Mitochondrial Disease: A Practical Approach for Primary Care Physicians - Part II

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PATHOGENESIS OF MITOCHONDRIAL DISEASE

Primary Mitochondrial Disease

The term “primary mitochondrial disease” refers specifically to mitochondrial dysfunction caused by genetic mutations, directly impacting the composition and function of the electron transport chain. These defects impair mitochondrial oxidative phosphorylation (OXPHOS), the process by which oxidation of the end products of metabolism in the electron transport chain is coupled to phosphorylation of adenosine diphosphate to produce energy in the form of adenosine triphosphate. These disorders are unique in that the electron transport chain is the only metabolic pathway under dual control of both the mtDNA and nDNA genomes. Therefore, the transmission of mitochondrial disease can occur by traditional mendelian genetics or by mitochondrial genetics, the latter of which is complicated by special considerations such as heteroplasmy, threshold effect, mitotic segregation, and maternal inheritance.5

nDNA-Based Primary Mitochondrial Diseases

Mutations in nuclear genes are increasingly becoming recognized as the major cause of pediatric mitochondrial disease.26 This occurrence is explained by the predominance of proteins expressed in the mitochondria that are synthesized by nDNA (~850 genes) compared with mtDNA (13 genes).26 Autosomal recessive inheritance of nuclear genetic defects is probably the most common etiology of mitochondrial disorders in children, although mild manifestations are occasionally observed in heterozygous carriers.10

Nuclear genes implicated to date in mitochondrial disease encode proteins that are structural subunits of mitochondrial enzyme complexes, cofactors, assembly factors, translation factors, mtDNA maintenance factors, and factors that are important for the fission and fusion of this dynamic organelle. However, the specific causative gene defect has yet to be identified in most patients with probable mitochondrial disease that is suspected to be nuclear in origin. For example, the genetic basis remains a mystery in 50% of patients with complex I dysfunction, which is the largest protein complex in the 5-complex electron-transport chain and the one most commonly implicated in mitochondrial disease. The nDNA diseases that cause severe coenzyme Q10 deficiency deserve special consideration as presenting a rare treatment opportunity in mitochondrial disease, because their symptoms, the onset of which may range from infancy to adulthood, usually respond to coenzyme Q10 supplementation.31 A full discussion of the clinical findings seen in individual nDNA defects (continued on page 2)
is beyond the scope of this article but has been addressed in several excellent reviews.\textsuperscript{32}

\textbf{mtDNA-Based Primary Mitochondrial Diseases}

Human mtDNA is a small 16 569-base pair molecule that encodes 37 genes. Primary mtDNA abnormalities consist of point mutations, deletions, or duplications. Point mutations are maternally inherited and may affect genes for mitochondrial transfer RNA, mRNA, ribosomal RNA, the control region, or the 13 mtDNA genes that encode electron-transport chain subunits. Deletions and duplications in mtDNA are usually sporadic. mtDNA disorders are clinically heterogeneous, but some phenotypes such as Leigh disease and MELAS are particularly common.\textsuperscript{5,33} Depletion of the number of copies of mtDNA in a tissue can occur, although the cause for this depletion is commonly a mutation in an nDNA gene.

Interestingly, with age, the genetic basis for mitochondrial disease is more likely to be found in mtDNA than in nDNA.\textsuperscript{4,34} Common primary mitochondrial diseases in older patients include mtDNA-deletion diseases (eg, chronic progressive ophthalmoplegia or Kearns-Sayre syndrome) and mtDNA point mutations in transfer RNA genes (including MELAS and Leber hereditary optic neuropathy).

\textbf{Secondary Mitochondrial Disease}

Even when mitochondrial dysfunction is confirmed by sophisticated biochemical testing, it can be challenging to distinguish whether the cause for this dysfunction is a gene that directly impacts the electron-transport chain or is secondary to an unrelated genetic or environmental cause. Thus, definitive diagnosis of mitochondrial disease cannot be based on biochemical findings alone, because in vitro electron transport chain enzyme activities in a patient's tissue sample may be reduced secondary to other metabolic diseases or to specimen-handling issues.

