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United Mitochondrial Disease Foundation

MITOCHONDRIAL NEWS

Volume 9 • Issue 3 • Summer/Fall 2004

Mitochondria and Alzheimer's Disease: A Powerful Link

*Paula I. Moreira, MSc and
George Perry, MD*

*Institute of Pathology, Case Western
Reserve University, Cleveland, Ohio*

Mitochondria convert the chemical energy from food into ATP, which cells use to function properly; in short, they are the powerhouses of the cell.

They are about the size of bacteria but may have different shapes depending on the cell type. They are membrane-bound organelles and, like the nucleus, have a double membrane. The double membrane divides mitochondria into two distinct parts: the intermembrane space and the

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Mark your Calendars NOW!
Mitochondrial
Medicine 2005
Gateway to a Cure

Scientific Sessions:
June 15-17, 2005

Family Sessions:
June 17-18, 2005

Clinician Sessions:
June 18, 2005

**Hyatt Union Station Hotel
St. Louis, Missouri**

**Scientific Program Chair
Bruce H. Cohen, MD**

**Check www.umdf.org on
December 1, 2004 for**

Call for Abstracts

Promoting Research for Cures and Treatments . . .

Providing Support to Patients and Families . . .

Mitochondrial Medicine 2004

Streams of Energy

"I am writing in order to say thanks to you for organizing such a great meeting. For me, it was the first time that I actually got in touch with parents of children affected by mitochondrial diseases -- this made a lasting impression on me. As a basic scientist working only at the bench, I do know that my research eventually will benefit patients, but putting names and above all faces to it, is different.

I was and I still am emotionally touched, and this adds a lot of additional motivation to do hard bench work, which unfortunately is too often not as successful as one wishes and therefore always comes with a great deal of frustration.

Attending this meeting was a unique and very valuable experience for me and I am looking forward to St. Louis."

*With best regards,
Volkmar Weissig, PhD, ScD*

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 -- 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: Salvatore DiMauro, M.D., Columbia University, NY; and Russell P. Saneto, D.O., Ph.D., Children's Hospital and Regional Medical Center/University of Washington, Seattle, WA.

The Question is:

Early this year I underwent surgery to plant scaffolding into my arteries [stents x 2] because of coronary cholesterol. I was actually in the hospital in the pulmonary unit when I became unwell and was transferred to the coronary unit where I underwent testing. A short while later I had this operation as above. A few weeks later back at the pulmonary department I had another period of sickness and was sent back to the coronary department for observation and eventually another operation to confirm that I was ok. I was told I needed medication for cholesterol and that statins were not good for me as they would destroy muscle which is the last thing I need. I am now waiting research to establish the best form of medication for me.

Response From: Salvatore DiMauro, M.D.

Statins do not actually "destroy muscle," but in some individuals and at certain doses they do seem to damage

muscle, as manifested by exercise intolerance, muscle pain, and increased levels of serum creatine kinase (CK). In rare occasions, muscle damage has been extensive, resulting in myoglobinuria (discoloration of the urine due to the passage of the muscle pigment myoglobin). So, as often in life, one has to find a balance between risks and advantages. If - as it sounds - your cholesterol problem is serious, statins are wonderful drugs. If you take them, however, you should also take coenzyme Q10 (CoQ10) by mouth because CoQ10 levels in blood decrease during statin treatment. Also, have your blood CK checked at regular intervals.

The Question is:

I would like your thoughts on the use of a Ketogenic Diet for seizure management. While my daughter's seizures are fairly well controlled by Keppra 1000 mg bid and Neurotin 900 mg tid, she is sleepy and lethargic throughout the day; some days sleeping 3 hours in school. We miss seeing our daughter interactive and excited by life. This is something we are looking into with the support of the pediatric neurologist with the hopes we can reduce her meds and wake her up.

She will be evaluated to determine if a Ketogenic diet is appropriate for her in July. I have been reading about it and am now wondering if it is a reasonable consideration. The limited carbs and protein, fluid restriction, the possible low blood sugars, etc. make me concerned we may tip the balance on a stable child.

Please share your ideas on the positive and negative aspects of this treatment in a child with Leigh's, clinical diagnosis not confirmed through testing done 14 years ago.

Response From: Russell P. Saneto, M.D.

I am sorry that your daughter's seizures are difficult to control. Whether to try the ketogenic diet depends on several factors:

(1) The diet is very regimented, meaning that strict account of all foods eaten is required. The diet is very bland in taste; although there are many ways to enhance taste in the diet, it still remains somewhat bland. This may cause a child not to eat the diet, which defeats the

purpose of the diet.

(2) If a child is mobile and capable of getting into other foods in the refrigerator or pantry, then this would sabotage the diet and could enhance seizure activity. We have seen patients go through the garbage to find food as well as even eating their pet's food.

(3) The diet requires a commitment of the family. This is necessary as other siblings will need to be aware that certain foods are "off limits" to the child on the diet. Sometimes other foods not on the diet will need to be out of the vision of the child so he/she is not aware or tempted to eat the forbidden food, i.e. cookie.

(4) A full metabolic workup is needed to make sure that the child does not have a beta oxidation problem (metabolizing fats for energy).

(5) Most ketogenic diet centers would like the patient to stick with the diet for at least 4 - 6 months for an adequate trial.

(6) There are some children who just do not tolerate the diet -- vomiting, severe constipation, or lack of seizure abatement.

I have good success with the ketogenic diet and mitochondrial disease, with respect to seizure control. My patient population consists of electron transport chain complex dysfunction, mitochondrial DNA mutations, and pyruvate dehydrogenase deficiency. Most of my patients are on the 4:1 diet but several are at 3.5:1 or 3.75:1 ratios. The variation in ratios is due to the caloric needs of the patients and ketone levels. Most are growing well, albeit a little slower than their peers (most of my patients are infants and young children). I have not had difficulties with increased lactic acid levels or patients becoming too hypoglycemic on the diet (after the initial phase of starting the diet). Seizure frequency has been from seizure-free to significantly reduced (>75% seizure reduction) in my patient population. Unfortunately, most have continued on their seizure medications. The majority of parents tell me that their child is brighter or at least more interactive on the diet. I think the reduction in seizure frequency has a large part in this subjective view of the parents. I hope this helps in your decision-making process.

