr., CEO/Executive Director of the UMDf. "This was an important meeting because it gave us the opportunity to share with the administration the tremendous benefits this research may provide in leading to the prevention of mitochondrial disease, other more well-known diseases, and the diseases that come with aging."

Johnson said the information presented at the meeting was incredibly useful and requested additional information as the conversation will continue between the UMDf, the scientific and medical community, and the Obama Administration.

**Edison Pharmaceuticals Initiates Phase 2 of EPI-743 Clinical Trial**

Edison Pharmaceuticals announced the initiation of a phase 2 clinical trial entitled, "Double-Blind, Placebo-Controlled Clinical Trial of EPI-743 in Patients with Cobalamin C Defect." Cobalamin C deficiency represents an inherited disorder that induces perturbations in the metabolism of glutathione. "Given the central role of glutathione in cellular redox balance and antioxidant defense systems, we are eager to explore whether a therapeutic that increases glutathione such as EPI-743 will provide clinical benefit," stated Professor Carlo Dionisi-Vici, Division of Metabolism, Bambino Gesù Children's Hospital- the trial's principal investigator. The rationale underlying this investigation is centered on understanding whether EPI-743 - a therapeutic targeting primary defects in electron transport - might hold therapeutic benefit in disorders of intermediary metabolism that also result in redox disturbances.

The trial is a placebo-controlled study lasting 12 months, preceded by a six-month run-in phase for all patients to establish a defined clinical and metabolic baseline. The primary endpoint is improvement in visual function with secondary outcome measurements assessing neurologic and neuromuscular function, glutathione biomarkers, quality of life, as well as safety parameters. More information on study specifics is available at [www.clinicaltrials.gov](http://www.clinicaltrials.gov).

Edison Pharmaceuticals continues to recruit patients for a double-blind randomized placebo-controlled trial of EPI-743 for children with Leigh syndrome. The study is being conducted at four centers in the United States: Akron Children's Hospital, Lucile Packard Children's Hospital at Stanford, Seattle Children's Hospital, and Texas Children's Hospital. For more information about trial enrollment please use the contact information below.

Akron Children's Hospital  
 Akron, OH  
 Contact Hilary (Wolf) Tonni at [htonni@chmca.org](mailto:htonni@chmca.org)

Seattle Children's Hospital  
 Seattle, WA  
 Contact Laurie Guidry at [laurie.guidry@seattlechildrens.org](mailto:laurie.guidry@seattlechildrens.org)

Lucile Packard Children's Hospital  
 Stanford University Medical Center  
 Palo Alto, CA  
 Contact Katherine Connors at [kconnors@stanford.edu](mailto:kconnors@stanford.edu)

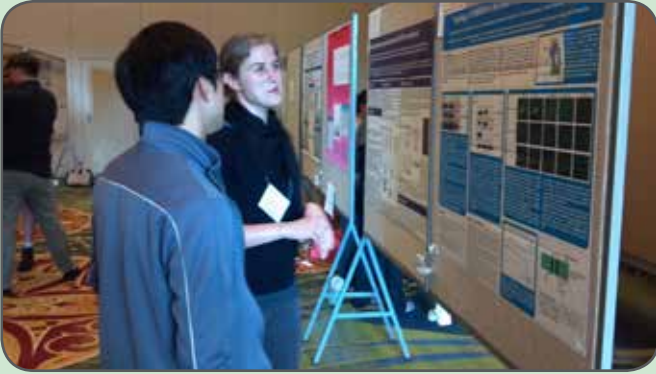
Texas Children's Hospital  
 Houston, TX  
 Contact Catherine Loffredo at [catherine.loffredo@bcm.edu](mailto:catherine.loffredo@bcm.edu)

# UMDF symposium

Providing education and networking.

(Continued from page 1)

## 2013 Scientific Program - June 12 - 15, 2013



*Attendees read some of the posters at the 2012 symposium.*

This activity has been planned and implemented in accordance with the Essential Areas and Policies of the Accreditation Council for Continuing Medical Education (ACCME) and the Accreditation Council for Pharmacy Education (ACPE); through the co/joint sponsorship of Northeast Ohio Medical University and The United Mitochondrial Disease Foundation. Northeast Ohio Medical University is accredited by the ACCME and the ACPE to provide continuing education for physicians and pharmacists.

**The scientific program and the invited faculty are posted on the symposium web page. Call for Abstracts is NOW open and will close on March 31, 2013. All abstracts must be submitted electronically online at [www.umdf.org/callforabstracts](http://www.umdf.org/callforabstracts).**

**For updates, online registration, and travel scholarships, please visit [www.umdf.org/symposium](http://www.umdf.org/symposium) or call 1-888-317-UMDF. Please use code SCI1303 if registering online.**

### Planned Topics Include:

- Mitochondrial Disorders in the 21st Century - Is Treatment Possible?
- The Science Behind Clinical Management
- Mitochondrial Imaging
- Next Gen Reporters of Cellular Metabolism
- Animal Models and Translational Research
- Translational Mitochondrial Medicine
- Bringing New Therapies to Market

**2013 Course Chair: Gregory Enns, M.B., Ch.B.  
Stanford University Medical Center, Palo Alto, CA**

**2013 CME Chair: Bruce H. Cohen, M.D.  
Akron Children's Hospital, Akron, OH**

## 2013 Family Program



*Dr. Russell Saneto talks to attendees at The Doctor Is In at the 2012 symposium.*

### Tentative Topics for 2013:

- Mitochondrial Disease Overview
- Clinical Trials Updates
- Reaching a Diagnosis of Mitochondrial Disease - Past, Present, and Future
- Current Treatments and Therapies
- Genetics of Mitochondrial Disease
- Ask the Mito Doc Panel Discussions – for Adults and Pediatric Patients
- Back by popular demand, our Special Teen Sessions

**For updates, online registration, scholarship, and hotel information, please visit [www.umdf.org/symposium](http://www.umdf.org/symposium) or call 1-888-317-UMDF. Please use code FAM1303 if registering online.**

# 2013 Scientific Registration Form

## Mitochondrial Medicine 2013: Newport Beach

June 12-15, 2013 • Newport Beach, CA

Newport Beach Marriott Hotel & Spa

### FEES (Full Four-day Registration) - Please check only ONE

- \$800 Physician/Researcher  
 \$750 UMDF/MMS/MRS/MIP Members (discount rate)  
 \$450 RNs, Allied Health Professionals/Residents/Fellows\*/Students  
(\*Letter from program director must be received in our office before June 3, 2013, in order to receive this fee.)

Total Full Registration Fee: \$ \_\_\_\_\_

**Can't attend all four days? Choose the days that you can attend with our daily rate. We offer a \$175 daily rate for Scientific/Medical Sessions. Please check ALL that apply.**

Wednesday, June 12  Thursday, June 13  Friday, June 14  Saturday, June 15

\_\_\_\_\_ of days x \$175 = Total \$ \_\_\_\_\_

I would also like to attend the UMDF Friday Night Banquet (included in a four day registration)

\_\_\_\_\_ of people x \$75 = Total \$ \_\_\_\_\_

Total daily rate fees: \$ \_\_\_\_\_

**Full Registration Fee** includes a hard copy syllabus, daily continental breakfast, refreshment breaks, lunch, and the Friday banquet. **Daily Rate Fee** includes all of the above except for Friday's banquet. Payment must be received prior to attendance at symposium. This year, the banquet will be outdoors. We suggest that you plan ahead in case of cool weather.