Mitochondrial dysfunction, which may or may not be clinically relevant, may be seen when the primary defect occurs in another energy-related metabolic pathway, such as fatty acid oxidation\textsuperscript{35} or amino acid metabolism.\textsuperscript{36} In addition, OXPHOS impairment with reduction of in vitro electron transport chain enzyme activity by as much as 50% has been seen in tissue samples from patients with other metabolic diseases. Indeed, other diagnoses that have ultimately been confirmed in individuals with suspected mitochondrial disease and biochemical evidence of in vitro mitochondrial dysfunction include copper-metabolism disorders (Wilson disease and Menkes disease\textsuperscript{37,38}), lysosomal disorders (neuronal ceroid-lipofuscinoses\textsuperscript{39} and Fabry disease\textsuperscript{40}), peroxisomal disorders,\textsuperscript{41,42} pantothenate kinase–associated neurodegeneration, holocarboxylase synthetase deficiency, molybdogen cofactor deficiency, and neonatal hemochromatosis.\textsuperscript{43}

It is increasingly recognized that OXPHOS impairment may be contributing to the disease pathology in some genetic conditions not typically classified as mitochondrial or metabolic disorders, including Rett syndrome,\textsuperscript{44} Aicardi-Goutie’res syndrome,\textsuperscript{45} various neuromuscular disorders,\textsuperscript{46} and Duchenne muscular dystrophy.\textsuperscript{47} In addition, activities of electron-transport complexes in skeletal muscle may be decreased in malnourished children, with correction to normal levels after improved nutrition.\textsuperscript{48}

Medications and toxins can also significantly affect mitochondrial function. Sodium valproate may impair mitochondrial function by the induction of carnitine deficiency, depression of intramitochondrial fatty acid oxidation, and/or inhibition of OXPHOS\textsuperscript{49–51}—this knowledge should prompt consideration of alternative anticonvulsant use in mitochondrial disease, particularly in patients with mitochondrial polymerase \_mutations.\textsuperscript{52}

Other important examples of drugs that may induce mitochondrial dysfunction include antiretroviral nucleoside analogues for HIV,\textsuperscript{52,54} as well as salicylates, which impair liver mitochondria in Reye syndrome.\textsuperscript{55}

Because many clinical features that may raise suspicion for mitochondrial diseases are nonspecific, the differential diagnosis can be very broad. The clinical presentation of mitochondrial disease in children can mimic other multisystem disorders such as congenital disorders of glycosylation or Marinesco-Sjogren syndrome\textsuperscript{56,57} or even be misinterpreted as a vascular or immunologic stroke syndrome. Although clinical and neuroimaging features of Leigh syndrome are generally strongly suggestive of a mitochondrial disorder, there are other conditions that may give rise to striatal necrosis that should be considered. Similarly, clinical and neuroimaging findings may sometimes suggest other leukoencephalopathies or neurodegenerative disorders.\textsuperscript{18}

\textbf{MITOCHONDRIAL NEWS}

\textbf{FROM PAGE ONE}

\textbf{DIAGNOSTIC EVALUATION OF MITOCHONDRIAL DISEASE}

The major challenge to properly establishing mitochondrial dysfunction as the cause of a patient's presentation is the absence of a definitive biomarker that characterizes mitochondrial disease in all patients. Thus, the diagnostic evaluation is necessarily multitiered and broadbased, with a focus on integrating information from many avenues: the complete medical and family history, clinical findings that may be suggestive of mitochondrial disease, biochemical laboratory abnormalities such as lactic acidosis (which, as discussed above, is neither sensitive nor specific as a single biomarker for many mitochondrial disorders), tissue-biopsy evidence of abnormal electron-transport chain enzyme activity or impaired respiratory capacity, and, if possible, the identification of a pathogenic mtDNA or nDNA mutation. This process often involves sophisticated assays that require invasive procedures such as muscle or liver biopsy to obtain tissue for testing in specialized laboratories. These investigations may give intermediate or ambiguous results, and decreased activities of electron transport chain enzymes may be secondary to nonrespiratory chain disorders.\textsuperscript{42} To aid interpretation, 2 diagnostic schemes for infants and children have been proposed to categorize the likelihood of mitochondrial disease in a given patient as definite, probable, possible, or unlikely.\textsuperscript{58,59} Guidelines for diagnosis and treatment of mitochondrial disorders in infants and children were proposed recently by a European working group and are available in English online (http://aps-med.de and select “leitlinien”). However, these complex and sophisticated diagnostic algorithms are directed at the metabolic specialist and limited in their clinical utility for the generalist who is contemplating initiation of the diagnostic evaluation for a particular patient.

The diagnostic evaluation typically proceeds from general clinical evaluation to imaging and metabolic screening tests and then to more specific biochemical and genetic assays. This process starts with less invasive assays and proceeds to more invasive biopsy-based analyses as required. Clearly, the complete diagnostic process can become complicated, and enlisting the early involvement of a local metabolic specialist may be quite helpful.