Chairman's Report

"Streams of Energy," the 6th symposium on Mitochondrial Diseases, co-sponsored by the Mitochondrial Research Society, the Mitochondrial Medicine Society, the Pittsburgh Mercy Health System and UMDF has brought, once again, the most up-to-date information on mitochondrial disorders to families, researchers, scientists and doctors. The UMDF office has received more positive comments from the largest number of attendees this year than ever before. UMDF Symposia are becoming the recognized symposium on mitochondrial disorders in the world.

"We are now as good as major conferences in other fields of medicine with a novel format of clinical and basic science all working towards the goal of understanding and curing mitochondrial disease. I think that this meeting is coming of age."

**Richard H. Haas, MB, BChir
UCSD, San Diego, CA**

In 1995, Matt and Lee Neff of Indiana decided that something had to be done to convey information and to educate families and professionals about mitochondrial disease, so they organized the 1st symposium on mitochondrial disorders one year before the formation of the UMDF. This gathering of parents and professionals revealed the need for annual symposia as well as providing the opportunity for the creation of the UMDF. From this beginning, UMDF symposia have traveled to Philadelphia in 1997, Ohio in 2000, San Diego in 2001, Dallas in 2002 and Pittsburgh in 2004, with our next stop in St. Louis, June 15-18, 2005.

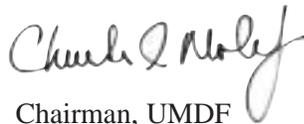
Have we had an impact on treatments and cures? Absolutely!

Attendance at symposia is increasing, more information is being exchanged between professionals and families and we are realizing more interest in research with better science being funded. Membership in UMDF is growing with over 24 support groups and chapters across the United States, Australia, and Canada.

UMDF continues to re-define hope for thousands of individuals who have been told there is no hope. UMDF continues to create a language of hope that empowers rather than restricts, that helps people become participators rather than spectators, that creates awareness not silence, activity not complacency. UMDF is helping acknowledge rather than deny, convincing people to commit, not quit, and to try again, not give in.

When everything in our life is wrong it's time to take inventory of everything in our life that is right. Hopelessness is what happens when we allow ourselves to become helpless. UMDF re-defines hope by turning tragedy into tribute and tribute into triumph. UMDF re-defines hope.

Yours toward a cure,



Chairman, UMDF



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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

Chapter Activities

NEW YORK METRO CHAPTER

Manhattan, NY

President: Tom Shubeck

Phone: 203-287-0655 (Mary Pisani)

Email: NYMetroChapter@umdf.org

Walk to Create Awareness raises \$5,990.92 in Connecticut.

In honor of her son, Matthew Pisani, Mary Pisani and her family have worked diligently each year to organize this annual walk. Keep up the great work!!!

Upcoming Events:

- The Annual Nunno Dinner -- Friday, October 22, 2004

Coins for a Cure

In honor of Todd McCartha of Rome, NY, Bellamy School raised \$360 to benefit UMDF. Thanks Kids!

NEW ENGLAND CHAPTER

Boston, MA

President: Bridget Willis

Phone: 413-593-5920 (Beverly Ingram)

Email: NEngChapter@umdf.org

Coins for a Cure

In Honor of Meaghan Brown, Miller Elementary in Holliston, MA raised \$130.25 to benefit UMDF.

Lego Walk raises \$985

Friends of Cameron Picnic raises \$8,572 in July

Energy for Life Campaign

The following businesses and individuals participated in the Energy for Life Campaign in Memory of Matthew James Bailey, and raised \$1,287.50: North Fork Bank, Children's Speech Services, Inc; Barbara Thompson; Salvatore & Germaine Vindigni; and Maureen Johnston. Matthew's family resides in Ellington, CT.

Upcoming Events

- October 3, 2004 -- Mito-What? 5K Run/Walk, Longmeadow, MA



KANSAS CITY CHAPTER

Kansas City, MO

President: Heidi Harmon

Phone: 816-554-8530

Email: KCChapter@umdf.org



1st Annual 5K Mito-What? 5K Race and 1 Mile Walk Raises More than \$60,000

Kansas City Chapter's First Annual "Mito What?" 5K Run and 1 mile walk was held on June 26th, 2004. Great work, Kansas City! The Chapter will start planning the 2005 Walk/Run in October. Please be thinking about any committees you might want to be part of and contact Heidi for more information.



Chapter and support group leaders participated in focus groups during the 2004 conference in Pittsburgh. The focus group information will be used in the UMDF Strategic Plan to move us *One Step Closer to a Cure!* We value our members' input!!!!

SOUTHERN CALIFORNIA CHAPTER

Lakewood, CA

Vice President: Kathy Fares

Phone: 858-271-9000

Email: SCalChapter@umdf.org

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!**

ARIZONA CHAPTER

Phoenix, AZ

Interim Leaders: Sharon Shaw, Thom Montgomery and Jane Shumaker

Phone: 480-563-8562

Email: AZChapter@umdf.org

Chapter Activities

OHIO CHAPTER

Cleveland, OH

President: Jennifer Lyman

Phone: 330-929-4430

Email: OHChapter@umdf.org

Coins for a Cure

In honor of Ann Marie Rady, St. Rita's School raises \$194.

Mitochondrial Medicine Seminar

The chapter hosted a seminar with featured speaker, Bruce H. Cohen, M.D., at Silver Lake Country Club on October 10th.



3rd Annual KFC/UMDF 5K Run/Walk raises more than \$150,000

Friends and family members enjoyed a day of triumph once again at the 3rd Annual KFC/UMDF 5K Run/Walk in Cleveland Heights, OH. The sun was shining and spirits were high as walkers and runners pulled together to move UMDF *One Step Closer to a Cure*.