**\$100 - Registrations made after May 21, 2013, must include a \$100 late fee.**

Total amount enclosed or to be charged: \$ \_\_\_\_\_

Make check payable to: **The United Mitochondrial Disease Foundation (UMDF).**

Or charge the following account:  Visa  MC  Disc  AmEx

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

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

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

**Special Dietary Requirements:**  Vegetarian or  other \_\_\_\_\_

### Please Print Clearly:

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

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

Address \_\_\_\_\_ City \_\_\_\_\_

State/Province \_\_\_\_\_ Country \_\_\_\_\_ Zip/Postal \_\_\_\_\_

E-mail \_\_\_\_\_

Phone Number (\_\_\_\_\_) \_\_\_\_\_ - \_\_\_\_\_ Fax Number (\_\_\_\_\_) \_\_\_\_\_ - \_\_\_\_\_

### SIGN UP NOW ON YOUR PHONE!

Scan this QR pattern on your smartphone and register online. Don't have a QR reader app?

You can download one free from the App Store. Don't have a smartphone?

Register online at [www.umdf.org/symposium/registration](http://www.umdf.org/symposium/registration).

Please use code SCI1303 if registering online.

Otherwise, send completed registration to

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



# 2013 Family Registration Form

## Mitochondrial Medicine 2013: Newport Beach

June 14 -15, 2013 • Newport Beach, CA  
Newport Beach Marriott Hotel & Spa

Last Name \_\_\_\_\_ First Name \_\_\_\_\_  
 Suffix/Degree \_\_\_\_\_  
 Address \_\_\_\_\_  
 City \_\_\_\_\_ State/Province \_\_\_\_\_ Country \_\_\_\_\_ Zip Postal \_\_\_\_\_  
 Phone Number (\_\_\_\_) \_\_\_\_\_ - \_\_\_\_\_ E-mail \_\_\_\_\_  
 Birthdate \_\_\_\_\_

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

**A \$25 late fee will automatically be added to any registrations received after May 28, 2013.**

**FULL REGISTRATION** - Fee includes hard copy of syllabus, two continental breakfasts, two lunches, refreshment breaks, Thursday LHON sessions, and the Friday Night Banquet.

**Fees per person:**

- Individual Registration by May 28, 2013 \$ 225
- Family Registration by May 28, 2013 (2 adults, same household) \$ 400
- Additional Friday Night Banquet tickets (for family NOT registered for the conference)  
(\$65 per person x \_\_\_\_\_) \$ \_\_\_\_\_
- LHON ONLY** (lunch and sessions) \$ 50

*Additional lunches may be purchased prior to the symposium for family or friends NOT registered.  
E-mail [symposium@umdf.org](mailto:symposium@umdf.org) for payment/ordering options.*

**TEEN REGISTRATION IS FREE BUT YOU MUST REGISTER!**

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

Credit card payees may fax their form to UMDF at 412-793-6477. Once faxed, DO NOT mail the original registration form.

**Questions?**  
Call 1-888-317-UMDF

**Would you like to make an additional donation?** \$ \_\_\_\_\_

**Total amount enclosed or to be charged:** \$ \_\_\_\_\_

Make check payable to **UMDF** in U.S. currency on a U.S. bank.

Or charge the following account:      Visa              MC              Disc              AmEx

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

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

Signature \_\_\_\_\_

*(invalid without signature)*

The UMDF would like to provide assistance to conference attendees who may have special needs as described below.

**Assistive Listening Devices** - If you have a hearing impairment, we can provide you with an Assistive Listening Device (ALD) that will enable you to hear the speakers clearly. The ALD has a headset that receives sound waves from the speaker's microphone that go directly into the user's ears.

- Yes, I would like an ALD.
- No, my needs are more demanding.

**Scooters** - The UMDF will secure scooters for those adults who need assistance and cannot bring their own scooters. A credit card is required to reserve a scooter.

- Yes, please reserve a scooter in my name.

**Special Dietary Needs:**

- I require vegetarian meals
- Other - contact us about special meal requests





# United Mitochondrial Disease Foundation LEAP Award

## Living, Encouraging, Achieving & Persisting

**Purpose:** To recognize an individual living positively with mitochondrial disease, highlighting the person's accomplishments and volunteer service.

**Eligibility:** Age 14 years or older.

**Criteria:** Individual with confirmed or suspected mitochondrial disease who overcomes daily challenges to achieve goals in career, family, and volunteer service. The individual demonstrates a positive attitude, hope for a brighter future, and an enthusiasm that inspires others.

**Instructions:** Any UMDF member can nominate an individual for this award. Fill out the form below and attach the requested information. The LEAP Award winner will be featured on the UMDF website and recognized in the UMDF newsletter.

In 100 words or less, please explain how this individual overcomes daily challenges to achieve goals in career, family, and volunteer service. Please provide examples of how the individual demonstrates a positive attitude, hope for a brighter future, and an enthusiasm that inspires others. You may also attach copies of articles about the nominee and lists of projects, activities, or clubs the nominee is involved with.

Nominations may be submitted online at [www.surveymonkey.com/s/LEAPAward2013](http://www.surveymonkey.com/s/LEAPAward2013) or type your essay and attach it to the nomination form.

Mail the nomination by April 15, 2013 to:

United Mitochondrial Disease Foundation  
Attn: LEAP Award  
8085 Saltsburg Road, Suite 201  
Pittsburgh, PA 15239

Or fax to 412-793-6477 or e-mail the nomination by April 15, 2013 to [info@umdf.org](mailto:info@umdf.org).

## Person Nominating

Name: \_\_\_\_\_

Address: \_\_\_\_\_

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

E-mail: \_\_\_\_\_

## Nominee for the Award

Name: \_\_\_\_\_

Address: \_\_\_\_\_

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

Diagnosis (if known): \_\_\_\_\_

Age (must be at least 14 years old): \_\_\_\_\_

Please remember to attach your 100 word nomination narrative to the application.

The winner will be announced at the symposium on Friday, June 14, 2013, at the awards ceremony.

# United Mitochondrial Disease Foundation Heartstrings Award

## Recognizing a youth commitment that tugs on the heartstrings

**Purpose:** To recognize a child or teen who has donated or raised funds for the UMDF, enabling the UMDF to continue its mission.

**Eligibility:** The individual recognized must be under 18 years of age at the time of the donation or fundraising activity.

**Criteria:** The winner is chosen based on related criteria of age, time invested, talents demonstrated, effectiveness, and generosity. For nominees who implement fundraising projects, the judges will consider the uniqueness and creativity of the project, communication, time invested and the amount raised in comparison to the age of the individual. For nominees who donate funds, the judges will consider the generous spirit shown, communication, and amount donated in relation to the age of the individual.

**Instructions:** Any UMDF member can nominate an individual for this award. Fill out the form below and attach the requested information. The Heartstrings Award winner will be featured on the UMDF website and recognized in the UMDF newsletter.

In 100 words or less, please explain how this individual has "tugged at your heartstrings" through fundraising for or donating to the UMDF. Identify important features of the nominee's activity, such as the time invested, creativity, communication skills, determination, effectiveness, and generosity. You may also attach supporting information on the fundraising project (published articles, pictures, comments from others involved with or participating in the project) or the communications of the nominee (letter explaining intended use of the gifted funds, thank you letters, letters sent with the donation, and so forth).