Referral to a metabolic specialist

(continued on page 8)
Living with mitochondrial disease presents many twists and turns and a maze of questions. UMDF is pleased to offer answers to some of those questions as taken from Ask the Mito Doc™ at UMDF.org. 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.

Responder for this issue: Russell P. Saneto, DO, PhD, of Children’s Hospital & Regional Medical Center, Seattle, WA, Mary Kay Koenig, MD, of the University of Texas Health Science Center at Houston, TX and Salvatore DiMauro, MD, of Columbia University, New York, NY.

THE QUESTION IS...
Our daughter is now six years old and is diagnosed with a possible mitochondrial encephalopathy complex IV. This has never been confirmed, but a very thorough workup has been done. We have seen more tremors within this past year. They used to be after a severe regression with a viral illness and would resolve as she came “back up” to baseline but now they are there most of the time and do not leave. In recent months she has started having intention tremors. This is concerning for us. She had an MRI a year ago and it did not show anything. She is on CoQ10 and carnitine. Would there be any reason to see the neurologist? Enhancing the carbohydrate level of the diet might help a little as well. I have not found the usual medications such as beta blockers or myosin to be very helpful, but you might ask your physician about these drugs.

There are some practical points. With intention tremors, when using writing or coloring instruments, use the big fat ones. The glass should be half full and not completely full. At school, see if it is okay to use a helper (such as a computer) for class projects and long writing assignments. When doing a lot of hand work, then a weighted glove can be helpful (although should not be used continuously). Velcro is a heaven sent item and can be used in many situations where the tremor becomes an issue.

RESPONSE FROM RUSSELL P. SANETO, DO, PHD:
I am sorry to hear about your daughter. I know it is so difficult to watch you daughter have all these problems. Tremors are difficult to control, especially when they are due to mitochondrial disorders. It is very common to see neurological deficits during illness as this is a strong stressor to our systems and energy demands go up (fighting infection, keeping the body going, etc) the poor mitochondria are faced with added energy demands that in the face of a mitochondrial disease, compromises their function even to a greater extent. This is why many patients have their first symptoms during a viral illness. Over time as the mitochondrial disorder progresses, these same symptoms are enhanced and may, as seemingly with your daughter, become continuous. You are doing the correct things, at least in my opinion with the coenzyme Q10 and carnitine. CoEnzyme Q10 or Carnitine.

THE QUESTION IS...
I’m a 50 year old male with diagnosed MERRF #8344 (8249 RFLP) and LHON #13708 through blood tests. My questions are: Can Mitochondrial problems cause a deficiency in Immunoglobulin IgM (low level)? Can Mito cause Folliculitis?

Also, lately I have been extremely tired, total mental/physical fatigue with no energy. However, other times, I am full of energy and extremely hyper. Usually these episodes happen every other day—from fatigue to hyperness—full of energy. I noticed that if I’m on my feet for 8-10 hours one day, then, the following day is where extreme fatigue sets in. Could this be a cause from Mito? I have other problems not mentioned here, but this fatigue is the worst and most worrisome. Please note: I am aware that other than a Mito problem, there are other ailments that could be causing these symptoms, but am wondering in the absence of this, could Mito also be playing a role.

RESPONSE FROM MARY KAY KOENIG, MD:
Mitochondria are present in all organs and therefore can potentially affect all organ systems. Although not typically associated with mitochondrial disorders, a deficiency of immune function can be seen. The immunodeficiency could then pre-dispose to infections, such as folliculitis.

Most patients with mitochondrial disorders experience episodic fatigue. It is common for patients to become fatigued after activity and fatigue can set in many hours after the activity has occurred. Many patients report improvement in their fatigue after beginning supplements such as CoEnzyme Q10 or Carnitine.

RESPONSE FROM SALVATORE DIMAURO, MD:
Your concern is certainly understandable, given the recent publicity about a potential relationship between mitochondrial disease, autism, and vaccination. We are dealing with three “dots” here: two of them (mitochondrial dysfunction and autism) have definitely been connected: although probably only a minority of autistic children have an underlying mitochondrial disease, some (like, apparently, your son) certainly do. The third “dot” (vaccination) however has NOT been connected to either mitochondrial diseases or to autism. I doubt very much that vaccination triggered the autistic features in your child, which can be simply explained by his mitochondrial dysfunction. children with mitochondrial diseases should be vaccinated to protect them from horrible infectious diseases!
CHAPTER ACTIVITIES

ATLANTA CHAPTER

- June 7, 2008. A Movie Night for Mito and Family Fun Fest was held at Joint Venture Park in Cumming, GA in memory of Samantha Mancuso. The event featured a special screening of the Chipmunks movie and had activities for the whole family! Thank you to Nadine Mancuso for organizing such a meaningful event!