Ohio Chapter Treasurer, Lisa Arnold, huddles with her family at the run/walk -- including the newest addition to the family, Joey, who was only one week old at the race.

INDIANA CHAPTER

Indianapolis, IN

President: Sue Ann Bube

Phone: 317-894-9099

Email: INChapter@umdf.org



John Flack, Bob Thomas and Bob Jenks took an incredible journey from Indianapolis to Pittsburgh in August. These heroic men raised more than \$19,900 to benefit UMDF and caught the attention of local media across Ohio and into Pennsylvania. Outstanding, gentlemen!!!

See page 9 for more details on the Miles for Mito Journey!

Atlanta Area Group Continues to Grow



The Atlanta Area Support Group raised more than \$6,300 during a special dinner with Dr. John Shoffner and Chuck Mohan as guest speakers this past May. Over 100 guests attended the event, held at the Peachtree Club in Midtown Atlanta.

Upcoming Event

- November 15, 2004 - 1st Annual Fore-A-Cure Golf Tournament, Stone Mountain Golf Club, GA

DELAWARE VALLEY CHAPTER

Philadelphia, PA

President: Maripat Shelly

Phone: 215-256-0273

Email: DelValChapter@umdf.org

Monte Carlo/James Bond Casino Night - raises \$3,120 to benefit UMDF.

Coins for a Cure

In honor of Abel & Averal Cruz, Dr. William Mennies School raised \$978.87.

Upcoming Events

- October 6, 2004 - You Go Girl Golf Outing
- November 6, 2004 - Fashion Show



1st Annual Brew at the Zoo raised \$8,500

Through the efforts of Lisa and Bob Polsky, and the Delaware Valley Chapter, the first annual "Brew at the Zoo" was quite the success.

Held at Elmwood Park Zoo, participants enjoyed sandwiches, chicken wings, soft pretzels, iced tea and bottled water. And, of course, Beer Tasting!!! Music was provided by Soniqboom and everyone in attendance had a great time! Thanks Lisa & Bob!!

Planning for a Gift

By Nick Nicholson

The first concern to be evaluated -- before any consideration of charitable giving can be contemplated -- must be "How will this impact me; does it fit with my plans for the future, for my children's future?"

Comprehensive financial analysis and planning is needed to give you that answer well before making any gift of a substantial nature. By virtue of tax incentives, yes, paradoxically, you could end up with more by giving some away, but this is not always the case for everyone. The question is even more important for those who may not improve their position monetarily but still want to give.

That's why UMDF has launched the "Taking Control" initiative and is providing access to The Monteverde Group and the resources of Partners Financial for members and donors.... access to expert assistance, coupled with complete confidentiality.

In many cases, charitable giving can not only fit your plans, it can enhance them, increasing cash flow and improving your bottom line. That's one of the primary goals of the program.

For others, substantial gifting may not currently be appropriate or feasible, but does that mean "Taking Control" is not an option... not an opportunity?

The answer is an unequivocal "No"!

"Taking Control" can benefit any UMDF member or contributor. Some things, including solid financial planning principals, are universal. The more we can save you, or the more we can help you to make and to keep, the better able you'll be to help in some way.

If not now, then possibly in the future.

Here's an idea based on two such widely accepted principals. First, "Never outlive your assets", and second, a well-balanced portfolio is important to retirement, to your future financial health. You should have so much in safety of principal, so much in income-producing vehicles, growth and income, growth and possibly aggressive growth positions.

And if you're never to outlive your resources, some funds must be considered "assets of last resort".... funds you never plan to touch, just in case you "live too long." These funds should be safe, earn at least some interest (to keep up with increased costs in the future), and since relatively safe interest income is almost always lower than expectations for higher risk investments, tax advantaged.

What if you could fund a vehicle offering minimum guarantees, tax advantages, and cash accumulation you could access should you need it? And what if, when you've planned well and never needed these "assets of last resort", money would pass to your children -- tax free

-- and still generate a gift to UMDF?

Find out how this, or other strategies, might work for you.

The worst case scenario is that a "Taking Control" analysis will show you to be one of those rare cases in which everything has been done as best it can be, with no viable alternatives of interest. In that case, you'll have received the benefit of a considered second opinion confirming that fact. Best case scenario.... it works for you and UMDF.

Call us.... It's in all our best interests.



Nick Nicholson, pictured above at the 2004 Mitochondrial Medicine Conference in Pittsburgh, is a Senior Financial Advisor with The Monteverde Group, a member of Partners Financial, and is a Registered Representative and Investment Advisor Representative offering securities and advisory services through NFP Securities, Inc. of Austin, Texas. Nick is our contact person, and is spearheading the development of all components of the "Taking Control" initiative sponsored by UMDF. He has been appointed to develop, coordinate and implement the necessary planning, strategies, and financial instruments for our members. He can be reached by phone at 800.722.0098, or via e-mail at nnicholson@monteverdegroupp.com

United Way and You

Many companies require United Way giving - why not take this as an opportunity to support UMDF and its new 5 year grant initiative. Start off by asking the employees you work closely with and maybe they'll help spread the word.

How to make it work . . .

The United Way campaign in your local area is designed to meet a diversity of health and human care needs. You may give an unrestricted gift and, in some areas, you may designate UMDF as the recipient of your gift to United Way. Contact your local United Way office to find out if there is a DONOR CHOICE program in your area. We have received funds from a variety of communities through the Donor Choice program and appreciate being included in this effort.

We will acknowledge each gift received unless otherwise indicated or if no address is provided.

Simply complete the appropriate form (available through your employer) as follows:

**United Mitochondrial
Disease Foundation**
8085 Saltsburg Road, Suite 201
Pittsburgh, PA. 15239
Phone: (412) 793-8077
Fax: (412) 793-6477
Attention: Toni Beasley

Matching Gifts . . . Your gift could be doubled!

Does your employer participate in a Corporate Matching Gift Program? If so, your gift could be doubled. Please obtain the appropriate form from your comptroller or personnel office using the same contact information as you would for a United Way designation.

