



United Mitochondrial Disease Foundation

MITOCHONDRIAL NEWS



Volume 11 • Issue 3

Ask the Mito Doc

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Incidence and Prevalence Rates of Mitochondrial Diseases

Bruce H. Cohen, MD, and Sumit Parikh, MD

Department of Neurology, Section of Pediatric Neurology, The Cleveland Clinic Foundation

There is much discussion as to the incidence and prevalence of mitochondrial disorders. The purpose of this article is to discuss why these statistics are not readily available and to give a best estimate on these numbers.

Some experts prefer to define mitochondrial disorders as those diseases only involving defects in the terminal stage of energy production: the electron transport chain. Other experts also would include disorders of fatty acid oxidation, the citric acid cycle and other enzymatic reactions that take place within the mitochondria. The UMDF serves those with all mitochondrial disorders, but for this discussion of prevalence and incidence, it is easier to only discuss the disorders of the electron transport chain.

It is first important to clearly define incidence and prevalence, which are rigid statistical terms. The incidence of a disease is defined as how many people in a defined population will get a particular disease each year. Prevalence is a term that indicates how many people or a percentage of people in a defined population already have a particular disease. The incidence and prevalence have very different meanings. For some disorders that are chronic and not life threatening, the incidence may be small while the prevalence is much larger because it is the cumulative sum of the incidence rates over time. For other disorders where death is rapid, the person that dies of the disease is no longer counted in the prevalence rate, in which case the incidence may be high but the prevalence relatively low. Many persons with mitochondrial disorders live long but very difficult lives. Others have disorders

that are rapidly fatal. Therefore a more common disorder that is rapidly fatal may have a relatively high incidence but a relatively low prevalence. Conversely, a less common disorder that is not life-threatening may have a relatively low incidence, but a relatively high prevalence. Because the individually named mitochondrial disorders are rare, we tend to group all of the mitochondrial diseases as one disorder when we ask the question as to how common mitochondrial disorders are. Combining all the disorders into one organization like the UMDF is both necessary and convenient, as most of our members share common goals. However, it is not a statistically valid question to ask what is the incidence and prevalence of all mitochondrial disorders, as the process of combining all the individual diseases together will result in inaccurate conclusions. Finally, incidence requires a point in time when a person is positively diagnosed, which is not always easy to do for a person with a mitochondrial disorder.

In addition to this problem, scientists have not reached complete agreement as to how to define a mitochondrial disorder. When experts discuss this in an open forum, most of us would agree that "we know which people certainly have a mitochondrial disorder and those that certainly do not," but there are many patients, possibly the larger than the combined certain and certainly not groups, where there is no firm agreement.

The complexity of this debate can be highlighted further if we take a well-defined and easily tested for A3243G mitochondrial DNA mutation. This is the mutation often causing MELAS. In the

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Ask the Mito Doc

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

Responders for this issue: Richard G. Boles, M.D., Children's Hospital, Los Angeles, and Russell Saneto, DO, Ph.D., Children's Hospital, Seattle.

The Question is:

Our 8-year-old daughter has been diagnosed with Oxphos Defect in Complex 1 and 3. She has also been diagnosed with Cyclic Vomiting syndrome. She has had periods of vomiting that last up to 12 days and then has a rapid recovery. We are now in day 45 of uncontrollable vomiting with no end in sight. She had a partial fundoplication when she was two. They did an endoscopy and found only a hiatal hernia. Other than the vomiting episodes that are few and far between, she is a normal functioning child. Our question is: Can Complex 1 and 3 develop into more severe complications or has she been misdiagnosed? We are searching for any ideas or answers. We are so frustrated and are unable to get any answers.

Response From: Richard G. Boles, M.D.

Regarding the Complex 1 and 3 deficiency, I doubt that there has been a misdiagnosis. Cyclic Vomiting

syndrome refers to multiple episodes of nausea, vomiting and lethargy, with the essential absence of these symptoms between episodes. Many patients do have other conditions/symptoms consistent with dysautonomia (migraine, irritable bowel, muscle cramps, rapid heart rate, chronic fatigue, etc.) and/or other neurological conditions (developmental delay, seizures, hypotonia/floppy muscles, depression, anxiety, etc.). However, many patients only have vomiting episodes and are otherwise healthy. In most of those patients both with and without other conditions, mitochondrial dysfunction/disease is present. The mitochondrial dysfunction is often, but not always, maternally inherited, with some of the above listed dysautonomic and/or other neurological symptoms being present in the mother and her other relatives that have the same mitochondrial DNA.

In those children with cyclic vomiting in which a muscle biopsy is performed, Complex 1 deficiency is common, and I have seen cases with Complex 3 deficiency as well.

In Cyclic Vomiting syndrome, vomiting episodes usually last from a few hours to several days. Episodes that last for 12 days can occur, but are uncommon.

In some cases, an episode can last much longer, but it is also possible that there is now an additional problem in your daughter that is contributing to the very prolonged episode of vomiting. There are far more possibilities than can be covered here, but malrotation of the gut and pancreatitis deserve mention.

In mitochondrial disease, additional problems can occur with time. However, in children with normal to near-normal intelligence in which cyclic vomiting is the only major problem, the vast majority do well and the episodes can be brought under good control. Mitochondrial-directed therapies such as frequent feedings/fasting avoidance, L-carnitine and co-enzyme Q10 are often helpful in my experience. An excellent resource is the Cyclic Vomiting Syndrome Association at www.cvsaonline.org, which can be helpful in terms of other conditions that might be present, how to prevent and treat vomiting episodes and

finding a physician who is an expert in the field.

The Question is:

My daughter has NARP 8993 T-C mutation. Since 1996, she has been unable to walk or talk and eats only soft meals. She was constipated for one year and afterwards had stool incontinence for several years until six months ago. Now she has constipation again, in spite of eating many fibers in cereals and fruits. Not even mineral oil helps. Fleet enema helps sometimes. She has been on two anticonvulsants (phenobarbital and lamotrigine) for several years. Any suggestion?

Response From: Russell Saneto, DO, Ph.D.

I am sorry that your daughter is having severe constipation. Unfortunately, this seems to be a common problem in children and adolescents with epilepsy as well as mitochondrial disease. In my patient population, those with both disorders seem to be more difficult to control. For the most part, most of us do not think that it is the seizure medications themselves, but the central nervous system and likely the enteric (GI tract) nervous system and its control of the muscles of the GI tract. The usual controls that interplay between the GI tract and the brain are disrupted in epilepsy. The movement of the GI tract is compromised, and there is likely a slowing of the constant pulsating GI tract and constipation. Likewise, when the muscles of the GI tract are affected with abnormal mitochondria (deficient energy) they become dysfunctional and add to the poor efficiency of GI movement. Together, all this adds up to a lot of stool in the bowel and constipation.

Treatment can be very challenging. We usually try a stepwise plan. Initially, we try high fiber and fruits (especially pears). The next step is mild laxatives and, if no effect, the use of stronger laxatives. Intermixed with this is the use of glycerin suppositories. When the problem is severe, we sometimes have to resort to enemas. Many of my patients are on chronic laxative dosing with intermittent enemas.



When embarking on a journey, whether large or small, it is best to set out with a detailed plan so that you will know where you are going, how and when you are going to get there and who will be helping you along the way. When you take a family vacation, for instance, you choose your destination, book your hotel, make airline or other travel arrangements with your agent, create an itinerary, put together a packing list, etc.

At UMDF, we are setting out on a challenging journey of great importance - to promote research and education for the diagnosis, treatment and cure of mitochondrial disorders and to provide support to affected individuals and families. Setting our mission is easy; we know exactly why we are here. The next step in our quest for a cure is to put together a comprehensive strategic plan so that we know where we are going, how and when we want to get there and whose help we will need to move forward.

To create our strategic plan, UMDF assembled a committee of board members, volunteer leaders and staff to assess the current status of UMDF, as well as our strengths and weaknesses. From this evaluation came five strategic objectives:

- To continually enhance the effectiveness of the UMDF by developing its volunteers, staff, technology and infrastructure.
- To develop and implement a multi-faceted fundraising strategy and plan that emphasizes

Chairman's Message

the development of strategic relationships, leveraging the membership and exploring the solicitation of grants and corporate sponsorships.

- To develop and effectively communicate the UMDF story and brand, both externally and internally.
- To continually enhance the UMDF's educational capabilities and resources with a special emphasis on members, the medical community, affected individuals and families.
- To continually enhance the UMDF's ability to guide and support its members by improving support to the chapters and groups and developing leaders and other volunteers.

In order to better reach these objectives, the planning committee set out seven goals for 2006-2007.

- To refine and complete the implementation of a comprehensive marketing and communications plan.
- To have a robust, effective and proactive committee structure.
- To present to the board a recommendation to upgrade UMDF's IT infrastructure.
- To successfully include language for government support of mitochondrial disease.
- To raise \$2.6 million in FY 2007.
- To increase net membership by 25 percent by the end of FY 2007.
- To restructure and broaden the scope of the Scientific Advisory Board (SAB) into a Scientific and Medical Advisory Board (SMAB).

While some of these goals may seem ambitious at first glance, the staff and volunteers of UMDF look ahead with optimism and dedication to reaching these ends for the benefit of

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UMDF MISSION

To promote research and education for the diagnosis, treatment and cure of mitochondrial disorders and to provide support to affected individuals and families.