CHICAGO CHAPTER

- May 10-17, 2008. The eighth annual Kites for Kristen was held in honor of Kristen Charleston. A special thanks to the students and staff of St. Daniel the Prophet School! Events included a kite decorating contest, dress down day activities, silent auction and raffles. Proceeds benefit the UMDF through the Kristen Charleston Research Fund.

DELAWARE VALLEY CHAPTER

- April 29, 2008. A Jamming for Jamie Talent Show was held at Shawnee High School in Medford, NJ in honor of Jamieson Smith. Jamie is a student at the high school. The event was sponsored by the faculty and staff of Shawnee High School and the Junior Executive Homeroom. The event raised $3,400 for the UMDF.

INDIANA CHAPTER

- April 5, 2008. A Hockey Night was held at an Indiana Ice hockey game at Conseco Fieldhouse in Indianapolis, IN. The Indiana Ice donated five dollars from each ticket purchased on behalf of the UMDF to the Foundation. Thank you to the Indiana Ice organization for your continued support!

- June 6, 2008. The second annual Green Gala 80’s revival event was held at the Allison Mansion on Marion College Campus. The event was held in honor of Riley Henderson.

NEW ENGLAND CHAPTER

- May 18, 2008. A Bike-E-Thon was held at Haines Park in Barrington, RI. The bikers rode along the East Bay Bike Path and a family picnic was held for the bikers after their ride. The event raising $1,500 was held in memory of Elijah Bonney. Thank you to the Bonney Family for organizing a great event!

OHIO CHAPTER

- April 11, 2008. The Kobunski fifth annual Guest Bartender Night was held at the 82nd Street Grill and Pub in North Royalton, OH. The event raised $5,025 for the UMDF in honor of Kyle Kobunski. Thank you to the Kobunski Family for another great event this year!

- April 20, 2008. Joey’s second annual Spaghetti Dinner was held at the Mogadore Moose Lodge in Mogadore, OH. Dinner included homemade spaghetti, salad, rolls and a bake sale table with homemade desserts. The event was held in honor of Joey Ricci. A special thanks to the Ricci Family!

- May 10, 2008. The seventh annual Run Wild for a Cure 5K Run 1 Mile walk was held again this year at the Cleveland Metroparks Zoo. More than 3,000 participants joined the Cleveland Ohio Chapter in raising over $200,000 for the UMDF! The Ohio Chapter has now surpassed the $1 MILLION dollar mark for the race! Thank you to the committee members for your hard work on another successful year!

SOUTHERN CALIFORNIA CHAPTER

- May 6, 2008. The Chevy’s Restaurant in Mission Valley, CA held a UMDF night and donated 25% of all food sales made from 5-8 pm to the UMDF. Thank you to Kappa Alpha Theta’s alumnae board for your support of this event!

- May 17, 2008. Motors for Mito, a motorcycle ride, was held in Norco, CA. The ride was hosted by officers from the Department of Corrections facility in Norco. The event included a motorcycle ride, family fun activities and a BBQ lunch after the ride. A special thanks to Steve Nolan, owner of Backwoods BBQ, for hosting the party! The post-ride events included live music, balloons, clowns, a magician and a puppet show. The ride and after party were held in honor of Taryn Fogel.

- May 17, 2008. On National Scrapbooking Day, Stacey Calk organized the activity for the Southern California Chapter. The event was held at Central California Children’s Hospital and raised $700 for the UMDF. A special thanks to Stacey for organizing the event in honor of Joshua and Nolan Calk.
GRAND ROUNDS HELD IN CINCINNATI - By: Jeff Salt

The Cincinnati Mito Group was pleased to welcome Dr. Bruce Cohen from the Cleveland Clinic and Chuck Mohan, CEO of the United Mitochondrial Disease Foundation to Cincinnati Children's Hospital Medical Center (CCHMC) on February 19th for a very successful Grand Rounds. The Sabin Center auditorium was packed with staff doctors, pediatricians and nurses and many more were tied in through the hospital’s web conferencing program from the many satellite locations of the hospital. Jeff and Cindy Salt, Cincinnati Mito group leaders, worked very hard to notify the different departments of the hospital by e-mailing the flyer for the meeting to every major department and to all the therapists. They also faxed the flyer to every pediatric office in the greater Cincinnati area as well as to school districts and the county boards of MRDD.