Attention Federal
Employees!
You can now donate
to UMDF through the
Combined Federal
Campaign. UMDF
has been approved
under the CFC Code
2921 for 2004.



Book for Sale

Shawnda Kizzie, mother of Ky'monie Onyx Kizzie, wrote a book called *She's An Angel Now* to tell her story about her daughter and the disease that took her daughter's life. The book can be found at www.authorhouse.com.

Do you ever shop online?

If so, you could be raising money for the UMDF at no extra cost to you! Several free websites will allow you to do your online shopping through them and will donate anywhere from .5-15% of each purchase to the charity of your choice. There are hundreds of online stores you can shop at (from GAP, to Barnes and Noble, to Sears) and part of the money will go to the UMDF-at no extra cost! So if you shop online, think about registering at one of these sites and having a portion of your purchase go to the UMDF.

Three websites that include the UMDF as a possible beneficiary include:

www.benevolink.com
www.igive.com
www.mycause.com

***Start your holiday shopping
and benefit UMDF at the
same time!!***

Couldn't Attend Mitochondrial
Medicine 2004?
Order Tapes/CDs and DVDS

**Order forms are available at
www.umdf.org/conferences or
call 412-793-8077 to have forms
faxed or mailed.**

Dr. Amy Goldstein, of Pittsburgh, served on the Family Program Committee for the Mitochondrial Medicine 2004 Conference and was essential in securing speakers for the clinical sessions. She also spearheaded and hosted the Center for Creative Play outing on Thursday night of the conference for our families. You are truly an inspiration, Dr. Goldstein -- we cannot thank you enough!

Special Thanks to Dr. Amy Goldstein!



Dr. Goldstein, 3rd from the left in the first picture, enjoyed spending time with the families in attendance -- both young and old had a wonderful time!

Mitochondria and Alzheimer's Disease: A Powerful Link

Continued from page 1

mitochondrial matrix. Several steps in cellular respiration occur in the matrix due to its high concentration of enzymes. Mitochondria are semiautonomous in that they can divide and grow to make more of themselves. They also have their own DNA and ribosomes.

Mitochondria are essential for neuronal function because the limited glycolytic capacity (production of energy without the presence of oxygen) of neurons make them highly dependent on mitochondrial aerobic oxidative phosphorylation (production of energy linked to respiratory chain) for their energetic needs. However, oxidative phosphorylation is a major source of endogenous toxic free radicals (harmful species), including hydrogen peroxide (H₂O₂), hydroxyl (HO-) and superoxide (O₂-•) that are products of normal cellular respiration. Besides the key role of mitochondria in the maintenance of cell energy, these organelles are also involved in cell death pathways.

Alzheimer's disease (AD) is a progressive neurodegenerative disorder in which nerve cells in the brain die. It is the fourth leading cause of death in the USA, affecting more than 4 million people. AD is devastating, and, despite great strides in recent years, still puzzling in its cause and mechanisms. AD patients suffer loss of neurons in large areas of the brain particularly concentrated in the lobes of the brain that control some of the higher intellectual functions. Two important hallmarks of AD are the extracellular accumulation of amyloid-β (Aβ) aggregates, or plaques, and intracellular accumulation of hyperphosphorylated tau protein (neurofibrillary tangles).

The pathology seen in AD patients involves oxidative stress events, leading to neuronal degeneration. Several studies have shown that oxidative stress is involved not only in damage to the proteins of neurofibrillary tangles and plaques but also involves extensive damage to the cytoplasm of neuronal populations vulnerable to death during AD

evolution (Smith et al., 1996; 1997; Sayre et al., 1997).

If the amount of free radical species is greater than antioxidant defenses (endogenous protective mechanisms that can be enzymatic and non-enzymatic), oxidative stress occurs, followed by mitochondrial dysfunction and neuronal damage. Furthermore, it has been proposed that the presence of increased oxygen free radicals and their accumulated effects during aging may contribute to AD.

Reactive species generated by mitochondria have several cellular targets including mitochondrial components themselves (lipids, proteins and DNA) (Perry et al., 2002). The lack of mitochondrial repair mechanisms renders mitochondria an easy target for oxidative stress events. Oxidative damage to mitochondrial function might explain why the risk of AD increases with age and why there has rarely been evidence that the disease has a familial pattern of mitochondrial inheritance.

Using diverse methodology, we determined whether mitochondria could be involved in AD pathophysiology because they can be both targets and sources of reactive oxygen species (Hirai et al., 2001; Castellani et al., 2002). We found that neurons showing increased oxidative damage in AD possess also a striking and significant increase in mitochondrial DNA (mtDNA) and cytochrome oxidase (an important enzyme involved in the respiratory chain). MtDNA and cytochrome oxidase are found in the neuronal cytoplasm. Cytochrome oxidase is also found in vulnerable cells, in the vacuoles containing the fatty pigment, lipofuscin (a conglomerate of lipids, metals, organic molecules and biomolecules that accumulate with aging and result from the degradation of mitochondria and other cell organelles).

Continued on page 15

Miles for Mito



Most parents would be willing to go to great lengths to help their sick child -- but a 400 mile bike ride? It seems extreme for even the most dedicated parent. Still, that's how far Bob Thomas, Vice President of the Indiana Chapter and father to Gabe Thomas, went on July 31, 2004. Bob and his team--fellow riders John Flack and Bob Jenks--left from Marion County Fairgrounds in Indianapolis and rode their bicycles through central Ohio all the way to Pittsburgh, Pennsylvania. They arrived at the Westin Convention Center Hotel on Thursday night, August 5, of the Pittsburgh conference and were greeted by UMDF

families! Special thanks to numerous Holiday Inns on the route who donated lodging!

The cyclists made several stops on the way and by the time they reached Pittsburgh, raised more than \$19,900 to benefit UMDF! The Indiana Chapter initiated the event and were responsible for more than \$13,000. In Bellefontaine, OH, Joni and James Holycross, parents of Collin Holycross, held a buffet-style dinner catered by Whitmore House at the First Lutheran Church and brought in more than \$1,800 for the Miles for Mito cause. Thank you all for a job well done!