Newsletter Editor

Kara Strittmatter
kara@umdf.org

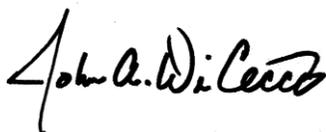
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Chairman's Message Cont.

our membership, the scientific community and the world at large.

In setting these goals, we are creating a road map to success in maximizing education and awareness, encouraging ever-expanding research and enhancing the support we can offer to patients and families. We are hopeful that analyzing and evaluating our successes and failures in achieving these specific goals and the broader objectives behind them will allow UMDF to stay on track toward reaching our ultimate destination: a cure.

Yours toward a cure,



Member Forums

One of the requests we receive most frequently at the UMDF office is a request for networking.

In answer to these requests, the UMDF recently made it easier than ever for members to connect with one another and share their experiences, questions and advice.

The UMDF website now offers a "Member Forums" feature. When members log on to the UMDF website using their User IDs and passwords, they will be greeted by a new menu item labeled "Member Forums."

The Member Forums are divided into three subcategories—Networking, Supportive Services and Behavioral Concerns. Members can use the forums to read messages left by others and post questions or topics for discussion.

Please visit the Member Forums today and start connecting with other members from around the world!

Congratulations to the recipients of the 2006 Heartstrings and LEAP Awards



UMDF 2006 *Heartstrings* Winner, 14-year-old Hannah Roberts of Cleveland, Ohio, has a mitochondrial disease.

Over the past year and half, Hannah has been working on a Jewish Holiday Cookbook. The project began in Speech Therapy – she and her therapist were looking for new ways to work with Hannah on her language skills.

Since Hannah enjoys baking and is active in her religion, they decided to try decorating cakes for the different Jewish holidays. Hannah came up with ideas on how to decorate cakes for different holidays and then wrote down directions so another person could create the cake on their own. The idea grew into a cookbook – Hannah sold the book to raise awareness about mitochondrial disease and dollars for UMDF.

Her project took over a year to put together from start to finish. Hannah sold all of her copies (100) within a week and half – raising \$1,000 for UMDF. Her parents said on their nomination form, "What tugged at us most about this project – it was never about Hannah. When she came up with the idea, it was 'to help others like me.'"



Our 2006 *LEAP* Winner, Pam Johnson, M.D., is the president of the Kansas City Chapter and newly appointed chapter liaison on the UMDF Board of Trustees.

Pam accepted the responsibility of Kansas City Chapter President in 2005 -- even though her disease symptoms cause her to tire easily. She willingly organized and reorganized the chapter to bring back key members to the board and works hard to motivate and support the KC Chapter Board, as well as striving to meet the needs of the families that call her.

Pam is a pediatrician but can no longer practice because of her mitochondrial disease. Instead of being discouraged by what she can no longer do, Pam channels her energy and knowledge to help parents and other adults. Her commitment to share information and inform the public about mitochondrial disease is never ending and she always gives 150% -- even when she should be resting!

Pam is Living with the disease, Encouraging others to help the UMDF and each other, Achieving by building the capacity of the Kansas City Chapter and Persisting every day to help others.

See pages 22-23 for applications for this year's awards!

Spotlight - UMDF Board Member

To keep UMDF members connected to our board volunteers, we will “spotlight” our trustees from time to time in the *Mitochondrial News*. In this issue, we will spotlight one of our trustees, Bruce H. Cohen, M.D., pediatric neurologist at the Cleveland Clinic Foundation.



During the 2002 UMDF epidemiology meeting, Cohen tries to keep up with workload.

For hundreds of patients and families, Dr. Bruce Cohen is no stranger, and the UMDF will never be able to thank him enough for his years



Cohen speaks to local media at the 2004 Ohio Chapter Run/Walk.



During the 2002 UMDF Ohio Chapter Run/Walk, Cohen chats with Lily Grossman and her dad, Steve.

of dedication to our mission by volunteering precious time that very few physicians are able or willing to give.

Dr. Cohen joined the UMDF Board in 1998 and rarely said no to our many, many requests. Write a newsletter article? Sure! Chair a UMDF symposium? No problem! How about doing grand rounds on mito? Absolutely! Dr. Cohen has served on symposium committees and chapter committees and has always



Cohen talking CoQ10 with exhibitor, Tishcon Corporation, at the 2004 UMDF Symposium in Pittsburgh, PA.

played a key role on the executive committee. He has given the board and staff the perspective necessary to communicate effectively with doctors in the mitochondrial arena.

As the course director for the 2000 and 2005 symposiums in Cleveland and St. Louis, Dr. Cohen and his committee brought together some of the top specialists and researchers from around the world, and even with



Cohen reviewing agenda with UMDF Chairman (and now CEO), Chuck Mohan, at the 2005 UMDF symposium in St. Louis.

his hectic schedule during those meetings, he made every effort to speak with patients and families.

His term on the UMDF board ends this year, but he has made a lifetime commitment to the quest for better treatments and cures for mitochondrial disease. And if the staff has its way, UMDF will continue to keep him busy.

***Thanks, Dr. Cohen!
You are an inspiration to us all!***

Incidence and Prevalence Rates of Mitochondrial Diseases

Continued from page 1

United States, this A3243G mutation occurs in 163 of every 1,000,000 people, or about one in 6135 people. However, not all of these people have the disorder MELAS; they simply have the mutation. A study of people living in Finland found a prevalence of the disease MELAS caused by the A3243G mutation of 102 per 1,000,000 people, or one in 9800 people. This prevalence is a statistic, which means that not

every person in Finland was examined and tested for MELAS, but based on known cases, a reasonable effort was made to estimate how common the mutation is. Although the studies that gave us these results may have some degree of error, this is the best data available, and for our discussion, we can give an estimated prevalence of MELAS due to the A3243G mutation as one in 10,000 persons. We will have

to assume the prevalence is similar in both the USA and Finland, which may or may not be true. At the time this article was written (Fall 2006), the U.S. population just climbed to 300,000,000. Therefore, there are about 30,000 people in the U.S. living with MELAS caused by this mutation. This prevalence is three times the membership of the UMDF, and we can

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Chapter Activities

ATLANTA AREA CHAPTER

Atlanta, GA

Upcoming Events:

- **November 11-December 31** - To celebrate her fourth birthday, Trinity Sumlin and her mother, Sheryl, started a letter writing campaign to benefit UMDF.
- **Sunday, March 25, 2007** -- ING Atlanta Half Marathon, Atlanta, Ga. Marilyn Arkin will be running in honor of her son, Ross. Marilyn has set her goal to complete the 1/2 marathon and to raise \$10,000! Good Luck Marilyn!

Special Events - THANK YOU!

- **September 16** - The Saint Andrew Rowing Club hosted a *Row for Mito* at Riverside Park, Roswell, Ga. Nearly \$6000 was raised in honor of Ty Seldes.
- **September 23** - In honor of Joseph Hendley, the Atlanta chapter held their *Going the Extra Mile for Josey* 24-hour Ultra Run. Thanks to all who participated!
- **October 8** - Organized by Sheri Seldes, the first *Mito What? Ty's Trot Toward a Cure*, honoring Ty Seldes, took place at the Georgia Tech Campus, raising more than \$35,000 for UMDF.



President: Chris Swinn

Phone: 770-270-5337

Email: AtlChapter@umdf.org

- **October 23** - The *Third Annual Fore-A-Cure Golf Tournament* was held at the Standard Club in Duluth, Ga. This event was a great success, with more than \$24,000 collected so far in honor of all affected.



INDIANA CHAPTER

Indianapolis, IN

President: Sue Ann Bube

Phone: 317-894-9099

Email: INChapter@umdf.org

Upcoming Events:

- **Saturday, February 10, 2007** -- UMDF will be teaming up for the fourth year with the Indiana ICE at Conesco Fieldhouse for the *Pack the House* event. UMDF is the only charity chosen this year, and funds will be raised in honor of all affected. For more information, contact Chapter President Sue Ann Bube.

For activities or information about other UMDF Chapters, please see page 21 for contact information.

Want to Get Involved?!

Contact your local chapter or group **today**. Chapters and Groups will continue to grow with your HELP. **YOU can make a difference.**

Strathmore-Vanderbilt Women's Club Holds Its 2006 Philanthropic Dinner Dance

The evening of November 3 was very magical in Manhasset, N.Y., when 250 people gathered to support UMDF at *Magic, Martinis and Music*.

Vita Amato hosted the event at the Strathmore-Vanderbilt Country Club in honor of her grand-niece Mary Rose Zuzzolo, daughter of Kim and Anthony Zuzzolo.

The silent auction had more than 40 outstanding items, the raffle boasted equally enticing items and the 50/50 was almost sold out before the evening even began!

After sampling a variety of martinis and

being delighted by the magic of Matt Furman, guests were treated to the sounds of the Heart Rob and Patty Meade Band, dinner and an inspiring talk from Chuck Mohan, UMDF's executive director.

Although donations are still coming in, more than \$70,000 has been raised so far. The Foundation sincerely thanks Vita and all who made the evening possible.