Nominations may be submitted online at [www.surveymonkey.com/s/HeartstringAward2013](http://www.surveymonkey.com/s/HeartstringAward2013) or type your essay and attach it to the nomination form.

Mail the nomination by April 15, 2013 to:

United Mitochondrial Disease Foundation  
Attn: Heartstrings Award  
8085 Saltsburg Road, Suite 201  
Pittsburgh, PA 15239

Or fax to 412-793-6477 or e-mail the nomination by April 15, 2013 to [info@umdf.org](mailto:info@umdf.org).

## Person Nominating

Name: \_\_\_\_\_

Address: \_\_\_\_\_

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

E-mail: \_\_\_\_\_

## Nominee for the Award

Name: \_\_\_\_\_

Address: \_\_\_\_\_

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

Diagnosis (if known and if applicable): \_\_\_\_\_

Age (must be less than 18 years old at time of donation or event): \_\_\_\_\_

Please remember to attach your 100 word nomination narrative to the application.

The winner will be announced at the symposium on Friday, June 14, 2013, at the awards ceremony.

# United Mitochondrial Disease Foundation Energy Award

## Energy

**Purpose:** To recognize an individual who embodies the spirit of the UMDF and its Mission...*promoting research and education for the diagnosis, treatment, and cure of mitochondrial disorders and providing support to affected individuals and families.*

**Eligibility:** Anyone

**Criteria/Instructions:** Any UMDF member can nominate an individual for this award. Fill out the form below and attach the requested information. The Energy Award winner will be featured on the UMDF website and recognized in the UMDF newsletter.

In 100 words or less, please explain how this individual embodies the spirit of the UMDF and its Mission. You may also e-mail supportive documents of your nomination to [info@umdf.org](mailto:info@umdf.org) (please put Energy Award Nominee and their name in the subject line) -- listing projects, activities, or other information that highlights their efforts in supporting the UMDF Mission. You may also send or e-mail copies of articles about the nominee and lists of projects, activities, or other information that supports your nomination.

Nominations may be submitted online at [www.surveymonkey.com/s/EnergyAward2013](http://www.surveymonkey.com/s/EnergyAward2013) or type your essay and attach it to the nomination form.

**Mail the nomination by April 15, 2013 to:**

United Mitochondrial Disease Foundation  
Attn: Energy Award  
8085 Saltsburg Road, Suite 201  
Pittsburgh, PA 15239

Or fax to 412-793-6477 or e-mail the nomination by April 15, 2013 to [info@umdf.org](mailto:info@umdf.org).

## Person Nominating

Name: \_\_\_\_\_

Address: \_\_\_\_\_

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

E-mail: \_\_\_\_\_

## Nominee for the Award

Name: \_\_\_\_\_

Address: \_\_\_\_\_

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

Diagnosis (if known and if applicable): \_\_\_\_\_

Age: \_\_\_\_\_

Please remember to attach your 100 word nomination narrative to the application.

The winner will be announced at the symposium on Friday, June 14, 2013, at the awards ceremony.

# UMDF support

Connecting our community.

## UMDF Support Events

**December 1, 2012.** A Holiday Breakfast with a special visit from Santa was held at The Peachtree Club in Atlanta, GA. The event was sponsored by TJ Maxx. A great time was had by all 150 people who attended the event. A special thank you to Atlanta Chapter Members Chris and Mary Swinn and Nancy McCue of TJ Maxx for organizing this event.

**December 8, 2012.** A Holiday Party was held in the Detroit area for Michigan Mito families. Over 40 people attended the party. The event was organized by Missy Leone and Julie Scott, with a special thanks to Cindy and Vic Bitel for scheduling Santa to visit the kids!

**December 9, 2012.** The Chicago Chapter held their annual holiday party in Oak Brook, IL. Approximately 35 UMDf members attended the event.

**December 29, 2012.** Dr. Vamsi Mootha, Professor of Systems Biology and of Medicine at Harvard Medical School, gave a special presentation to Dallas, Texas, members on new techniques and discoveries from his research lab.

**January 6, 2013.** The Annual Indiana Chapter Night at the Children's Museum was held in Indianapolis, IN. Close to 100 attended the event, enjoying dinner and family-fun at the Museum's exhibits. Thank you Jennifer Demotte, Jackie Parrish, and Darla Cohen for organizing the event!

**January 12, 2013.** Twenty-eight UMDf volunteers from the Minneapolis/St. Paul area represented the UMDf at the Young Brain Carnival at the Mall of America. Anne Simonsen, Great Lakes Regional Coordinator, was in attendance along with the Mighty Mito and "Mini" Cheerleaders. Attendees received information about the UMDf and mitochondrial disease.

**January 26, 2013.** Trisha Kranz and her daughter, Anastasia, organized and held the first Mito Muffins Support Breakfast in The Woodlands, Texas. Look for more muffins and support in The Woodlands throughout 2013!

**February 7, 2013.** A Patient and Family Meeting was held at the University of South Florida in Tampa, FL featuring speaker Dr. Bruce Cohen of Akron Children's Hospital. Dr. Cohen presented an update on mitochondrial medicine and answered many questions from the patients and families that attended. More than 80 people attended the event. Thank you Chapter Members Cecilia Coloret and Marla Tobia for helping out with this event.

**February 12, 2013.** A Patient and Family Meeting was held at Mercer University in Atlanta, GA. Dr. Bruce Cohen of Akron Children's Hospital was the presenter. Dr. Cohen gave an overview of mitochondrial medicine and answered many questions by the 60 people who attended this event. Thank you Atlanta Chapter Members Amy Blackwell, Sebastien Cotte, and Mary Beth Morris for making this wonderful event happen. A special thank you to Dr. Stanley Pollock and Mercer University for helping with this event.

**February 17, 2013.** The Ohio Chapter held a Mini-Symposium at Akron Children's Hospital. Over 80 medical professionals and UMDf families attended the event. Dr. Sumit Parikh, Dr. Bruce Cohen, Dr. Douglas Kerr, and Dr. Amy Goldstein presented a variety of topics and provided research updates to the attendees. The event concluded with a special presentation from Ari and Stacy Goldberg, the premier of the movie trailer for "The Magic Bracelet," a film conceptualized by their late daughter, Rina Goldberg. A special thanks to Dr. Cohen and his staff for their help in organizing the event.

**February 18, 2013.** Approximately 45 UMDf members attended the Cincinnati Support Group Meeting at Cincinnati Children's Hospital. The attendees received a welcome from Dr. Gregory Grabowski, Director of the Division of Human Genetics,

and a presentation from Dr. Taosheng Huang, Director of the Mitochondrial Diseases Program.

**February 23, 2013.** A Special Educational Meeting was held in Durham, NC featuring Medical Geneticists Dr. Muge Calikoglu of UNC Chapel Hill and Dr. Dwight Koeberl of Duke University. The physicians gave presentations to the 25 attendees on mitochondrial disease diagnosis and treatment. A question and answer session followed. Thank you Adriana Smith and Jenny Hobbs for organizing this event!



*The Mighty Mito Cheerleaders are in the back row, posing at the Young Brain Carnival: Megan Elston, Mada Tautges, and Korbin Kvaas. The "Mini" Mighty Mito Cheerleaders are in the front row: Sophia DiPaola, Linae Larson, and Kathryn Simonsen*

### Create Your Own Support Event

Interested in creating  
your own support event?