A local physician in Bellefontaine called wanting more information about the disease and contacts to make. He felt that he may have a patient that needs further evaluation in that regard. Right or not, he only was thinking of that after having read the article in the Examiner about the Miles for Mito. I would say that makes Bob's trip a success in getting more awareness out there.

- James Holycross

Tomato Face Foods - Inspiration Beyond Words



Tomato Face Foods Make Special Presentations at 2004 Symposium

An inspiration to all in attendance during the Saturday luncheon, Dana Segal, daughter of Barbara Bruck and Dr. Allen Segal, gave a heart warming tribute to her mother and made a plea to the audience to help her with a campaign she has been working on over the past year -- to get her mom's story on the Oprah Winfrey Show.

After Dana's beautiful speech, Barbara presented the first Tomato Face Foods awards and a check for \$1,000 to the UMDF in honor of Caroline Lyman and In Memory of Gina Marie Mohan. The Lymans and the Mohans have touched their hearts in so many ways - this was their way of saying thank you.

Allen did his part too during the luncheon -- he requested help from the audience in expanding their market. **YOU can help Tomato Face Foods** continue reaching thousands across the U.S. Do you know someone in your local supermarket, school cafeteria, college food service, or grocery chain? If so, please contact Barbara and Allen at 216-382-0232 or seebrook1@aol.com.

Dana recently started working on a tennis fundraiser too! This is one amazing family!



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DSW

FILENE'S
BASEMENT

VALUE City
Department Stores



The Scientific Program:

- "While I was only able to attend the conference Friday and Saturday, I derived a great deal of information from that 24 hour period as well as being able to talk to patients/parents to help answer their questions. The session on Therapeutics and Clinical Trials was very interesting in terms of the different approaches used by Drs. Haas, Haller, Stacpoole, and Tarnopolsky to provide therapy for mitochondrial disease."
- G. Vladutiu, PhD
- "Once again, the combined conference format of basic and clinical science sessions overlapping with the parents meeting proved very successful. I was able to catch up with some patients and old friends as well as participate in a very exciting science program. The quality of the abstracts and the didactic science presentations was first class."
-R. Haas, MB, BChir
- "The meeting was amazing this year. There was an inspiring group of young scientists, including locals David Kelley and Clayton Matthews. Some of the research is starting to be aimed at therapeutics/gene therapy, which was very promising for the parents and families. I learned new things about my patients, especially about complex III and symptomatology. I gained new information about which supplements I should be using and I had the opportunity to ask questions of the experts."
-A. Goldstein, MD
- "I just wanted to thank UMDF and all involved in the conference. The scientific session was very helpful. As usual, I very much enjoyed interacting with the families. The patient/family interactions are clearly the most important aspect of the conference."
-J. Shoffner, MD



"Thank you for creating the only environment that provided me with understanding and acceptance."

-Barbara Bruck, OH



What Did You Like?

(Anonymous Responses from Evaluations)

- "Seeing people we had only met online. Having time to chat with others. The program syllabus that included slides and room for notes."
- "Dr. Shoffner's session on the Mito Basics was great. Meeting others from our state/region with mito kids -- this was one of the best aspects of the weekend."
- "The opportunity to hear and interact with the specialists in the field is always a high point."
- "I enjoyed the sessions on fundraising - they will be very useful."
- "Learning more about the UMDF and the stories and struggles of other families."
- "My passion for a cure for mito and to bring awareness to others was elevated to a whole new level. I have a desire to do what I can to reach my community. I am looking forward to becoming more involved in the UMDF."
- "Thank you for holding this conference. My husband and I have not had a weekend without the children for five years. It has been good for our marriage and what we have learned will help our son."

Special Thanks to our Foundation Sponsors and Exhibitors and In-Kind Donors

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2004 UMDF Grant Recipients

The United Mitochondrial Disease Foundation has made a commitment to raise \$5,250,000 by 2006. We are pleased to introduce our newest grant recipients. We hope to highlight our grant recipients in future issues of Mitochondrial News to provide more details on these exciting new projects.

David Chan, MD, PhD, *California Institute of Technology, Pasadena, CA* **Award Amount: \$128,000**

Project Title: Understanding the role of mitochondrial fusion in mitochondrial myopathies

Summary: This project will produce lab mice that have undergone tissue-specific deactivation of factors needed for mitochondrial fusion and will study the effects that this has on skeletal and cardiac myopathies.

Miriam Meisler, PhD, *University of Michigan*

Award Amount: \$108,305

Project Title: The nuclear-encoded gene OMI and Mitochondrial Disease

Summary: Their experience with a mouse model possessing a mutation that causes abnormal mitochondrial function leading to neuromuscular disease has led them to design a study in which they will screen 300 mitochondrial disease patients for the same mutation.

Volkmar Weissig, PhD, Sc, *Northeastern University, Boston, MA*

Award Amount: \$99,360

Project Title: Development of a method for transforming mitochondria in living mammalian cells with exogenous DNA

Summary: They plan to develop and perfect a mechanism for transporting exogenous plasmid DNA through the interior of cells to the mitochondria, with subsequent mitochondrial uptake of the DNA.



Mitochondrial Medicine 2004

UMDF Members Present Checks to Researchers

Top Row: V. Mootha, R. Marti, V. Weissig, D. Chan, J. Garcia, and R. Merrill (for S., Strack).

Bottom Row: Pam Johnson, of Kansas City Chapter; The Morgan Family, of Pittsburgh; Bob Thomas, of Indiana Chapter; Tova & Christopher Sido of Texas; Jennifer Lyman, of Ohio Chapter; and Stan Davis, UMDF Trustee.

Vamsi Mootha, MD, *Massachusetts Institute of Technology*

Award Amount: \$90,200

Project Title: Genomic Approaches to Human Cytochrome c Oxidase Deficiency

Summary: They will use a comprehensive computational strategy to identify genes that code for assembly factors responsible for cytochrome c oxidase; candidate sequences will be validated through the use of RNA interference in cultured cells with subsequent biochemical assay of potential changes in respiratory function.