OHIO CHAPTER

Cleveland, OH

Upcoming Events

- **Saturday, May 12, 2007** -- *Run Wild for a Cure Walk/Run 5K*. New date, new location, new image! Contact the Ohio chapter for further information.

Special Events - THANK YOU!

- **August 5-6** - Nisha Amos' neices, Rachel and Madison, set up a cookie and drink stand at their family's garage sale and raised \$122 for UMDF in honor of Caroline Lyman. Great job, girls!
- **October 14** - The Ohio chapter held their *2nd Annual Clams for a Cure* at Hoggy's Restaurant in Valley View, Ohio. Thanks to all who organized and attended!

President: Bill Hodges
Phone: 440-235-2451
Email: OHChapter@umdf.org

Ohio Chapter Holds Mini Symposium

The Ohio Chapter held their mini-symposium November 5 at the Solon Community Center.

The mini conference featured seminars by Dr. Bruce H. Cohen, M.D., of The Cleveland Clinic, Dr. Charles Hoppel and Dr. Robert Hostoffer of Case Western Reserve University and Dr. Mini Wallace of Rainbow Babies and Children's Hospital. Topics covered included "Symptom Management in Mitochondrial Disease," "Mitochondria: Then, Now, Future" and "Mitochondrial Disorders and Gamma Globulin."

The Ohio Chapter would like to



Drs. Chuck Hoppel, Bruce Cohen and Robert Hostoffer at the Ohio Mini Symposium

give special thanks to the doctors for their dedication in sharing their time and knowledge and for the compassion they show the children, family members and friends who have been affected by mitochondrial disease.

NEW ENGLAND CHAPTER

Boston, MA

Special Events - THANK YOU!

- **August 27** - Becky and Daniel Genie of the New England chapter held their *Friends of Cameron* picnic, raising more than \$9,700 in honor of Cameron Genie.
- **October 4** - The *Windermere Elementary School Walk*, organized by Heidi Bailey, raised more than \$2,600 in honor of Matthew Bailey. Thanks, walkers!



Windermere Elementary School Students

- **October 12** - Thanks to Harry and Virginia Ingram and Chris and Angelo Polumbo for organizing their casino bus trip from Springfield, Mass., to the Fox Woods Casino in Connecticut. The group passed out candy, played bingo and watched a movie on the way to the casino, and a good time was had by all -- all while raising more than \$400 for the UMDF in honor of Dominique Ingram.

President: Bridget Willis
Phone: 508-224-7165
Email: NEngChapter@umdf.org

Weekend of Hope Raises \$75,000+

- **October 7-8** - The New England chapter's *Mito What? Weekend of Hope* was a great success. On Saturday, more than 400 walkers registered for the Plymouth 5K Walk, and 600 participated in the Family Fun Zone activities. On Sunday, there were 300



participants in the 7th Annual Longmeadow 5K Walk and Family Fun day. Massachusetts state representatives were on hand both days to read and

present the governor's "Mitochondrial Disease Awareness Week" proclamation. The weekend celebration raised more than \$75,000 in honor of all affected.



Fundraisers

Coins for a Cure Campaign!

There's a very simple way to raise money for mitochondrial disease research! The project is called *Coins for a Cure*, and it's as easy as 1, 2, 3! The *Coins* program shows children that they can make a difference in the world through their actions, one coin at a time.



UMDF needs your help to organize events, contact local schools and businesses, encourage students or patrons to participate, count the coins and return the final total to UMDF.

The special events department will supply stickers for your coin containers, coin containers (optional), informational literature on mitochondrial disease, promotional information and sample correspondence for your campaign and certificates for

winning classes or stores.

Here are some helpful hints for a successful *Coins for a Cure* campaign:

- Make *Coins for a Cure* a friendly competition between classes or stores.
- Hold a *Coins* fundraiser in honor of a child or adult affected with mitochondrial disease.
- Use incentives for the class or store with the highest totals.
- Be sure to keep your containers and totals separate for contests.

Marathon runners race for UMDF

In a different kind of “miles for mito,” Chris and Tova Sido ran in the Chicago Marathon to raise money for the Charlie and Louisa Fund through UMDF. Despite the cool, misty weather, they had a great time, and *great times!* Chris finished the 26.2 mile run in three hours and 54 minutes, and Tova finished in three hours and 58 minutes. Congratulations on this amazing accomplishment, and our heartfelt thanks for raising nearly \$14,000 for mitochondrial research!



For more information, contact UMDF at 412-793-8077 or info@umdf.org.

If you have an event announcement or an idea for an article for the Mitochondrial News, please email kara@umdf.org. We want to hear from YOU!

For information on starting a fundraiser in your area, email jodie@umdf.org.

You Go Girl! Golf Tournament Raises \$9,500+



One hundred twenty golfers turned out September 9 for a fun-filled day at the *You Go Girl!* golf tournament in Pittsburgh. The event raised more than \$9,500 for UMDF.



Fundraisers

- **July 8** - The *Second Annual Carter Martin Classic*, a golf outing and dinner in Maquoketa, Iowa, organized by Greg & Carolyn Martin raised almost \$11,800 in honor of their son, Carter.
- **August 26** - Kathy Gillen kicked off the *Merritt Gillen Birthday Letter Writing Campaign* on August 26 in Pittsburgh, raising more than \$6000 so far!
- **August 27** - Alyssa Rogers' lemonade stand earned almost \$20 in honor of Carson Rogers of Greer, S.C.
- **Summer 2006** - Emily Fischer & Joe Gilsdorf held various fundraisers throughout the summer at Nessun Dorma Restaurant in Milwaukee, Wis., raising \$600 for UMDF in memory of Sam Juhlman and in honor of Zachary Juhlman.
- **September 15** - The *Elena's Hope Research Fund Benefit Dinner*, organized by Mindy Wellhouse, raised nearly \$25,000 in Kimberly, Wis., and Sandra Began Greenery offered \$10 for every wreath purchase received before October 15.
- **September 16** - A horseshoe tournament, prize raffle and 50/50 raffle were among the fundraisers held at the Nathan Mowrer Memorial Playground in Pottstown, Pa., which raised \$3,000 for UMDF.
- **September 22-24** - *Will Freeman's Birthday Trail Ride* was a three-day weekend packed with horseback riding, fresh grilled food, fun and fellowship! More than 300 were in attendance at the Demopolis, Ala., event, and \$15,700 was raised over the weekend!
- **September 24** - After some early rain, the sun was out for the *6th Annual Cruisin' for a Cure* event in Pittsburgh. About 200 cars participated in the cruise, raffles and other fundraisers, raising \$4,350 in honor of all affected.
- **October 2** - It was the first year for the *Kovalcik and Geraghty Wealth Partners Charity Golf Outing* in Columbus, Ohio, and UMDF was chosen as the first charity. More than \$28,000 was raised in honor of Ellie Kovalcik.
- **October 7** - The *MPS and MITO Walk/Run* in Eagan, Minn., raised more than \$2,800 for UMDF in honor of Patrick Luskey.
- **October 13** - Sarah Vincent raised \$400 in honor of James Vincent by hosting a *Casual Day for a Cause* at work in Slippery Rock, Pa. Great job, Sarah!
- **October 13** - Pamela Kania organized the *Bellamy Middle School Dress Down Days* in Chicopee, Mass., raising \$100 in honor of Dominique Ingram.
- **November 11** - Monica Canty ran in the Richmond, Va., Marathon in honor of her cousin Emma Jurek. Thanks, Monica, for your dedication -- and for raising \$2,500!
- **November 15** - The *Tory Burch After Hours Shopping Night* was held in cooperation with the Sewell Family of Dealerships. Twenty percent of sales made during the night of shopping at their Dallas, Texas, location will be donated to UMDF for the Charles and Louisa Sido Research Fund.

Karaoke for Kate!

Fun was had by all at the *3rd Annual Karaoke for Kate* on October 7 at the Port Vue Firehall in Port Vue, Pa.

Activities included a 50/50 drawing, a Chinese Auction, which included a Steeler autographed football, and of course, some singing fun!

This event was in loving memory of Kaitlin Vasilich, who lost her battle with mitochondrial disease on February 8, 2004. A donation of \$2,000 was made to the UMDF in memory of Kaitlin. The families would like to extend their deepest appreciation to all who attend the event and to those who donated items to make the evening a success.

Gifts from the Heart - Thank You ALL

- Ronald & Margaret Donohoe of Maine celebrated their 50th Wedding Anniversary in August. In lieu of gifts, \$1,200 was donated to UMDF in honor of Michael Pastorelli.
- Erin Hylka and Kevin Polansky from Massachusetts gave a donation of \$250 in lieu of wedding favors.
- In September, Katharine Lewis of California and her parents asked friends and family to donate to UMDF in lieu of birthday presents for Katharine's third birthday, raising a total of \$250. Thanks to all who participated.
- Mark and Kathy Campbell donated \$300 in memory of Gina Marie Mohan in lieu of wedding favors for their daughter, Kate's, wedding in Pittsburgh.
- Stephen and Jackie Pace of North Carolina, grandparents of Alex Crisp, celebrated their 30th wedding anniversary in September. Donations to UMDF were encouraged in lieu of anniversary gifts, and more than \$1,500 was received. Thanks to Stephen and Jackie and their friends and family for their generosity.