Contact Melinda O'Toole in the  
UMDF Member Services Department  
by telephone, toll-free, at 1-888-317-8633  
or via e-mail at [melinda@umdf.org](mailto:melinda@umdf.org).

# Do You Have Some Form of Medical Identification?

Liz Kennerley said, "I updated my MedicAlert® information and for the first time noticed a more comprehensive list of mitochondrial diseases. When I first got my bracelet, it wasn't even recognized by the company, now they have many of the complexes, as well as many of the metabolic issues we have as a result! Way to go us [UMDF] for raising awareness."



MedicAlert® ID bracelet

MedicAlert® Foundation is a non-profit organization that provides 24/7 emergency response service with a team of medically trained specialists that will speak on your behalf and relay your health information to emergency responders and medical personnel, as well as loved

ones. Some of the items that MedicAlert® offers members are customized medical identification bracelets and emergency response cards. For details, go to [www.medicalert.org](http://www.medicalert.org).

CARE Memory Wrist Band offers electronic medical ID bracelets that can hold an entire medical history and can be plugged into a USB port to download information onto a computer. Go to [www.carememoryband.com](http://www.carememoryband.com) for details.

Road ID sells ID wrist bands for outdoor athletes but is recommended by a mito mom. The interactive version allows users to access and update emergency response profile information online. Go to [www.roadid.com](http://www.roadid.com) for details.

The United Mitochondrial Disease Foundation (UMDF) provides these type of resources on its website. Go to [www.umdf.org](http://www.umdf.org) and look under "Find Support" and "Resource Guide."

**Disclaimer:** Inclusion of particular professionals, services, products, or organizations does not imply endorsement by the UMDF.

## Free Camp for Children with Life-Threatening Medical Conditions

Camp Korey, located in Carnation, Washington, is a non-profit organization, which provides camping programs to children living with chronic or life-threatening medical conditions free of charge. The children are between the ages of 7-17, who are living with illnesses such as skeletal dysplasia, metabolic disorders, cranio-facial differences, organ transplants, and other blood-borne illnesses. The camping programs include activities such as arts and crafts, swimming, horseback riding, team building, and more with the goal of allowing them to simply let loose, have fun, and be kids. According to Emma Thier, Camper Recruiter, Camp Korey works with organizations that will fly kids in for free! For more information about Camp Korey, contact Emma at 425-844-3226 or at 949-701-0059. You can also view the Camp Korey website, website: <http://campkorey.org/camper-programs/apply>.

### 2013 Summer Schedule for Children with Mitochondrial Disease

*Each session serves 50 campers.*

- Session 4 July 15 -19 - Metabolic Bone Conditions/Mitochondrial Disease/General Conditions
- Session 6 July 29 - Aug 2 - Mitochondrial Disease/General Conditions
- Session 7 Aug 5 - 9 - Friends of the Craniofacial Center/Mitochondrial Disease/General Conditions

There are other facilities that offer camps for special needs campers. Check them out at [www.umdf.org](http://www.umdf.org). Go to "Find Support" and click on "Resource Guide."

## Ways to Help Those with Mitochondrial Disease

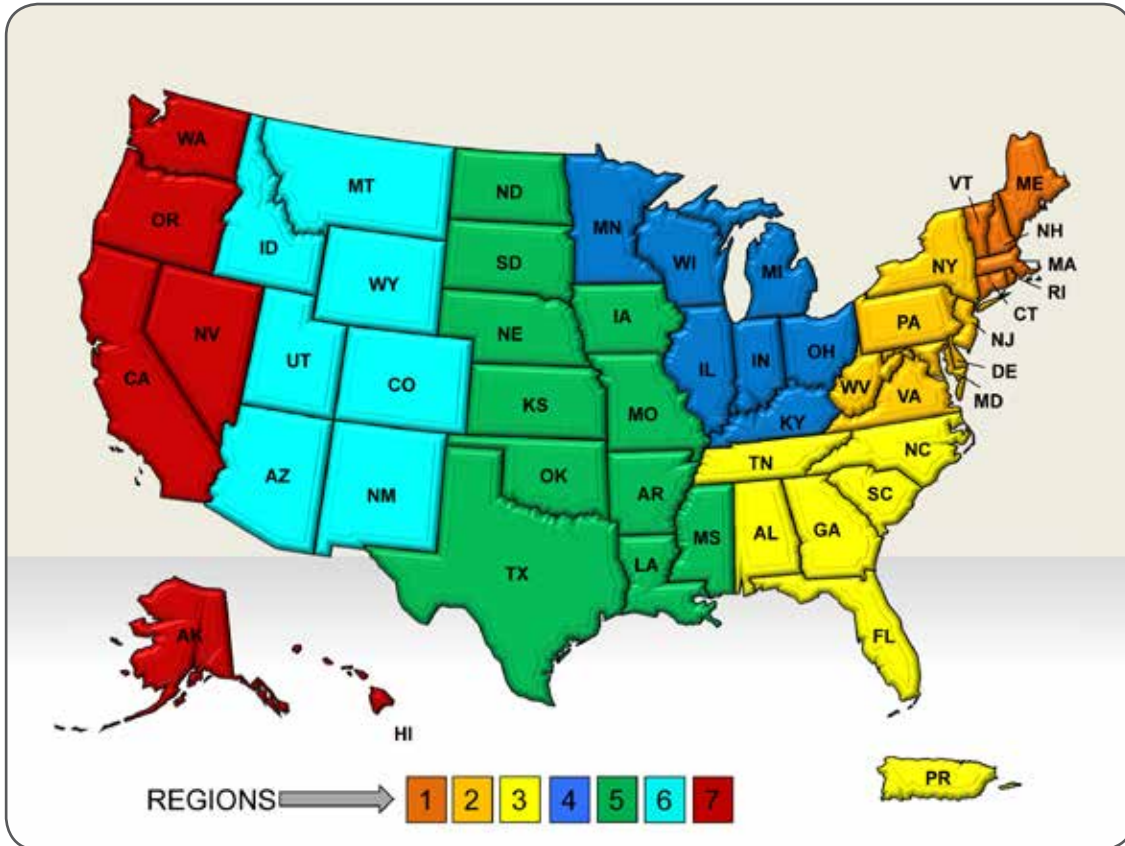
Do friends and family members want to know how to help you? Perhaps you want to help someone who has mitochondrial disease and don't know how best to help them. After a recent post about this on Facebook, the suggestions listed below were made.

- Ask the family if there are specific restaurants that can accommodate their dietary needs and purchase gift cards from there.
- Ask the individual or family if they need anything when you are going to the grocery store, especially during the winter months. Gift cards to grocery stores are also helpful.
- Buy gift cards for popular restaurants that deliver, that way they can be used at home or at the local hospital.
- Purchase gas cards for that emergency run to the hospital when funds are low and gas is short.
- Gift cards to the hospital cafeteria are helpful. Parents often forget to eat and it's very expensive to eat at the hospital.
- Gift cards for supplements or Dr. office co-pays are useful.
- Offer to run errands, do housework or yardwork, or walk their dog.
- Make a gift basket for the caregiver at the hospital and include change for the vending machines, purse size hand sanitizer, a puzzle book or novel, note cards, postage, and a gel pen.
- Offer to drive your friend or family member to an appointment; that way they can attend to their child and it will offer them companionship on long drives.
- Make goodie bags or cards for loved ones in the hospital or who are sick at home.
- If you work at the same company as a caregiver, ask the Human Resources Department if you can donate some of your banked sick hours to them.
- Give cards that say, "thinking of you" or "we care." Often families with mito kids spend a lot of time in hospitals and feel very alone.
- Visit them with their favorite snack and pray for them.