Joseph Garcia, MD, PhD, *University of Texas SW Medical Center*

Award Amount: \$88,852

Project Title: The hypoxia sensing transcription factor EPAS1/HIF-2a is a novel mitochondrial disease candidate in mice and man

Summary: They will screen cell lines that have been collected from 200 pediatric mitochondrial disease patients for the presence of a mutated protein that normally stimulates the production of anti-oxidant enzymes and will express the coding errors in cultured cells to examine the specific dysfunction associated with the protein.

Stefan Strack, PhD, *University of Iowa, Iowa City*

Award Amount: \$88,000

Project Title: Protein phosphatase 2A in mitochondrial function and disease

Summary: Methods will be developed to block the activity of a mitochondrial regulatory protein that normally renders brain cells vulnerable to toxin-induced degeneration, with the goal of rescuing the cells from the effects of the toxins.

2004 UMDF Grant Recipients



Chuck Mohan and Tova & Christopher Sido presenting Dr. Garcia with a grant check during Friday night's banquet.

Brian Robinson, PhD, *Hospital for Sick Children, Toronto, Canada* **Award Amount: \$44,000**

Project Title: Drug development for the regulation of respiratory chain components in mitochondria

Summary: They will screen a large number of chemicals from a family of heterocyclic compounds that show promise for stimulating the production of mitochondrial respiratory proteins.

Gregory Enns, MB, ChB, *Stanford University, CA* **Award Amount: \$34,179**

Project Title: GSH levels, reactive oxygen species production, lipid peroxidation, products and mitochondrial membrane potential in patients with mitochondrial disease

Summary: They will look for correlations between the relative health status of mitochondrial disease patients and various factors associated with oxidative stress in the patients' cells.

Ramon Marti, PhD, *Fundacio Institut Hospital, Barcelona, Spain* **Award Amount: \$33,776**

Project Title: Restoration of thymidine phosphorylase activity in MNGIE patients through platelets infusion

Summary: Patients with the rare mitochondrial disease MNGIE will be infused with platelets that contain the enzyme they are missing, with the goal of decreasing plasma levels of toxic nucleosides that usually accumulate and damage the patients' mitochondria.



Jack Black Receives 2004 UMDF Humanitarian Award

During the Friday night banquet, UMDF Trustee, Sharon Shaw, accepted the Humanitarian Award on behalf of actor/musician, Jack Black. Chuck Mohan presented the award to thank Jack for his ongoing efforts in raising funds for research and bringing the term mitochondrial disease to the general public through various mediums.

Jack Black, well known for his roles in movies such as *School of Rock* and *Shallow Hall*, played *Who Wants to be a Millionaire* and *Celebrity Poker* to benefit UMDF. Since 2001, more than \$200,000 has been donated to UMDF thanks to Jack's efforts. Through various events and venues, mitochondrial disease and UMDF has been brought to the media's attention. Awareness is PRICELESS and we cannot thank Jack enough for helping us Redefine HOPE for thousands affected by mitochondrial disease.



UMDF Board Strives for Excellence through NEW Strategic Plan

Five focus groups were held during the 2004 conference and included doctors, UMDF members, staff, chapter/support leaders and the board. Feedback from the sessions was extremely positive and we look forward to seeing a strategic plan that will capture the vision of all our supporters and keep us focused on the UMDF mission. Please note that the mission has been revised:

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

Mito Adults Corner

by Laureta Fitzgerald, aka Laurie on the Mitoldies List

Many adults with mitochondrial disease (mito) have more to deal with than the health issues of mito. Most have had to give up a job, give up college, or feel inadequate as a parent. We all have to grieve for that loss. Many deal with depression, withdrawal from a social life and feelings of frustration.

After grieving the loss of what our life used to be or what we had envisioned, then what?

Finding a purpose is the next important thing we can do for ourselves. Without a purpose, it is hard to get past the grief and depression. The purposes we find may have to be adjusted from day to day or year to year. There are many things that we can do that we may not think of as a purpose in life, but finding something to make us feel worthwhile, puts a spark back into our lives. It gives us a reason to go on and be a happy person.

So, what can we find as our purpose? It really depends on the level of disease. If you are still able to get out and about, find a nursing home that needs someone to come in and talk with lonely residents. Find a hospital that needs someone to work as a volunteer. Working as a big sister or big brother to a child needing this can be very rewarding. Volunteer at the local library. The people who are in charge of these facilities need to know that you will come when you can. This might mean being a part of a team who trades off the duties if you are unable to go on a certain day.

If you are unable to get around the community or you don't have transportation, there are things you can also do. If you are into crafts, you can make decorations for a nursing home or hospital. You can knit or crochet little hats for the newborn nursery in your local hospital. You can give support to others an on-line group such as "mitoldies". You don't have to

have knowledge to contribute, but you can always send a "cyberhug" or let someone know you are thinking of him or her. You can find a senior who is living alone who needs someone to call them each day to make sure they are okay.

These ideas will not pay the bills, but we all need a reason to be alive - a purpose. It may take some trials to see what fits you the best, but that is okay. Helping others gives us a purpose, but finding something to challenge yourself can also give you a reason to feel good about yourself. Make sure your goals are realistic and you start out slow. The reason is to be of value as a person, not to set the world on fire. You just need to feel you have a purpose.



Mitoldies held a Silent Auction during the Mitochondrial Medicine 2004 Conference in Pittsburgh and raised \$437.50 to continue the UMDF mission! These ladies are hard workers and made some beautiful items for the auction.

Thank you so much! Hope to see you all in 2005 in St. Louis!

Mito Adults Corner

Calling all Mito Adults: The Mitochondrial News Needs YOU!

UMDF could use more helpers. Please consider joining the committee, submitting an article for review or send us your experiences with a specific topic of interest. If you are willing to help, please email Kara Strittmatter at kara@umdf.org or call 412-793-8077, ext. 106. We look forward to hearing from you!