★ Mark your Calendars ★

See pages 6-7 for more details on chapter events

- **Saturday, February 10, 2007** -- UMDF will be teaming up for the fourth year with the Indiana ICE at Conseco Fieldhouse for the *Pack the House* event. UMDF is the only charity chosen this year, and funds will be raised in honor of all affected. For more information, contact Sue Ann Bube at INChapter@umdf.org or at 317-894-9099.
- **Saturday, February 17, 2007** -- The *Third annual Curl-O-Rama*, organized by Chesapeake Physical & Aquatic Therapy, will be held at the National Capital Curling Center in Laurel, Md. No prior experience necessary. Funds raised will be donated to UMDF in honor of Peter Lubelczyk. For more information, contact Stephanie Coleman at 301-498-2212 or scoleman@chesapeakept.com.
- **Saturday, February 17, 2007** -- Artist Peter Max will be the guest of honor at the second annual *Meet the Artist* event in Florida. A percentage of the art show sales will be donated to UMDF in memory of R.J. Victor, event organizer Jenah Victor's brother. For more information, contact Jenah Victor at gopjenah@hotmail.com.
- **Saturday, March 10, 2007** -- The *Fourth Annual Bet on Baylee Casino Night* will be held in Zanesville, Ohio. Activities will include Texas Hold'em and special auctions! Proceeds will be donated in honor of Baylee Thompson. For more information, contact Jody Thompson at 740-982-1244.
- **Saturday, March 10, 2007** -- The *Brittany Wilkinson Dinner & Silent Auction*, hosted by Linda and Brittany Wilkinson, will benefit the Brittany Wilkinson Research Fund. This is the second year for this event, which will be held in Clover, Ca. For more information, contact Linda Wilkinson at 559-299-1767 or dotoheven@aol.com.
- **March 16-18, 2007** -- The third annual *Writers at the Beach* event, a three-day writer's conference held in memory of Sam Juhlman and in honor of Zachary Juhlman, will take place in Rehoboth Beach, Del. For more information, contact Maribeth Fischer at mbfischer1@verizon.net or visit www.writersatthebeach.com.
- **March 17-18, 2007** -- For the third year, the Lonely Leprechaun will run in the Shamrock Marathon in Virginia Beach. *Preston's March* will be held on Sunday to benefit Preston Buenaga. For more information, contact Deb Buenaga at dsb6891@earthlink.net.
- **March 23-24, 2007** -- The *Caroline's On My Mind* weekend will be held in Spartanburg, S.C. Events will include a golf tournament and a 5K walk/run in honor of the Caroline Virginia Pulliam Mitochondrial Disease Fund. For more information, contact UMDF at info@umdf.org.
- **Sunday, March 25, 2007** -- ING Atlanta Half Marathon, Atlanta, Ga. Marilyn Arkin will be running in honor of her son, Ross. Marilyn has set her goal to complete the 1/2 marathon and to raise \$10,000! Good Luck Marilyn!
- **May 2007** -- *Brittany Wilkinson Golf Tournament*, Clover, Ca. For more information, contact Linda Wilkinson at 55-299-1767 or dotoheven@aol.com.
- **Saturday, May 12, 2007** -- Ohio's *Run Wild for a Cure Walk/Run 5K*. New date, new location, new image! Contact the Ohio chapter at 440-235-2451 or at OHChapter@umdf.org for further information.
- **Saturday, June 2, 2007** -- The 5th Annual *One Step Closer to a Cure 5K Run/1 Mile Walk* will be held again at the North Hills Boat House in North Park, Pittsburgh. For more information, please contact info@umdf.org.

Coins for a Cure Kickoff!

The *Coins for a Cure* campaign is off to a great start with more than ten participants, including St. Bernadette's School in Pittsburgh, which is participating for the fifth year in memory of Gina Mohan! Coin collectors will also be placed in Wine & Spirit stores throughout Western PA through the month of December. Below is a list of our individual *Coins* participants so far:

Participant

Paul & Maura Buckley
Jennifer Bukovac
Dustin & Kelly Ann
Conover
Nicole Goerges
Rita Grosso

In Honor Of

Neil & Tyler Buckley
Stephen & Samuel Bukovac
Brandon Leach Research
Fund
Abigail Goerges
Riley Henderson

Participant

Beth Hartman
Susie Leach

Diana Nieves
Kim Stover
Melissa Weiner

In Honor Of

Haley Hartman
Brandon Leach Research
Fund
Nicole Nieves-Roberts
Andrew Stover
Melissa Weiner

For more information on how you can start a *Coins* campaign of your own, contact taniah@umdf.org.

Upcoming Clinical Trials

Phase 3 Clinical Trial of CoQ₁₀ in Genetic Mitochondrial Diseases

A new research study at the University of Florida will be the first controlled clinical trial of chronic oral CoQ₁₀ for patients with genetic mitochondrial diseases. Peter W. Stacpoole, Ph.D., M.D., will be the principal investigator for the trial.

This is an FDA-approved multicenter study funded by both the U.S. FDA and the NIH. Eligible patients are children ages 12 months to 17 years with suspected or proven defects in one or more enzymes of the respiratory chain or with a mutation in mitochondrial DNA.

The participating study sites are:

- General Clinical Research Center, University of Florida, Gainesville, Fla. (lead institution)
- General Clinical Research Center, Rainbow Babies and Children's Hospital, Case Western Reserve University, Cleveland, Ohio
- General Clinical Research Center, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio
- Hospital for Sick Children, University of Toronto, Toronto, Canada

If you have a patient who has a suspected or proven mitochondrial disease whose family might be interested in this study, or if you have additional questions regarding the trial, please contact:

Ms. Lesa Gilbert, RN, CoQ₁₀ Clinical Trial Coordinator
Phone: (352) 392-2321 Fax: (352) 846-0990
Toll-free Pager: (888) 961-9068
Email: gilbelr@medicine.ufl.edu

Clinical Trials for Patients with Primary and Secondary OXPHOS Defects

Horizon Molecular Medicine is currently evaluating patients for eligibility for clinical trials and for review of diagnosis. Patients fall into three main categories: Primary OXPHOS defect, Secondary OXPHOS defect and mistaken diagnosis.

In order to appropriately screen patients and perform useful clinical studies, individuals are being carefully evaluated for eligibility. The study will involve about 300 patients. Agents being used will be released at a later date. The medications used will depend on the clinical and laboratory features displayed by the patients. Patients will be segregated into appropriate groups based on these criteria.

For more information, please contact:

Horizon Molecular Medicine
One Dunwoody Park, Suite 250
Atlanta, GA 30338
Phone: 678.225.0222
Email: info@horizonmedicine.com
Web: www.horizonmedicine.com

EPI-A0001 Evaluation Planned for 2007 for Patients with Respiratory Chain Diseases

Edison Pharmaceuticals is in the strategic planning stages for the evaluation of EPI-A0001 for respiratory chain diseases in 2007. More information will be forthcoming for patients in the next Mitochondrial News publication. Physicians and researchers wishing information please contact Guy Miller, gmiller@edisonpharma.com.

What is a Clinical Trial?

(Adapted from CLIMB (UK) article, Vol. 1, No. 4, October 2000)

Clinical trials are used to determine whether new drugs or treatments are both safe and effective. Carefully conducted clinical trials are the fastest and safest way to find treatments that work. New therapies are tested on people only after laboratory and animal studies show promising results.

What are clinical trial phases?

Clinical trials of experimental drugs proceed through four phases. In Phase I, a new drug or treatment is initially tested on a small group to evaluate safety, dosage and side effects. In Phase II, the drug or treatment is given to a larger group to test effectiveness and further evaluate safety. In Phase III, the drug or treatment is given to large groups to confirm effectiveness, monitor side effects, compare to current treatments and collect safe usage information. In Phase IV, studies continue testing the drug or treatment after it has been marketed to collect further information about effectiveness and side effects associated with long-term use.

What is a protocol?

All clinical trials are based on a set of rules called a protocol. A protocol identifies the participants; the schedule of tests, procedures, medications and dosages and the length of the study. While in a clinical trial, participants are seen regularly by the research staff to monitor their health and to determine the effectiveness of their treatment.

Who can participate in a clinical trial?

All clinical trials have guidelines about who can get into the program. The factors that allow someone to participate in a clinical trial are called inclusion criteria. The criteria are used to identify appropriate participants and keep them safe, and they help to ensure that researchers will be able to answer the questions they plan to study.

What is a placebo?

A placebo is an inactive pill, liquid or powder that has no treatment value. In clinical trials, experimental treatments are often compared with placebos to assess the treatment's effectiveness.

What is a control or control group?

A control is the standard by which experimental observations are evaluated. In many clinical trials, one group of patients will be given an experimental drug or treatment, while the control group is given either a standard treatment or a placebo.

What is a blinded or masked study?

A blinded or masked study is one in which participants do not know whether they are in the experimental or control group in a research study.

What is a double-blind or double-masked study?

A double-blind or double-masked study is one in which neither the participants nor the study staff know which participants are in the experimental group and which are in the control group. These studies are performed so neither the patients' nor the doctors' expectations about the experimental drug can influence the outcome.

What protections are there for study participants?