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, MASSACHUSETTS, NEW HAMPSHIRE, RHODE ISLAND**  
Contact the National Office to Connect

**Northeast Region (2)**

**DELAWARE**  
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 Slempp, 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 Slauter, 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 Edmundson, Evansville Area Support Group  
Lori Meyer, Evansville Area Support Group  
Camille Gaughan, Indianapolis Area  
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 Penninsula  
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

### **MISSISSIPPI**

Tracy Shedd, Vicksburg

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

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

### **TEXAS**

Manuel Castro, Austin  
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

### **NORTH DAKOTA, SOUTH DAKOTA, & MONTANA**

Marty Campbell, Beach, ND

Contact the National Office to connect

### **ARIZONA**

Gina Blair, Peoria

### **IDAHO**

Jennifer Pfefferle, Boise, Idaho Support Group

### **NEW MEXICO**

Stephanie Cassady, Albuquerque

### **UTAH**

Laura McCluskey, Orem

### **COLORADO, 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 Freiling, Monmouth Support Group

### **WASHINGTON**

Joy Krumdiack, NW Washington

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

Contact the National Office to Connect

### **INTERNATIONAL**

Rob Ryan, Australia  
John Carreiro, British Columbia  
Nilam Agrawal, India  
Saijad 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*

## **The Adult Advisory Council Team Continues to Grow**

Sharon Shaw, the Adult Advisory Council Team (AACT) Chairman, is pleased to announce its newest members: Kirah Fasano, Joy Krumdiack, and David McNees.



*Kirah Fasano*

Kirah Fasano lives in Doylestown, Pennsylvania, and was diagnosed with Chronic Progressive External Ophthalmoplegia (CPEO) in 2010, but she has been living with Mitochondrial Myopathy for 13 years.



*Joy Krumdiack*

Joy Krumdiack lives in Bellingham, Washington. She was diagnosed with mitochondrial disease in 2000, and she is the UMDF Ambassador for Northwest Washington.



*David McNees*

David McNees lives in Northeastern Ohio. In 2011, he was diagnosed with Complex IV Deficiency, Mitochondrial Myopathy, Neuropathy, and Dysautonomia. He is a Montessori teacher who is very excited about joining the AACT.

Shaw stated, "We have updated our AACT pages on the UMDF website. We always

welcome ideas from our adult mitochondrial community for any resources and services that some of you may know about, that are not already listed. Our goal on our AACT website pages is to have a growing and comprehensive library to assist the newly diagnosed and those of us that have been navigating this disease and its many complexities."

She continued, "Thank you to those that have given feedback regarding our annual symposium. We have incorporated many of your ideas and have added 'adult' sessions and topics during our symposium. Each year the number of 'adult issues and topics' grow. We also look for your stories and ask you to please send them in. Describe your journey getting a diagnosis and how you live life dealing with mitochondrial disease (500 to 700 words). There is nothing more comforting than for one adult mitochondrial patient to share with another... as we all travel this road." Feel free to e-mail your story to [aact@umdf.org](mailto:aact@umdf.org).

AACT is not a support group, but an advisory council. The purpose of the AACT is to represent and serve the unique needs of the affected adult community and to ensure that those needs are adequately represented to the United Mitochondrial Disease Foundation (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.

The group convenes on a conference call about four to five times per year and meets in person once a year at the UMDF's annual symposium. If being a member of AACT sounds like something that you would be interested in, contact Sharon Shaw by telephone at 602-692-0001 or via e-mail at [shshaw@aol.com](mailto:shshaw@aol.com).

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

Sharon Shaw, 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.umdff.org](http://www.umdff.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: Fran Kendall, M.D., Virtual Medical Practice in Atlanta, GA and Northside Alpharetta Medical Campus in Alpharetta, GA and Jerry Vockley, M.D., Ph.D., Children's Hospital of Pittsburgh, Pittsburgh, PA.

**The Question is...** Is there any reason that a child would be Carnitine deficient that has nothing to do with Mito? In other words, my child had a low test & retest. We supplement him and his energy & cognition is greatly improved. He used to lie on the floor a lot, and when tired or sick he was 'limp' and acted drunk and talked very slow (for example it would take him about 5 seconds to say a one syllable word). Carnitine changed all of this. I wonder if this is a clear mito case, or if a child can have low carnitine as an isolated issue.



**Fran Kendall, M.D.**

**Response from Fran Kendall, M.D.**

While low carnitine values can be and are often seen with mito, carnitine deficiency can also occur for other reasons ranging

from primary carnitine deficiency, another inborn error of metabolism, GI absorption problems, and for dietary reasons (specifically, the ingestion of a diet low in high carnitine-containing foods). If someone responds well to carnitine supplementation, looking into primary carnitine deficiency may be a consideration depending on previous studies and clinical features.

## ASK THE MITO DOC<sup>SM</sup>

**The Question is...** I have mitochondrial myopathy and have been having some problems with my dysautonomia. My blood pressure gets as low as 70/44 while up and moving around at work. (I am a registered nurse in cardiology.) My cardiologist wants to start me on Florinef but I am leery. Is it a safe drug for mito patients? Another question I have is I have been extremely cold lately.

I am not ill and have no idea what my blood pressure is running. It will be 75 degrees in my house and I will be cold to the bone. Is that the dysautonomia?

**Response from Jerry Vockley, M.D., Ph.D.**

Florinef has been used safely in patients with mitochondrial-induced dysautonomia. Midodrine (generic name proamatine) is also frequently used, often as a first line treatment depending on other aspects of your clinical condition. Hot and cold flashes and temperature intolerance are common in mitochondrial disease. It may be improved by increasing your blood pressure but it also may not. The blood vessels of the skin that control your body temperature can have abnormal regulation that is independent of blood pressure and still lead to temperature instability. Unfortunately there are no good treatments for this problem except adjusting your environment.



**Jerry Vockley, M.D., Ph.D.**

### DID YOU KNOW?

You can quickly and easily find "Ask the Mito Doc" Q/As on topics of your choice by going to the UMDF home page at [www.umdff.org](http://www.umdff.org). Go to "Find Support" and click on "Ask the Mito Doc." Click on the search "Ask the Mito Doc" link; this will pull up a search box. Type in a keyword or phrase you are interested in and click "Go." The search engine will pull up every Q/A that mentions your word or phrase. If you are not satisfied with the results, try variations or synonyms of your word/phrase.