Networking Requests

During a transition period in upgrading our computer system, we are currently unable to process networking requests. We hope to have a new and improved system operating in the next quarter. Your patience is appreciated. If you have any questions, please contact the UMDF office.

Morphometric analysis showed that mitochondria are significantly reduced in AD. These results indicate that vulnerable neurons in AD have increased mitochondrial degradation products, suggesting either greater turnover of mitochondria by autophagy (removal of mitochondria by being digested within secondary lysosomes) or a reduction of proteolytic turnover (removal of damaged mitochondria from cytoplasm to avoid cell damage) leading to accumulation of mtDNA and mitochondrial protein.

We also observed an overall reduction in microtubules (structures involved in the transport of organelles including mitochondria) in AD compared to control subjects (Cash et al., 2003). Altogether, these data indicated that the abnormal mitochondrial turnover (as indicated by increased cytosolic mtDNA and mitochondrial protein accumulation in the face of reduced numbers of mitochondria) could be due to a defective microtubule metabolism resulting in deficient mitochondrial transport. This may, in turn, set up a pathological cascade of events in the cytosol of neurons. These abnormalities occur in neurons lacking neurofibrillary tangles, placing mitochondria abnormalities as the earliest cytopathological change in AD.

In summary, mitochondrial defects are now described in a wide spectrum of human conditions including degenerative and metabolic diseases, aging, and cancer. The evidence that supports a critical role of mitochondria in neurodegenerative diseases and AD, in particular, is compelling. Studies from our laboratory have further elucidated the role of mitochondria in AD by showing that neurons in AD brains accumulate mitochondria debris in their cell body, which results from oxidative damage to mtDNA and mitochondrial proteins and may be related a priori to an impaired mitochondrial transport. Further studies examining the importance of mitochondrial pathophysiology in aging and AD may provide insight into neurodegenerative

disease pathogenesis and may indeed provide a target for specific therapies.

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An Inspiration

Lauren Surprenant and her friend Christopher Clark, of Belchertown, MA, designed and developed a remote control, power wheelchair for their 8th grade invention convention. They got an A+ and were on TV. Rides were given for \$1 and proceeds will go to UMDF. Very ingenious Lauren and Christopher - keep up the great grades and spreading the word about mitochondrial disease and UMDF!

Fundraisers

Allison Rogers contacted UMDF in the Spring of 2004 and asked if Chuck Mohan would speak to Eastside High School students in Taylors, SC and encourage them to choose UMDF as their charity for the 2004 Spirit Week.

Six months later, Spirit Week started on September 11 and raised \$62,204 to benefit UMDF. Spirit week is a tradition that began as a way to channel the destructive rivalry between two schools, Eastside High and Wade Hampton, and change it to a positive and successful fundraising competition.

Allison and Todd Rogers, parents of 3-year-old Carson, kept the UMDF informed of all the activities that occurred during the week. The family attended a spaghetti dinner

Spirit Week Competition Raises \$62,204 to Benefit UMDF

one night followed by a bachelor auction. High school students were auctioned off and the organizers included Carson. The girls went crazy when Carson walked on stage in his little tuxedo, blowing kisses and waving. UMDF members, the Peek Family, of Atlanta, and the Newton Family, of Clemson, also attended the dinner and auction.

40 fundraisers were held within this one week time frame -- from spaghetti dinners and yard sales to car washes and pageants. Thank you Eastside High School for choosing UMDF as your charity and helping us Redefine HOPE for thousands of kids like Carson.



Pictured above - Carson in his tux, Waynette and Riley Peek and Allison and Carson with decorated van, and Jenny Spork, Student Council President, with Carson.

Keep the "Coins for a Cure" Campaigns Coming, Kids!!!



Schools have been in session for at least a month - time to get those Coins for a Cure campaigns started in your local school!!! See pages 4 & 5 for chapter coin campaigns!



- ▲ In honor of Charles Darche of Chicago, IL, George F. Cassell Elementary raised \$500.
- In honor of Todd McCartha of Rome, NY, Bellamy School raised \$360.
- Grandmother, Melissa Cook, initiated a Coins campaign at her grandson's school in Ohio. Blanchester Local School District raised \$313.41 in honor of Dakota Mitchell.
- The Unified School District No. 434 in Overbrook, KS, raised \$53.65 in honor of Shelby Sund.

Energy for Life

In the Spring of 2004, UMDF sent members information about the *Coins for a Cure* and the *Energy for Life Campaigns*. The *Energy for Life* campaign targets businesses who are willing to sell printed cutouts of a light bulb, with the *Energy for Life* slogan, to their customers for \$1. Special thanks to the following for initiating a campaign in their communities:

- In honor of Harrison Montague - \$115.30 was raised at BP Connect; Paulding Gymnastics; and Stylers Salon in Georgia.
- Martin Family - In honor of Madelaine and Nathaniel Martin, parents Doug and Tammy of Parchment, MI have been busy. A *Coins for a Cure* was held at Schoolcraft High School and raised \$2168.09. An *Energy for Life* campaign at Mattawan Hardware and Big Joe's Pizza & Deli raised \$129.27.

- In Memory of Kevin and Eric Withum - Sir William Hair Fashions in Penn Hills, PA raised \$125 with the *Energy for Life* light bulbs.



Ring a Bell and Raise \$2,346 to benefit UMDF

On Friday, August 13th, Charlie Darche rang the opening bell to start the day's trading at the Chicago Stock Exchange. Charlie's aunt, Colleen Zimmerman, works at the exchange and helped organize the fundraiser to benefit UMDF. Special thanks to Charlie and all his family and friends at The Chicago Stock Exchange, Inc.

Fundraisers

Activities in Western PA Continue to Grow

Styx Vocalist and Keyboardist Decorates a Kite for Kristen!

The 4th Annual "Kites for Kristen" was another success raising more than \$13,000 to benefit UMDF.

Pat Charleston, Kristen's mother, was thrilled to have Dennis DeYoung and his wife, Suzanne, decorate a kite this year for the cause. The DeYoungs also donated autographed CDs to raffle off during the festivities! His kindness touched her heart and put a smile on Kristen's face. Thanks Dennis and Suzanne for helping us Redefine HOPE for thousands of children like Kristen!