The government has strict guidelines and safeguards to protect people who choose to participate in clinical trials. Every clinical trial in the U.S. must be approved and monitored by an Institutional Review Board to make sure the risks are as low as possible and worth any potential benefits. All institutions that conduct or support biomedical research involving people must have an institutional review board that initially approves and periodically reviews the research.

Mitochondrial Riding the Wave

Preliminary Scientific Program June 13-16, 2007

Wednesday, June 13: Mitochondrial Mechanisms and Disease

- Aging, Apoptosis and the Mitochondrial Permeability Pore
- Mitochondria in Neurodegenerative Diseases
- Aging and Mitochondrial DNA
- Mitochondrial DNA and Longevity
- Mitochondrial Toxins and Disease

Thursday, June 14: The Role of Mitochondria in Diseases of Aging

- Mechanisms of Mitochondrial Morphology and Inheritance
- Mitochondrial Signaling
- Cytochrome c Oxidase and Regulation of the Electron Transport Chain
- Mitochondrial Polymerase γ and Disease

Friday, June 15: Current Options for Treatment

- Treatment of Mitochondrial Disease: Overcoming the Obstacles to Clinical Trials
- Exercise and Mitochondria
- 'Mitopharma' Targeting Treatments to Mitochondria
- Mitochondria and Stem Cells

Saturday, June 16: Future Treatment Prospects

- New Genes in Mitochondrial Disease
- Anthony Linnane Young Investigator Award
- The Clinical Utility of Polargraphic Analysis
- Diagnosis of Disease: Workshop on High Resolution Respirometry

Call for Abstracts opened November 15, 2006

This event is AMA-PRA credit approved through joint sponsorship through the Univ. of Calif., San Diego, School of Medicine. For more symposium and speaker information, please visit www.umdf.org/symposium/upcomingsymposium.aspx

Catamaran Resort Hotel and Spa,

Medicine 2007 of the Future

Preliminary Family Program June 15-16, 2007

Friday, June 15

- Genetics
- Research Panel
- Mito Basics
- Fundraising 101
- Neurological Manifestations- Adult
- Parent's Panel
- Cardiology and Mitochondrial Disease
- The Many Faces of Grief

Saturday, June 16

- Scientific Meeting Review
- Ask the Mito Doc Panel - Pediatric
- Advanced Mito Basics
- Adapting Your Home and Activities of Daily Living
- Neurological Manifestations - Child
- Ask the Mito Doc Panel - Adult
- Mitococktails, Nutrition and Mitochondrial Disease
- Marriage and Family Stress
- The Art of Letter Writing/Organization
- Respite and Regional Care Centers

**ATTENTION FAMILIES! Did you know that there are many ways to help fund your symposium costs, including government grants?
For more information, please visit
www.umdf.org/symposium/Symposium_Fundraising.htm**

Mission Bay, San Diego, California

Development Corner

UMDF Named a '☆☆☆☆' Charity

UMDF has been named a four-star charity by CharityNavigator.org, a national website designed to help charitable givers make intelligent giving decisions by providing information and evaluating the financial health of more than 5,000 charities.

The four-star rating is described as "Excellent: Exceeds industry standards and outperforms most charities in its cause." UMDF has earned this rating two years in a row. Less than 12 percent of the charities rated received at least two consecutive 4-star evaluations.

For more information, visit www.charitynavigator.org.

UMDF Holiday Card Art Contest

The United Mitochondrial Disease Foundation is holding its first contest for next year's holiday card artwork. The contest is open to children up to age 18. One winner will be featured on the foundation's holiday greeting card solicitation in 2007 and will also receive a gift certificate. Blank holiday cards will also be available for purchase.

The winner will be selected by a group of judges based on a number of factors. The winner does not have to be the best artist in the group. Each participant is also required to write up to 50 words about what their drawing means to them and how they are affected by mitochondrial disease. Participants do not have to be diagnosed with a mitochondrial disorder to enter. Each contestant will receive recognition for his or her work.

The theme of the drawings should show holiday or winter spirit rather than reflect a specific holiday. For more information about the art contest or to download the submission form, please visit www.umdf.org/pdf/ArtContest.pdf.

2006 Grant Recipients

The United Mitochondrial Disease Foundation awarded \$1,025,021 in research grants in 2006. A list of the grant recipients is below. For more information about the studies, please visit www.umdf.org/research/researchgrants.aspx.

Stephane Chiron, University of California-San Diego:

Utilization of fission yeast as a model for mitochondrial

morphology: a new approach to discover novel genes involved in animal cells.

Vishal Gohil, Massachusetts General Hospital: *Molecular signatures of mitochondrial disorders.*

Zaza Khuchua, Vanderbilt University Medical Center: *Animal models of human Barth Syndrome, a mitochondrial cardiolipin disorder.*

John Gordon Lindsay, University of Glasgow: *Enzymatic, assembly and genetic studies on the human pyruvate dehydrogenase multi-enzyme complex.*

Haya Lorberboum-Galski, Hebrew University of Jerusalem: *Enzyme-replacement therapy: A novel approach for treating a mitochondrial disease-LAD deficiency.*

Thomas W. O'Brien, University of Florida: *Mitochondrial ribosomal proteins: candidate genes for mitochondrial disease.*

Michael J. Palladino, University of Pittsburgh: *Developing therapies for mitochondrial disease.*

Doron Rapaport, Institute for Physiological Chemistry, Germany: *Defective biogenesis of mitochondrial beta-barrel proteins as a cause for Mohr-Tranebjaerg syndrome.*

Brian H. Robinson, Hospital for Sick Children, Toronto: *High throughput screening for mitochondrial enhancers.*

Håkan Westerblad, Karolinska Institute, Sweden: *Mechanisms of muscle dysfunction studied in mouse models of mitochondrial myopathies.*

Attention United Way Donors!

You may designate UMDF as your recipient for United Way funds. Call your local United Way office to find out if there is a Donor Choice program in your area. If there is no such program in your area, unfortunately you cannot donate to UMDF through the United Way.

Please remember that all donations given to UMDF through the United Way will have an administration fee taken out by the United Way, so only a portion of your donation will be forwarded to UMDF. We have received funds from a variety of communities through the Donor Choice program and appreciate being included in this effort. A list of United Way Agencies that have DONOR CHOICE is available at www.umdf.org/about_umdf/UW_List.htm.

Appreciating the People Along the Way

While we would like to list ALL our donors in the newsletter, please understand that it would be difficult to do so. If we did, we would have more than 5576 names that donated \$1-99 (THANK YOU); 1241 names that donated \$100-249 (THANK YOU) and more than 390 names that donated \$250-499 (THANK YOU). Please know that we have not forgotten and deeply appreciate each and every individual, organization and corporation that has generously donated money to support our mission. We know who you are and you are appreciated. Your contributions continue to make a difference in the lives of those affected with mitochondrial disease.

The following acknowledgements include **donations of \$500 or more (entered into the system) between January 1, 2006 to June 30, 2006**. If your name or company's name is not listed, please contact kara@umdf.org. We continue to move through a database transition, so we need to hear from you if our information is not accurate!

Benefactor \$10,000 and over Corporations, Organizations and Foundations

Delta Pilots Charitable Fund, Inc.
Edith L. Trees Charitable Trust
Fifth-Third Bank
Matthew Dudgeon Memorial Fund
Precision Performance
Premier Restaurant Management
Rehoboth Beach Writers Guild Assn.
Sage Foundation
The Spartanburg County Foundation
Sterling Foundation Management
Time Warner Book Group
Tishcon, Corp.

Individual/Families

Frank Scavio
Linda & Doug Wilkinson

Patron \$5,000-9,999

Corporations, Organizations and Foundations

Blueline Enterprises, LLC
Shamrock Sports Fest, Inc.
Thrivent Financial for Lutherans
Foundation
United Way of Greater Cleveland

Individual/Families

Joan Evans
Gordon & Shirley Kidd
Gary & Jody Thompson

Guardians \$1,000-4,999

Corporations, Organizations and Foundations

AFNA, Inc.
Atlantic Five Realty
Butler Middle School
George F. Cassell Elementary School
Greystone Power Corporation
Holland Enterprises, Inc.
The IDT Charitable Foundation
Jay Honda
Kenneth & Loretta Schatz Foundation
KFC Inc.
Mayer Electric Supply Company, Inc.
MicroCorp
The Millard Group
Minerva Properties, LLP
Mogadore Moose Lodge 1082
Morgan's Foods, Inc.
P&B Partitions Inc.
Praxair Inc.
Ramsey Elementary School
Teamster Local Union #541
Telephone Operating Systems
Title America Agency Corp

Individual/Families

Marcia & Vincent Cassiani
Russell & Patricia Charleston
Pat Conway
Barry Feinstein
Patrick & Mary Beth Kelley
Kathy & John Kern
Douglas Kerr, M.D., P.H.D.
Warren & Kathy Lammert
Wendy & Scott Link
Marc & Janis McGrath
Todd & M. Elisabeth Pallo
Ronald & Susan Rice
Ronald J. Ross
Teresa Rowe

Tracey & Edwin Spencer
Chuck Steiner
Dennis Weisenberg

Sustaining \$500-999 Corporations, Organizations and Foundations

4M Emergency Systems, Inc.
Allenberry Resort Inn & Playhouse
Andreano's D. & J., Inc.
Archer & Greiner Foundation
Barrand, Inc.
Beverage Builders
Blue Cross Blue Shield KC Credit
Union

Byers Choice, Ltd.
CDM Services Group
Cleveland Browns
Clovis East High School
Collector's Corner
Consigning Closets
Croft Consulting, L.L.C.
Electric Sales Unlimited
Electrical Sales Associates, Inc.
Fidelity Access Networks, LLC
Fusetek
G. Kidd, Inc.
Hampton Road SQN
The Hartford
Hats Off Entertainment Inc.
Horizon Molecular Medicine, LLC
Hull & Associates, Inc.
Hyland Software, Inc.
Jack Peek's Sales, Inc.
James Sassano Associates, Inc.
The Jan S. Moskowitz Revocable
Trust

JE Dunn Construction
The K Company
Klein's Pharmacy and Medical
Equipment
Lanier Middle School
Mary Ann Donut Shoppe, Inc.
Massung Poultry Sales
Max & Erma's
Maxim Crane Works
Meineke Car Care Center
Mitoscience LLC
Motorcars
NATCA Local ZTL
National Air Controllers Assn.
Charitable Fdtn.
National City Corporation
National Property Management
Association, Inc.