## **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. Perhaps you can get a 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>
4/15/2013	Family Meeting	East Tennessee Children's Hsp., Knoxville, TN
4/16/2013	Grand Rounds	East Tennessee Children's Hsp., Knoxville, TN
5/2/2013	Family Meeting	Univ. of Nevada School of Medicine, Las Vegas, NV
5/3/2013	Grand Rounds	Univ of Nevada School of Medicine, Las Vegas, NV
5/20/2013	Family Meeting with guest speaker, Dmitriy Niyazov, M.D.	(location to be determined - New Orleans, LA area)
5/20/2013	Grand Rounds	Tulane Univ. School of Med., New Orleans, LA
6/26/2013	Family Meeting	A I Dupont Hospital for Children, Wilmington DE
6/26/2013	Grand Rounds	A I Dupont Hospital for Children, Wilmington DE
7/24/2013	Family Meeting	Texas State University, San Marcos, TX
7/25/2013	Grand Rounds	Texas State University, San Marcos, TX
8/7 or 8/8/2013	Family Meeting	Location to be determined in Minnesota
8/9/2013	Grant Rounds	Mayo Clinic Childrens Center, Rochester MN

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

## **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 would like the UMDF to reach out to them, please let us know by calling the UMDF National Office, toll-free, at 1-888-317-8633.

# In the Clinic

In the last issue of UMDF Connect, we premiered “In the Clinic...” In this edition, we take you to the Children’s Hospital of Philadelphia (CHOP) Mitochondrial-Genetics Diagnostic Clinic. Board certified in Pediatrics and Clinical Genetics, Marni J. Falk, M.D., directs the CHOP Mitochondrial-Genetics Diagnostic Clinic to aid in the evaluation of individuals with suspected mitochondrial disease. She is actively involved in developing improved diagnostic approaches for mitochondrial disease, including whole exome sequencing approaches in a translational research laboratory at CHOP that investigates the global metabolic consequences of mitochondrial disease, and targeted pharmacologic therapies, in *C. elegans*, mouse, and human tissue genetic models. Dr. Falk, M.D., has been an Assistant Professor in the Division of Human Genetics, Department of Pediatrics at CHOP and at the University of Pennsylvania (Upenn) Perelman School of Medicine since 2006. She is the Chair of the United Mitochondrial Disease Foundation (UMDF) Scientific and Medical Advisory Board and she is on the UMDF Board of Trustees.



Marni Falk, M.D.

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

I completed training as a Clinical Geneticist and Pediatrician in Cleveland, Ohio, at Rainbow Babies and Children’s Hospital. During that period, I had regular exposure to very ill patients with suspected mitochondrial disease both in the clinic, the inpatient wards, and in the diagnostic laboratory setting at the Center for Disorders of Energy Metabolism under the direction of Drs. Charles Hoppel and Douglas Kerr. I was intrigued with the breadth and range of disease manifestations patients experienced, the complexity of their biochemical findings, the limited number of “definitive” genetic-based answers that could be determined for individual patients, and the clear need for improved therapies to alleviate the symptoms and prevent the progressive disease course in mitochondrial disease. I remember one young child, in particular, whom we would visit regularly in an intensive care unit at a community hospital, where she lay for months on a ventilator from which she was unable to be weaned due to a loss of her natural respiratory drive from progressive brainstem strokes caused by her unspecified mitochondrial disease. She was awake and able to understand what was going on, recognized and seemed comforted by looking at and holding hands with her family, but could not talk and eventually became so weak that she simply could not continue to live like that and the ventilator had to be stopped. Her parents never knew the underlying cause of her disease.

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

Yes, we are happy to evaluate patients of all ages in our Mitochondrial-Genetics Diagnostic Clinic. Approximately three-quarters of our patients are children and one-quarter are adult. Often, we evaluate or follow multiple generations in the same family.

## 3. Have you made any discoveries in research?

Yes. We have identified a conserved cellular signature of mitochondrial respiratory chain disease that exists across evolutionary species from worms to mice to humans. This signature involves specific biochemical and gene expression changes that affect entire pathways. Using this knowledge, we have begun to identify drugs that can target the common biochemical and gene expression changes to improve the health outcome of the model animals. For example, we have studied a drug called probucol, which was previously used in people to treat elevated cholesterol, in a mouse model of lethal kidney disease that is caused by their genetic defect that prevents the biosynthesis of Coenzyme Q, which is necessary for the respiratory chain to function and make energy. Remarkably, probucol could prevent renal disease in these mice if started pre-symptomatically, and could even significantly reverse kidney disease if started after disease manifestations had begun. We already know that probucol lead to an increase in the Coenzyme Q content within the animals’ tissues and reversed some of the changes in the central signaling pathways that

respond to respiratory chain disease. However, we are still working to understand all of the reasons why this drug is so effective in this model, and to further understand whether it is safe and may be of potential benefit in other causes and for other manifestations of mitochondrial disease. There are multiple drugs that affect related pathways that we are also studying in mouse and worm models of mitochondrial disease.

We have also discovered the genetic basis for multiple patients’ suspected mitochondrial disease. Sometimes, this information conclusively demonstrates that a patient’s problems are actually not due to a mitochondrial disease but instead clarifies the precise cause of their complex problems. Other times, we have identified a novel gene that is the cause of their mitochondrial disease. We have done this for isolated medical problems such as blindness, and also for more complex mitochondrial diseases that involve multiple systems.

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

Our clinic evaluates approximately 80 to 100 new patients each year for suspected mitochondrial disease, and we follow-up a similar number of patients for management of known mitochondrial disease or for continued evaluation of suspected mitochondrial disease.

## 5. Are their openings for new patients?

Yes, we are happy to evaluate patients of all ages in the CHOP Mitochondrial-Genetics Diagnostic Clinic. Interested patients should contact our Clinic Coordinator, Elizabeth McCormick, MS, CGC, at 267-426-9650. We strive to see all interested patients, but given our limited staffing we typically have a several months’ wait.

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

We are working to build a multi-disciplinary clinic at CHOP in which the patient can be evaluated on the same day by a team of physicians with diverse expertise in their individual manifestations of mitochondrial disease.

Our goal is to diagnose and understand each patient’s individual disorder. Once diagnosed, we would like to facilitate their optimal clinical care. In addition, we can enroll interested patients with known or suspected mitochondrial diseases in the North American Mitochondrial Disease Consortium (NAMDC) registry and biorepository, which will facilitate understanding of disease course across many types of mitochondrial diseases, and be useful to identify patients for specific clinical trials. We are currently enrolling undiagnosed patients with suspected mitochondrial disease into an NIH-sponsored whole-exome sequencing research study we are conducting at CHOP, which involves patients in the process of tailoring this powerful diagnostic analysis tool to the needs of the mitochondrial disease community. Ultimately, we plan to initiate clinical research trials of promising therapies identified in our cell and animal model studies in mitochondrial disease patients with clearly understood genetic causes for their disorders, in conjunction with NAMDC and the CHOP Center for Mitochondrial and Epigenomic Medicine (CMEM).

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

The Mitochondrial-Genetics Diagnostic Clinic is currently a focused effort of Dr. Marni Falk and a dedicated Genetic Counselor, Ms. Elizabeth McCormick, MS, CGC. We have a monthly Clinical Mitochondrial Disease Multi-Disciplinary Conference held in conjunction with the CHOP Center for Mitochondrial and Epigenomic Medicine (CMEM) where we review complex cases to discuss further evaluation and treatment strategies.

## Fundraisers Benefiting the UMDF

**November 10, 2012.** The family of Rachel Kahoe held a benefit event in honor of Rachel and her battle with MELAS (mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes). The first annual event raised nearly \$13,000 for the UMDF in Rachel's memory to go toward research. A very special thank you to Rachel's brother, Rodney Jackson, for organizing such an amazing event!

**November 28, 2012.** Joe Auth and Molly Manning held a happy hour event in Boston, MA in honor of their brother, Charlie Auth. The event was attended by friends and family of the Auth and Manning families and raised nearly \$2,000. Thank you Joe and Molly for putting on such a great event!