Thank you, Charleston Family for your hard work and dedication toward finding a cure!!



▲ The **2nd Annual Pittsburgh UMDF 5K Run/Walk** raised **more than \$33,000** and attracted hundreds of participants in the rain! Next year - we'll have sunshine!!! Pictured above is Austin Manz, one of the top pledge collectors, with his mom, Jeanne.

▼ Caddies are lined up to be auctioned during the 1st Annual **Pittsburgh You Go Girl Golf Outing**. Organized by the LaCava, Mohan and Strittmatter Families, this first time event raised more than **\$4,900** and the ladies (and men) had a blast!



▲ Guess how many golf balls are in the golf ball ice sculpture and win a \$500 watch! Just one of the activities that helped UMDF raise more than **\$25,000** during the **7th Annual UMDF Pittsburgh Golf Outing**.

▼ The **Obley Family** held their first **"Rounds for Research" Golf Outing** in September to benefit UMDF and to honor their little girl, Kendall. The outing raised **more than \$5,000** and this event was thrown together in just three weeks! Outstanding!!!



▲ The **4th Annual Ohio Golf Outing** raised **more than \$78,000** in July and the above volunteers were key to the success. From left to right, Amy Rogoff, Sandy Babbush, Mara Bernon, Chuck Babbush and Jill Platt. Carol and Stan Davis, Jill's parents, dedicate countless hours to this annual event in honor of their grandchildren!



Chuck Mohan entertains the Deasy kids, of the Western PA Support Group, during the 4th Annual Cruisin' Toward a Cure Car Cruise which has raised more than \$11,000. Thank you, Ron and Donna Miklos, and all your cruiser friends, for their continued dedication to UMDF!



Help UMDF Identify Everyday Heroes

UMDF 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.

UMDF 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.

Nomination Forms and more instructions are available on the UMDF Web Site at www.umdf.org. Or request forms by calling Jill at 412-793-8077 or emailing JillG@umdf.org.



Pictured left to right: Kevin Shelly, winner of the LEAP award, Chuck Mohan and Samantha Stahler, winner of the Heartstrings award.

Congratulations to our 2004 LEAP and Heartstrings Winners!

Kevin Shelly Receives LEAP Award

21-year-old Kevin Shelly, of Harleysville, PA has not let his diagnosis of MERRF/MELAS slow him down. Kevin has been involved for years volunteering in his church outreach program and is an active member of the Knights of Columbus. As a talented singer/actor, he has performed in several community theaters, choral groups and a barbershop quartet group -- plus, he teaches piano to children. He is also taking classes to earn a degree in music therapy.

It is Kevin's magic that has touched so many. He is an inspiring magician and performs at many Delaware Valley Chapter functions and offers his service for raffle prizes. Kevin demonstrates that by having a positive attitude, working hard and believing in yourself, so much can be accomplished. A great lesson for us all!!!

Samantha Stahler Receives Heartstrings Award

As a loving cousin, 13-year-old Samantha Stahler has been dedicated to raising funds for research and has raised nearly \$3,000. From asking for donations in lieu of her 10th birthday to lemonade stands and selling friendship bracelets to baking holiday cookies to sell, this young lady never stops working toward a cure for her cousin Bobby Arnold. Her biggest accomplishment was getting her handmade bookmarks into several boutiques. Each year, Sam spends countless hours preparing the raffle for the Ohio race and collects pledges too! She tugged the heartstrings of her cousin's parents, Lisa and Bob Arnold, and she tugged at ours too! Keep up the great work, Sam!

UMDF Chapters & Groups

ARIZONA

★Arizona Chapter

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CALIFORNIA

★Southern California Chapter

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MICHIGAN

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New group forming in Western Michigan.

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Fundraisers



Two-year-old Laynee Brunk was the official starter for the **Ally Brunk Memorial 5K Run/Walk**. Julie, Dan, Laynee and Carson, and all their friends, raised **\$3,900** to benefit UMDF and send their prayers to all the families who are affected by mitochondrial disease.

Special Thanks to the following fundraisers:

- Mohan Family Fourth of July celebration raised \$158.
 - The family of Brayden Suman raised \$1,875.
 - The Billy Hackett Open raised \$2,825 in memory of Kayla Elizabeth Naughton.
 - Angelica/UMDF Family Fun Day raised more than \$1,300 to date.
 - In honor of Dominique and Kevin Ingram, the Mohegan Sun Casino Bus Trip event raised \$447.
 - A lemonade stand for Tyler Young raised \$6.00 and Sara Strittmatter raised \$10.05 for the UMDF with her Kool-aid stand. Keep up the great work, kids!!!
- Cans for Ashley sent in their recent collection of \$150 to benefit UMDF. Keep it up, Randy and Sandi, and Ashley!
 - Candy Machines in Mohan's, Ribkens and LaCava's Italian Market restaurants have raised \$500 in recent months.

THANK YOU!!!

The Annual Gibson Barbecue/In Memory of Heidi Marie raised \$6,025 this summer. Love those Gibsons and Daniels!!

★ **Mark your Calendars** ★

See pages 4 and 5 for Upcoming Chapter Events

- October 3, 2004 - Fifth Annual Mito-What? 5K Walk/Run, Longmeadow, MA
- October 9, 2004 - Carson's Crop: A Scrapbooking Event, Taylors, SC
- October 9, 2004 - 5K Walk/One Step Closer to a Cure, Birmingham, AL (rescheduled from Sept 18)
- October 16, 2004 - 2nd Annual Dinner & Wine Party for Charlie's Fund, Dallas, TX
- November 15, 2004 - 1st Annual Fore-a-Cure Golf Tournament, Stone Mountain, GA

UMDF Annual Membership Meeting Notice

8085 Saltsburg Road, Suite
Pittsburgh, PA 15239

Saturday, December 4, 2004



UNITED MITOCHONDRIAL DISEASE FOUNDATION®

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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 next issue is 1/3/05