New Millennium Wireless
Nordec, Inc.
Northcoast Infusion Therapies, Ltd.
Oak Mortgage Company
O'Leary Bros. Signs and Awnings,
Inc.
OLIS Inc.
Oz Pizza, Inc.
Patriot Communities LLC
Plumbers & Steam Fitters Local 72
Porsche Foundation
Price Management Company
Riffle & Associates, Inc.
RMC Vending
Roadway Express
Rothhammer Sprint Intl. Inc.
Royalty America, Ltd.
Salem Logistics

Salon 54
Serenbe Real Estate
Shirt Tales
The Sign Company, Inc.
S K C Inc. & Employees
South Jersey Engineers LLC
Summit Strength Physical Therapy
Transgenomic, Inc.
Twinsburg Fire Dept.
UCDHC, Dept. of Pediatrics, MS 8313
Unique Concepts
United Way
Wachovia Foundation

Individual/Families

Howard & Anne Adelstein
Steven & Betsy Adelstein
Ruthann Arnold
Stephen & Rosalind Atkins
Molly Auth
Charles Babbush
Thomas Bailey
Franklin & Melanie Baldwin
Kathleen Wagner Barton
Shirley Blaine
Steve & Jean Ann Brock
Sue Ann & Karl Bube
Oliva & Alejandro Buentello
Mark Byrnes
Rick & Robyn Cali
John Carlson
George & Jo Ann Caruth
Celanie Christensen
Stacy & Colin Coburn
Susan Cohen
Kenneth Cox
Jean Daprano
William & Judy Daprano-Rachau
Brian & Stacey Davis
Andrew Deckman
Shannon & Chris DeLacy
Ronald DeLuca
John & Julie DiCecco
Thomas Dickerson
Linda & William Dinkes
Catherine, Nick & Aengus Fanoie
Ralph & Joan Ferguson
Patrick Ferreira
Dale Fisher
Debbie & EJ Fogel
Leslie Friedman
Kenji Fujii
Kent Gordon
Beverly & Brett Hetrick
Bill & Jody Hodges
Beth Madans & David M. Janes
John & Michelle Janusz
Stephen & Jacqueline Johnstone
Glenn & Stacy Kalick
Joanne Kelley
Jerome & Jacqueline Klamerus
Matthew & Kristy Kovalcik
Del & Marge Kreiser
Richard W. Kubach Jr.
Malinda & David Lamarca
Coryn Layfield
Susanne & Richard Leach
Jeraldine Lemacks
Mary Liedloff
Stan Lilleberg
Jack & Joan Lococo
Michael & Shari Lococo
Katherine & Donald Lyons

Stacy & Michael Macaluso
George & Parthana Makropoulos
John & Vicki Marsh
Marjorie Melamed
Richard & Karen Mendelsohn
Russell & Jane Meyer
Ron & Donna Miklos
R.S. Miller
James Mindala
Sister Clara Mohan
Bob & Sandra Montgomery
Linda Nathanson -Lippitt
Godert Noltes
Andy & Julie Norman
Paul Ortman
Steven & Andrea Orwig
David Osolnik
Kenneth Overbey
Kathy Ozan
Gene Pache
Maria & Gregory Parsenow
Christine Pedalino
Robert & Bessie Pelchat
Mary Pisani
Robert Preziosi
Douglas Price
Carol & George Quinn
Leslie & Bernard Reynolds
Jack & Kathy Riley
Sean & Laura Riley
Kathy Rivers
Frederick & Nancy Robertson
Marc & Joan Rubinstein
Jeffrey & Tracey Ruby
Ted & Lynne Rudolph
Arthur & MaryAnn Russell
James & Elizabeth Schmid
Michael Sekara
Daniel & Jennifer Sheridan
Laura & Trey Sheridan
Ramachandra & Veydehi
Shivashankar
Nicholas Simpson
Jon & Jane Smalley
Bob Sudalnik
Anita Surkes
George & Kathy Svilar
Lisa & Jim Swanson
Joyce Temple
Vincent & Blanche Thibodeau
Jennifer Thrush
William & Sherry Thrush
Paul Vajda
John & Rosemary Vangraafeiland
Donald Van Oast
J. Thomas Viall
Diane & Michael Vincent
Joseph Vislocky
Carrie Wachowiak
Krista Wakefield
Declan Walsh
Kristie & James Warner
Elaine & Gerald Weinberg
Morton Weisberg
D.J. & K.M. Welhouse
Bridget Willis
Deborah & Gregory Winfield
Jane & James Wolf
Shan & Lee-Jun Wong
Jim & Janice Yahres

Incidence and Prevalence Rates of Mitochondrial Diseases

Continued from page 5

conclude that either the estimate of 1 in 10,000 is a gross overestimate (and the statisticians did not do their job properly) or that most people with this disorder have not been diagnosed.

The A3243G mutation is likely the most common mutation that is known to cause severe mitochondrial disease. There are about 50 known

mitochondrial DNA mutations that cause diseases, including several mutations that cause LHON (Leber Hereditary Optic

Neuropathy), but many of those persons carrying these mutations may never develop the disorder. Why some people develop the disease and others do not is simply not understood. For a variety of reasons not entirely clear, there are very few persons with LHON that are members of the UMDF. The mutations that cause MERRF and NARP/Leigh are much less common. Most of the remaining mtDNA mutations have been described only in single families. In recent years there have been a number of nuclear DNA mutations that cause mitochondrial disease, but there is no estimate of the prevalence of these

mutations. Based on what we know, it is reasonable to assume that the prevalence of mitochondrial disorders is about one in 4,000 - 8,000, or about 37,000 to 75,000 in the USA. Because the life expectancy of people with mitochondrial disease is so variable, the incidence is difficult to predict based on available data.

Every fifteen minutes, a child is born with mitochondrial disease or will develop one by the age of five.

Before one thinks this discussion is over, we must consider that the majority of persons with a diagnosis of a mitochondrial disorder do not have an identified mitochondrial or nuclear DNA mutation, despite having been tested for these mutations. Many patients come to diagnosis based on established diagnostic criteria using a combination of clinical information, biochemical screening tests, MRI and MRS data, muscle enzymology and results of light and electron microscopy. In most centers that evaluate large numbers of patients, most patients reach a positive diagnosis without having an

identified mutation. With improvements in technology over time, some of these people will ultimately have a genetic mutation identified or the cause of the mitochondrial biochemical and microscopic abnormalities further clarified. However, some experts are skeptical of a certain diagnosis without an identified mutation. Therefore, if we

are conservative and say only one-half the patients with true mitochondrial disease do not have a known mtDNA or nDNA mutation, the

prevalence may double to one in 2,000 - 4,000, or between 75,000 and 150,000 affected in the USA. Of course, these numbers are based only on our personal opinion having reviewed the literature and informally surveyed our colleagues over the years.

Statistics exist for the purpose of making estimates based on available data. For this reason, statistics and their conclusions are open for debate, and other authors may draw different conclusions than we have as to the incidence and prevalence of mitochondrial diseases.

Announcements and Merchandise

A New look for *Mitochondrial News*

UMDF is currently working to develop a new design for the quarterly newsletter. Be on the lookout for a new format in upcoming issues!

UMDF Heartstrings and LEAP Awards

Nominations are open for the 2007 UMDF *Heartstrings* and *LEAP* awards. UMDF wants to recognize our champions and heroes and we need your help. Please see pages 22-23 for applications. All applications must be submitted to the national office by April 27, 2007.

Email Alert

For those of you who have a spam or junk email filter enabled in your email, please add news@umdf.org to your address book as an acceptable address so that you are sure to receive any important information that may be sent from this email address!

Energy Bands

Available in Youth and Adult Sizes

Visit www.umdf.org for more information on how to order or call 412-793-8077.



Awareness Car Magnets

Awareness magnets are also available and can be purchased online at www.umdf.org. The cost is \$5 each or 10 for \$25 (postage and handling included). The magnets are UMDF green with yellow trim and wording.



UMDF Windshirts

The windshirt is green nylon and cost \$30 (includes shipping).