**January 2013.** The students at St. Bernadette's Roman Catholic School in Monroeville, PA held their annual Coins for a Cure fundraiser, which they call the "Gina Jug." The annual event is held in memory of Gina Mohan who was a student at the school. Thank you to the staff and students who raised over \$1,600 for the UMDF this year!

**January 25, 2013.** The Charlotte Checkers held a special night for Savannah Greenawalt entitled, Saving Savannah. Mito facts were displayed throughout the hockey arena, and proceeds from tickets sales of the game were donated to the UMDF. Highlights from the event include: Savannah signing autographs for fans and dancing with Cinderella, Zander Shields, age 7, from Charlotte, NC came out with the hockey players, and mito families did the chicken dance. Over \$4,500 was donated to the UMDF. Thank you to the Greenawalt family for your continued support!

**February 2, 2013.** The Energy for Life Walkathon in Houston, TX was held on a beautiful spring day! The event brought out over 550 excited walkers who raised over \$75,000 for the UMDF in honor of their loved ones. Thank you to the walk committee for all of your hard work! We can't wait to see you all next year!



*Team Energy for Sarah poses for a photo at the Energy for Life Walkathon in Houston, TX. Sarah James is on the left, her best friend is on the right, and her friends and family are behind her. Photo used with permission from Halcyon Claire Photography.*

## Gifts from the Heart

**September 2012.** Trisha and Jim Mowrer of Pottstown, PA held their annual Horseshoe Tournament and Raffle in memory of their son, Nathan D. Mowrer. Thank you Mowrer family and friends for your support of the UMDF!

**September 2012.** The Brittany Wilkinson Research Fund with the UMDF held a fundraising night at Chevy's Restaurant during Awareness Week 2012. The event raised over \$50 in memory of Brittany. Thank you to the Wilkinson family for your continued support!

**September 2012.** A Jeans for Jessica event was held at Blue Cross Blue Shield of South Carolina in the month of September. The event was organized by Wendy Smith in memory of her daughter Jessica. A special thank you to Wendy and Blue Cross Blue Shield of South Carolina for raising over \$1,200 for the UMDF in Jessica's memory.

**December 19, 2012.** Kasey McDaniel and her family sold energy band bracelets in honor of Owen Lee Ruffner. Over \$701 was raised for the UMDF!

**December 2012.** Rachael Pipp, who has mitochondrial disease, sold candy canes in the month of December and donated \$111 to the UMDF. She also organized a snowflake fundraiser and donated \$44. Thank you for participating in the season of giving and thinking of the UMDF!

**January 2013.** Jason, Jessica, and Alyssa Jacobs held an event in Plano, TX in memory of their son and brother, Caleb. "Remembering Caleb" was held at Plano West. The event raised money for Caleb's research fund that was set up by his family. Thank you to all who participated!

**January 2013.** A pedicure-of-the-month special was held at polished by Clair V nail boutique and spa in Roanoke, Virginia with proceeds benefiting the UMDF. A special thank you to Emily Goldberg for organizing this event in honor of her daughter, Olivia Paige Goldberg.

# Upcoming Events to Benefit the UMDF

**March 23, 2013.** The fourth annual Jackson - Culley Mito What 5k will be held in Millington, TN at USA Stadium. For details, contact Angie Hayes at [angie.hayes07@gmail.com](mailto:angie.hayes07@gmail.com) or visit [www.mitowhat5k.blogspot.com](http://www.mitowhat5k.blogspot.com).

**March 24, 2013.** Think Spring! Bruster's of Ingomar on Perry Highway in Pittsburgh, PA will host an Easter Egg Hunt with sales from the silent auction being donated to the UMDF. For more information, contact [brustersofingomar@gmail.com](mailto:brustersofingomar@gmail.com).

**April 6, 2013.** The Middle Tennessee Chapter will be holding their third annual Energy for Life Walkathon at Centennial Park in Nashville, TN. We will be at a new venue this year and looking forward to a great event! Register your team today at [www.energyforlifewalk.org/nashville](http://www.energyforlifewalk.org/nashville).

**April 13, 2013.** The California Chapter will be hosting their second annual Energy for Life Walkathon at Golden Gate Park in San Francisco, CA. Please consider starting or joining a team today! For information, visit [www.energyforlifewalk.org/sanfrancisco](http://www.energyforlifewalk.org/sanfrancisco).

**April 13, 2013.** The tenth annual Bet on Baylee event will be held at the Roseville Community Center in Roseville, OH. This annual event is held in honor of Baylee Thompson and features a Texas hold'em tournament, a cornhole tournament, silent auctions, and tons of fun. Contact Jody Thompson at [buff2506@hotmail.com](mailto:buff2506@hotmail.com) for more information!

**April 15, 2013.** Please join us for the second annual Carson's Classic golf tournament at the University of Georgia Golf Course in Athens, GA. The Dawgs for Mito collegiate chapter of the UMDF is hosting the event. All proceeds will benefit the UMDF in honor of Carson Coburn. For more information on registering or sponsoring the event, go to [www.carsonclassic.org](http://www.carsonclassic.org).

**April 20, 2013.** The St. Louis Metro Area Mito Group will be holding their third annual Energy for Life Walkathon at Tower Grove Park in St. Louis, MO. Join them and help raise funds to find a cure. For details, visit [www.energyforlifewalk.org/stlouis](http://www.energyforlifewalk.org/stlouis).

**April 20, 2013.** The Atlanta Chapter will be holding their second annual Energy for Life Walkathon at Centennial Olympic Park in Atlanta, GA. Please join members of the chapter to make their walk a success! For additional information, go to [www.energyforlifewalk.org/atlanta](http://www.energyforlifewalk.org/atlanta).

**April 27, 2013.** The Binghamton Group will be holding their second annual Energy for Life Walkathon at Otsiningo Park in

Binghamton, NY. Please join the group for a fun filled day of awareness activities and networking. For more information, go to [www.energyforlifewalk.org/binghamton](http://www.energyforlifewalk.org/binghamton).

**May 4, 2013.** The Evansville Walk Committee is excited to announce the second annual Energy for Life Walkathon, at Burdette Park in Evansville, IN. Please join us again this year! For details, visit [www.energyforlifewalk.org/evansville](http://www.energyforlifewalk.org/evansville).

**May 5, 2013.** Josh Clarke, along with other runners, will be participating in the Cincinnati Flying Pig Marathon to raise funds in honor of Leslie Whitt-Williams, who lost her battle with mitochondrial disease in the fall of 2012. If you are interested in running or donating, visit [www.umdf.org/flyingpigmarathon](http://www.umdf.org/flyingpigmarathon).

**May 18, 2013.** The Indiana Chapter will be holding their third annual Energy for Life Walkathon at the IUPUI Campus outside of Taylor Hall. Please join us again this year and help us to spread awareness and raise funds to find a cure. Visit [www.energyforlifewalk.org/indianapolis](http://www.energyforlifewalk.org/indianapolis) for details.

**June 1, 2013.** The UMDF is an official charity partner of the 2013 Hospital Hill Run in Kansas City! If you are interested in running or volunteering on behalf of the UMDF, contact Robin Jones for details at [1fastnurse@gmail.com](mailto:1fastnurse@gmail.com).