UMDF Throw Blankets

The blankets come in Black, Royal Blue, Khaki, and Forest Green colors and cost \$20



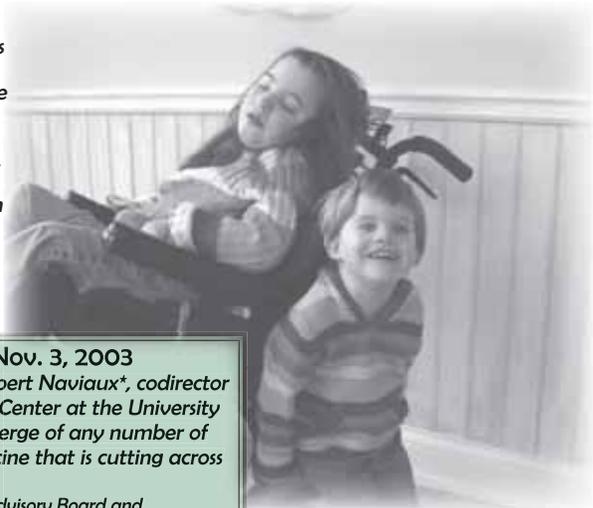
(shipping included). Available soon with new logo. Visit www.umdf.org for more information on how to order or call 412-793-8077.

Keeping *YOU* Informed

Did you know that UMDF has been rated a **FOUR-STAR CHARITY**? In order to keep you better informed of the latest UMDF and Mito news, the next two pages highlight recent media coverage and excerpts from the FY06 Annual Report. Visit www.umdf.org for regular news updates and UMDF announcements!



Caroline Lyman, pictured right with sister Abbey, was featured in the 2003 US News article at age six. She is now nine years old and continues to brighten her family's day with her smile. The Lyman Family has been extremely active with the Ohio Chapter since its inception.



U.S. News & World Report, Nov. 3, 2003
 "Although therapies remain elusive, says Robert Naviaux*, codirector of the Mitochondrial and Metabolic Disease Center at the University of California-San Diego, the field is on the verge of any number of breakthroughs. "This is a revolution in medicine that is cutting across all disciplines."
 *Dr. Naviaux is a member of the UMDF Scientific Advisory Board and Symposium Planning Committee

Asher Gould, pictured below, five-year-old son to UMDF members Anne Reckling and David Gould, was featured in the recent Time article. Anne has been busy revitalizing the Columbus Mito Group and volunteers on the foundation's communications committee.



Time Magazine, Nov. 13, 2006
 "But because defects in mitochondria may underlie an astonishing range of very familiar illnesses, researchers are beginning to suspect that the real number is vastly higher. In the past few weeks alone, reports have come out in Cell, Nature and the Journal of Neuroscience implicating the mitochondria as factors in diseases such as Alzheimer's and Parkinson's. Indeed, says Dr. Vamsi Mootha*, a Harvard Medical School researcher who won a MacArthur Foundation "genius" grant in 2004 for his work on mitochondria, "it looks like they're really important in diabetes, hypertension and many other common diseases – even in the aging process itself."
 *Dr. Mootha is a UMDF-funded researcher.

Time Magazine, Nov. 13, 2006
 "This helps explain why mitochondrial disease occurs in such bewildering variety and why it can be so difficult to diagnose. Even now that it's better understood, parents sometimes face doctors' accusations of deliberately poisoning children to draw attention to themselves."



Mito in the News

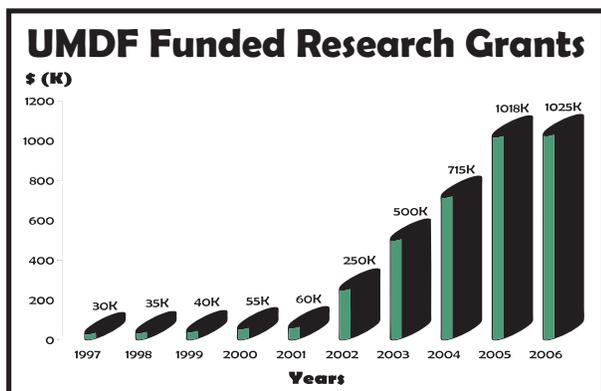
Foundation Progress Report 2005-2006

Our Mission:

To promote research and education for the diagnosis, treatment and cure of mitochondrial disorders and to provide support to affected individuals and families.

Research

The United Mitochondrial Disease Foundation funded more than \$1 million in research in 2006. The Foundation is one of the largest funders of mitochondrial disease-specific research in the world.



In the 2005-2006 grant cycle, UMDF received 211 letters of intent, almost double from the previous year. Research proposals were approximately 94% basic research and 6% clinical research.



Dr. Charles Hoppel, chair of the UMDF Scientific and Medical Advisory Board, applauds UMDF's ability to attract a mix of older and younger researchers, the "salt and pepper."

"They're the pepper in the meeting. They're what make things move forward, the spice of life, if you will. The salt is us old timers who have been around a long time... At the meeting in Atlanta, we had a lot of dark-haired individuals, which is just phenomenal. That's the UMDF. This is an organization that's committed to trying to find ways to learn more about mitochondria with the ultimate goal to find a cure for this devastating set of disorders."

Symposia

Next year's symposium, Mitochondrial Medicine 2007, UMDF's 7th annual symposium on mitochondrial diseases co-sponsored by the Mitochondrial Research Society and the Mitochondrial Medicine Society, will continue to provide the most up-to-date information on mitochondrial disorders to families, researchers, scientists and doctors. The UMDF symposium is the most recognized symposium on mitochondrial disorders in the world.

Services and Support

UMDF has ten Chapters and 14 Mito Groups across the United States.

In March 2006, UMDF pioneered the Ambassador program to serve areas without enough members or support to start a Chapter or Mito Group. At the end of FY 06, there are already 13 Mito Ambassadors nationwide. The MitoConnect program was also launched in 2006 to connect members with others of similar age, diagnosis, symptoms and/or location.

Raising Awareness

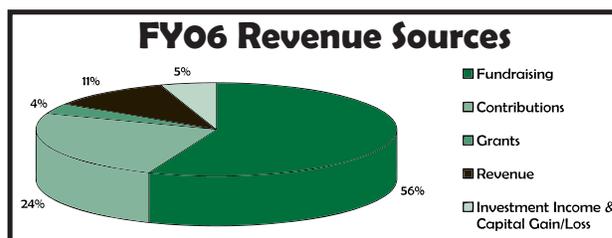
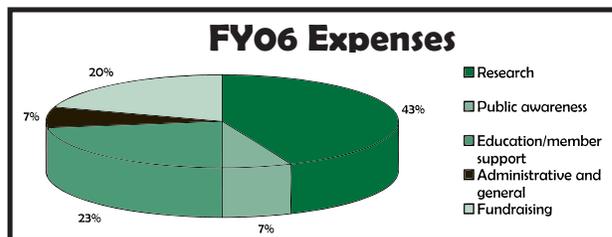
UMDF has been highlighted in more than 60 media outlets across the United States – including Time Magazine, New York Post, Boston Globe and Atlanta's TBS Storyline – gaining national exposure as the leading organization dedicated solely to mitochondrial disorders.

One article featured UMDF Board of Trustees Secretary Sharon Shaw and popular musician and movie star Jack Black:

"Within a year, he did a tremendous amount of work with [UMDF]. He did a House of Blues fundraiser with Tenacious D... And then he went on Who Wants to Be a Millionaire? ...He also donates a percentage of the Tenacious D merchandise."
-Sharon Shaw, New York Post, June 11, 2006

The Foundation is also raising awareness amongst clinicians, pediatricians, general practitioners and other front-line physicians across the country by exhibiting at annual medical meetings and hosting grand rounds, in which a mitochondrial disease expert addresses large groups of physicians at leading hospitals.

In 2006, UMDF supported grand rounds in Missouri, South Carolina and Illinois and plans to support five more in 2007.



***Mito Adults Corner* - Getting a VPAP: A Personal Odyssey**

by *Melissa Nixon*

For as long as I can remember, I have been unable to breathe properly when laying down, especially if positioned on my back. As a child I coughed night after night, unable to breathe deeply or clear secretions. Vivid nightmares and gasping wake-ups with a pounding heart and headache were facts of life. Every cold seemed to become a major medical event, and I spent weeks dozing in a plastic tent filled with medicated vapors every winter. I nearly died from rapid pneumonia. My brother suffered similar issues, and we were told he only had 40% functional lung capacity.

As a teenager, I was unable to participate in physical education, missed school for weeks on end and was told I had chronic bronchitis despite never having smoked. Nobody realized the scoliosis might be making the breathing even more difficult. My tossing and turning at night, as well as the coughing, kept my mother awake until her frustration erupted into abuse. I took to sitting on the floor next to my bed through the long nights and catching cat naps in the school library during the days.

After I married, it was not long before my husband moved out of the bedroom - even in a king sized bed he could not get any sleep as I struggled to get through each night. I began working a half shift so I could catch a few winks after dawn, the only time I seemed able to sleep at all. Doctors tried psychiatric drugs and sleep aids to no avail as my diagnosis evolved from ALS to MS to dystonia.

As I entered my forties, I finally had to retire. I was diagnosed with cardiac and respiratory failure, but even the six liters per minute of oxygen my doctor prescribed just did not seem to help. Flowing any amount of oxygen past my nostrils just frustrated me as I struggled to breathe any of it in, even when sitting propped up in a hospital bed at home. More and more medications were prescribed and more and more cardiac unit hospitalizations

occurred, but the respiratory failure continued basically untreated.

In 2000, two major events happened: I lost my beloved brother, and I discovered the Internet. Utilizing my medical knowledge as a retired veterinarian, I became determined to name the disease that had devastated my family. Within months, I became convinced it was mitochondrial cytopathy. On my 46th birthday, I found Mitoldies.

Finally, I had found other people who could actually understand my terrible sleep issues. By this point, I was having nightly chest pain and myoclonus; I went night after night, day after day, with no sleep because I simply could not breathe. A list-leader told me about something called a BiPAP.

Enquiring of my respiratory therapist with the oxygen supplier, I was told there were lots of BiPAPs on the shelf in his office, but he knew little about them. He suggested I get a sleep study.

My doctor willingly ordered the sleep study, but it turned out to be its own kind of nightmare. Told to lay flat on my back with medical equipment running up both nostrils, I could not get my breath. Sleep was impossible and I suffered terrible myoclonus all night as my muscles screamed for oxygen. In the morning, they told me the values were unbelievable so their equipment must be malfunctioning, and by the way, I did not have restless leg syndrome.

A second sleep study was scheduled, this one to run through the following day since I could not sleep at all until after dawn. I was propped up in a bed with pillows and oxygen was run into my nose at my prescription level of 4 L/min with special non-occlusive monitoring equipment. I slept for 30 minutes and since my CO₂ levels remained within normal limits, I was woken up and sent home. A week later my husband received a call informing him that I was faking my symptoms and had no medical sleep problem. Medicare was

billed for two complete sleep studies.

Angrily, I made the trip to the home office of my respiratory supplier and demanded to speak with their head respiratory therapist. I believe that, by taking control at that point, I saved my own life. As it turned out, since my diagnosis at that time was mitochondrial myopathy (clinical diagnosis) with secondary dystonia and hypokalemic periodic paralysis, any one of which was a qualifying neuromuscular condition, by Medicare regulations I needed only an overnight oximetry at home with my prescribed oxygen. If my oximetry records showed oxygen values below a specific level for longer than a specific time period, I would qualify for a BiPAP. Wow.

The oximetry was ordered through an independent testing laboratory and accomplished within a week. Once the values were downloaded from the unit the day after testing, I received a call that changed my life. My oximetry values were so low that they were surprised I had stayed alive long enough to get the testing done. I had my first BiPAP the next morning.

As a disabled veteran, my husband receives his medical care through the VA system. He is currently undergoing a sleep study that seems very leading edge. The sleep lab first shipped a package with oximeter, respiratory effort detector and airflow monitor plus instructions. He wore the equipment for three nights and shipped it back. Since abnormalities were found, he will be trying a CPAP for four nights this week. I am happy with this approach, as it allows the testing to occur in the home environment, and – pertinent in our case – at the usual geographic elevation.

Unfortunately, after I received my first BiPAP, I was still dealing with a local respiratory therapist who knew little about BiPAPs and nothing about fitting masks. My primary care doctor was as cooperative as I could wish but acknowledged I was his first and

Continued on page 20

only BiPAP patient. Still, the very first time I nestled my face into that mask and lay down to sleep, I knew we were on the right path at last.

Subsequently, I have seen several different respiratory therapists, two pulmonologists and a new primary care practitioner after my previous one retired. I have changed medical equipment suppliers twice. I am now using my third style of VPAP. I have learned that BiPAP is a brand name, while VPAP is a more general term for a variable pressure unit.

Eventually, my muscle biopsy showed ragged red fibers, thus confirming the Mito diagnosis. Further tests at a university respiratory laboratory have shown that I have a mismatch of ventilation and perfusion (normal carbon dioxide with low oxygen) as well as hypoventilation due to problems with my diaphragm muscle, chest muscles and the part of my brain that tells me to breathe. I also have trouble with the tissue in my throat collapsing during REM sleep. Because part of my problem is at the brain level, I do not always remember to breathe, even when awake. For that reason, I use a VPAP-ST that triggers a breath for me if I do not initiate one during the set time period.

Surely, without the benefit of variable positive airway pressure, I would be extremely ill, if not dead, by now. One respiratory therapist took the time to explain to me that with my condition a CPAP could be deadly and I should be sure that I never again try to use one. Well, that certainly explained my feelings of suffocation during a hospitalization that involved being strapped down on my back with a CPAP on my face!

Last year, my Medicare-provided VPAP machine began behaving strangely and after consultation with my respiratory therapist, I learned that the problem is called chattering. He recommended a volume ventilator. However, since I owned a different model of VPAP as a back

up that still worked well for me, my supplier agreed to order that particular model for me to use routinely. It seems to have a better sensitivity to my respiratory efforts and a more laminar, less turbulent, airflow.

Like many patients on respiratory support, I use a heated humidifier between my VPAP and the hose leading to my mask. I also keep saline nasal wash nearby to help moisten my nasal tissue and keep the stuffy nose problems at a minimum. I make a special effort to stay well hydrated in the hours before bed.

My non-Mito friends joke about my habit of brewing fresh espresso coffee at midnight every night, but I find the vapors help me breathe and that drinking a cup or two of coffee further improves my breathing. I know from my veterinary training that prescription medications with active ingredients similar to coffee strengthen the diaphragm temporarily and help the brain remember to breathe, so I just laugh right back at my friends and tell them maybe they should try coffee as a sleep aid.

While I still do not sleep perfectly every night, I rest comfortably for the most part, free of myoclonus and nightmares. Because I have cardiomyopathy as part of my Mito, I do still have some episodes of pulmonary edema and tachycardia at night, but they come less often these days. I sleep on my side, propped up on pillows, to reduce these cardiac problems as much as possible. Since my first BiPAP arrived five years ago, I find my life has improved dramatically. My muscle function is better during the day, my ability to think clearly has returned and I require far fewer prescription medications. I no longer get sick for weeks with every cold virus in town.

I love my 2.7-pound VPAP. I have the cable that connects the VPAP to my car cigarette lighter for power so that I can use it while driving or riding in the car. I also have a five-pound battery that provides several hours of power and a 25-pound

battery that will keep me breathing through a night without electrical power connection. I can fit the VPAP, the smaller battery and a small oxygen tank with regulator into a special backpack developed by one of my respiratory therapists.

While I still have a prescription for oxygen both day and night, I find I rarely need it during the day except when traveling at higher altitudes or talking. I get much greater benefit from an hour on my VPAP, especially if I take the time to do stacked respirations first to re-expand my lungs. At night, the oxygen is fed into my respiratory system via a valve positioned at the junction of the humidifier and the hose leading to my mask.

I am so very thankful for the support of my friends in the mitochondrial community. With the free exchange of information we enjoy, I know my life will not only last longer but will be lived better. I am so thankful to live in the era of the Internet, and I will be eternally grateful to the inventor who created the first BiPAP sometime around 1993.

***Calling All Mito Adults:
Mitochondrial News
Needs YOU!***

**Please consider
submitting an article on
your experiences with a
specific topic of interest.
If you want to contribute,
please email Kara
Strittmatter at
kara@umdf.org or call
412-793-8077, ext. 114.
We look forward to
hearing from you!**

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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 UMDF, enabling 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 fund raising projects, the judges will consider the uniqueness and creativity of the project, communication, the 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. UMDF will announce the winner at the annual symposium and will present the winner with a plaque. The Heartstrings Award winner will be featured on the UMDF web site and recognized in the UMDF Mitochondrial News newsletter.

In 100 words or less, please explain how this individual has “tugged at your heartstrings” through fundraising for or donation to 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, letter sent with the donation, and so forth).

Please type your essay and attach it to the nomination form. Mail the nomination by April 27, 2007 to:

**Heartstrings Award
UMDF
8085 Saltsburg Road, Suite 201
Pittsburgh, PA 15239**

Or fax to 412-793-6477 or email the nomination by April 27, 2007 to info@umdf.org.

Person Nominating

Name: _____

Address: _____

Phone: _____

Email: _____

Nominee for the Award

Name: _____

Address: _____

Phone: _____

Diagnosis (if known): _____

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

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. UMDF will announce the LEAP Award winner at the annual symposium and will present the winner with a plaque. The LEAP Award winner will be featured on the UMDF web site and recognized in the UMDF Mitochondrial News 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 nominees is involved with.

Please type your essay and attach it to the nomination form. Mail the nomination by April 27, 2007 to:
LEAP Award
UMDF
8085 Saltsburg Road, Suite 201
Pittsburgh, PA 15239

Or fax to 412-793-6477 or email the nomination by April 27, 2007 to info@umdf.org.

Person Nominating

Name: _____

Address: _____

Phone: _____

Email: _____

Nominee for the Award

Name: _____

Address: _____

Phone: _____

Diagnosis (if known): _____

Age (must be at least 14 years old): _____

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We're Here to Help You! 412-793-8077**

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UMDF MISSION

To promote research and education for the diagnosis, treatment and cure of mitochondrial disorders and to provide support to affected individuals and families.

Deadline for the next issue is January 31, 2007.