**June 1, 2013.** The second annual Energy for Life Walkathon: Pittsburgh will be held on the Great Lawn at Heinz Field in Pittsburgh, PA. Join us for another great year as we walk along the scenic Allegheny River. Go to [www.energyforlifewalk.org/pittsburgh](http://www.energyforlifewalk.org/pittsburgh) for information!

**Ongoing.** Mary Jo Balistreri has recently released her newest poetry book "Gathering the Harvest" and will be donating the proceeds of the book sales to the UMDF in memory of her grandsons, Zachery and Sam Juhlmann. To purchase Mary Jo's book, go to [www.maryjobalistreripoet.com](http://www.maryjobalistreripoet.com).

**Ongoing.** Louann Carnahan of River Forest, IL continues to sell her Piano Masterpiece CD with proceeds benefitting the UMDF. To purchase this magical CD, please visit [www.umdf.org/pianomasterpieces](http://www.umdf.org/pianomasterpieces).



*A logo created by the UMDF for the Cincinnati Flying Pig Marathon.*



*Two of the Directors for Dawgs for Mito, Meredith Frobose and Michael Trainer, are pictured above during Mitochondrial Disease Awareness Week.*

# Elementary Students Create Mito Club

by Alison Cooley, UMDF National Staff

"You never know where this can lead." That's what Charles A. Mohan, Jr., Chief Executive Officer/Executive Director of the United Mitochondrial Disease Foundation (UMDF), said to Cecilia Coloret, at a recent visit. Coloret teaches gifted children in grades 3, 4, and 5 at Hunter's Green Elementary School in Tampa, Florida.

Mohan and Margaret Moore, Southeast Regional Coordinator, recently came to visit Coloret and her students. Mohan and the students both asked one another questions. Coloret said it was a beautiful experience. Mohan explained how he started the UMDF and told them about his daughter, Gina. One of the students asked Mohan what his daughter wanted to be when she grew up, and he said, "a teacher," which brought a tear to Coloret's eyes.

Prior to the visit from UMDF staff, Coloret asked her two fifth grade classes if they would be interested in researching mitochondrial disease, a disease that she and a kindergartner named Emily, who attends the school, both have. Twelve of her fifth graders have shown interest. Each of them did their own research and created Microsoft PowerPoint presentations explaining what the disease is. These slide shows have been aired on the school's television station. The group also created a song about mitochondrial disease and is working on putting a video together about a day in the life of Emily, which will help everyone understand her needs, including the fact that she has a gastric feeding tube, which is why she never takes her backpack off.

Coloret just intended this to be an optional research project for the kids. However, she said that the enthusiasm of the children has been amazing. They not only wanted to learn about the disease, but they told Coloret that they want to spread the word about it so they can help Emily and their teacher. The children's enthusiasm has led to the creation of Mighty Mito Fighters, a club at the school, which hopes to create awareness and education about mitochondrial disease, as well as raise money for the UMDF.

The Mighty Mito Fighters already have the support of the school's PTA and supporting their initiatives will be the school's

ongoing philanthropy. Coloret thought that she would be the one to suggest awareness and fundraising activities; she never thought it would be the other way around. The students have planned a Go Crazy for a Cure day on March 20, 2013, where the students will wear green for the day. This will be the kick-off day to introduce the Mighty Mito Fighters Club to the student body. They also plan to continue to educate students and bring about awareness by creating posters, having a table at the school's spring carnival, and launching a Coins for a Cure campaign next year. In addition, they want to coordinate a talent show where they can display facts about

mito and raise money for the UMDF. The students hope to make a big impact next year during Awareness Week, as Emily's birthday happens to fall during that week. Some of her students will also be illustrating a children's book, which was written by UMDF employee, Jean Bassett, and explains to children what mitochondrial disease is.

Coloret is proud of her students. She said, "It has been fun to watch my students make a difference. The students came up with all of these ideas on their own." She told them that this is our way of giving back to the community and that they can make a difference in the world if they do something they believe in. Coloret, who was diagnosed with mitochondrial disease last year, also said that she has been able to deal with her diagnosis better since she has started focusing on others. She hopes that by raising awareness, others who are affected by mitochondrial disease won't have to wait until they are 37, like she did, to receive their diagnosis.



Cecilia Coloret poses with the Mighty Mito Fighters.

## Awareness Week 2013

It's not too early to start thinking about Awareness Week 2013.

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For ideas, go to the UMDF website, [www.umdf.org](http://www.umdf.org) and look under "Get Involved."

You will see a section about Awareness Week there.

# Coins for a Cure Inspires Longtime Support



Paula Quinn and Christine Glock's winning door.

Paula Quinn and Christine Glock, employees of PaTTAN (Pennsylvania Training and Technical Assistance Network) won a holiday door decorating contest that the organization's Pittsburgh Office held in December of 2012. Glock and Quinn are office mates and decorated their door with a Steeler snowman. The employees voted by putting money in a donation envelope on the door that they liked best. The two doors that collected the

largest donations split the total money collected to donate to the charity of their choice. Glock and Quinn were top winners and gave \$250 in donations to the United Mitochondrial Disease Foundation (UMDF).

Quinn and her family have been supporters of the UMDF for many years. Quinn's daughter, Nina, was a student at St. Bernadette Roman Catholic School in Pittsburgh, PA and participated in their annual UMDF Coins for a Cure while she was growing up. As noted in the events section of this newsletter, the staff and students raised over \$1,600 for the UMDF this year.



Chuck Mohan's late daughter, Gina, holding Nina Quinn in 1993.

Coins for a Cure is an easy way to raise money for mitochondrial disease research. It shows children that they can make a difference in the world by giving, one coin at a time.

All you need is a few coin collection containers and some willing participants. Simply approach your school or a local business owner you may know and ask for their approval to hold a Coins for a Cure campaign for the UMDF. Then, set out the containers and let the fun begin!

If you are interested in holding a Coins for a Cure campaign, go to [www.umdf.org/coinsforacure](http://www.umdf.org/coinsforacure), to register online. For questions, contact [events@umdf.org](mailto:events@umdf.org).



Nina Quinn helping Chuck Mohan teach the students about mitochondrial disease at St. Bernadette's School in 2006.

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# UMDF national

News from the national office.

## **Jack Black Honors UMDF with Red Carpet Toast at the Golden Globes**

Actor Jack Black, a longtime supporter of the United Mitochondrial Disease Foundation (UMDF), honored those battling mitochondrial disease when he offered a “red carpet toast” during the January 13th broadcast of the 70th Annual Golden Globe Awards. For each toast raised, Moët & Chandon donated \$1,000 to the chosen charity in the nominees’ names. Black chose the UMDF and Moët & Chandon donated the \$1,000 in his name to the UMDF. Including this year’s outstanding participation, nearly \$200,000 has been raised for over 130 charities as part of its “Toast for a Cause.” The UMDF thanks Jack Black and Moët & Chandon for this very generous donation.

### **UMDF Meets with Patient Group from Japan**



On February 11, 2013, some of the UMDF staff met with representatives from Tokyo who were interested in learning from UMDF how to start a patient advocacy group for mitochondrial disease patients in Japan.

### **UMDF Receives \$10,000**



On February 4, 2013, Eric J. Werner, Executive Director of J.P. Morgan, presented UMDF CEO/Executive Director Chuck Mohan with a generous gift of \$10,000. Thank you Chase Community Giving and all of you for your help!

**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.