Chuck Mohan and Dr. Cohen also made themselves available for a very successful and well attended family night the evening before. Over 80 family members along with a few medical staff that would not be able to make the Tuesday morning medical session packed the Sabin Center auditorium for Dr. Cohen and Chuck’s presentations. After the presentations, Dr. Cohen spent quite a long time answering questions for the families that attended. There were families that had driven in from as far away as Buffalo, NY, Charleston, WV and South Bend, IN to hear Dr. Cohen speak. Their children are treated at CCHMC.

The Cincinnati Mito Group would like to thank Dr. Cohen and Chuck Mohan for giving of their time to come to Cincinnati for these wonderful and informative events. They both did an outstanding job! We would also like to thank our sponsor, Transgenomic, for funding the Grand Rounds program. We have already noticed an increased awareness of mitochondrial disease by several of the departments at CCHMC.

The following Grand Rounds are scheduled for 2008:
- July 8 and 9, 2008 in Chicago, IL
- August 11, 2008 in Philadelphia, PA
- September 17, 2008 in Ann Arbor, MI
- September 18 and 19, 2008 in St. Louis, MO

For more information on the Grand Rounds program, contact Janet at 888-317-UMDF, email janet@umdf.org or visit www.umdf.org

OTHER NOTABLE EVENTS

- **April 19, 2008.** The Emmyfest 2008 Charity Benefit Concert featuring MOSSSA, Crucial Elements, William Walter and Tucker Rogers was held at the Capital Ale House in Richmond, VA. The event raised $3,300 for the UMDF in honor of Emily Jurek and all affected by mitochondrial disease.

- **April 19, 2008.** The St. Louis Mito Group hosted the fifth annual Mito-What? Family Fun Day at the Shrine of Our Lady of the Snows in Belleville, IL. This year the walk raised more than $14,000 for the UMDF! A special thank you to the race committee for all of your hard work!

- **May 4, 2008.** Stephanie Watkins and friends participated in the Frederick Running Festival in Frederick, MD. Team Noah raised over $4,175 for the UMDF in honor of her son Noah. Congratulations, Team Noah! We knew you could do it!

- **May 9, 2008.** Over 350 students from Fordwood Elementary School in Wilmington, DE walked for their buddy Preston Buenaga in conjunction with Preston’s March. The pouring rain did not hamper the fun as the children walked in the gym, made banners and were treated to ice cream and colorful pencils. The walk raised $1,200 toward Preston’s March. Bringing the total to over $45,000 for the UMDF. Thank you to the Buenaga and Frostick Families!

- **May 18, 2008.** Christopher Walker participated in the Ford Ironman Triathlon at Walt Disney World in Florida. Christopher, who is currently a pre-med student at Duke University, has taken on this mission to give hope to his cousin Rachel and to all who have been diagnosed with a mitochondrial disease. To date, 70.3 Miles for Rachel has raised more than $4,600 for the UMDF.

- **May 18, 2008.** The Corvette Club of Western Pennsylvania hosted their annual Corvette Cruise at Day Chevrolet in Monroeville, PA. A portion of the proceeds raised benefitted the UMDF. Thank you to Joe Haver and the Corvette Cruisers of Western PA for your support!

- **May 25, 2008.** The third annual Golf Tournament in honor of Brittany Wilkinson was held at the Madera Golf Course in Clovis, CA. The tournament was a four-man golf scramble, and fun was had by all. Proceeds from the event benefit the UMDF through the Brittany Wilkinson Research Fund. Thank you to the friends and family of the Wilkinson’s for your continued support!

- **June 31, 2008.** The Dobke Family of Waukesha, WI hosted the second annual Greater Mito Open Golf Classic at Old Highlander Golf Course in Delafield, WI. The outing was held in memory of Zachary Juhlmann who recently lost his battle to mitochondrial disease and is now in heaven with his best brother Sam. A special thank you to the Dobke Family for your continued support!

- **June 7, 2008.** The sixth annual Pittsburgh One Step Closer to a Cure 5K Run 1 Mile Walk was held at North Park Boathouse in Allison Park, PA. A special thank you to all the families who came out to support their teams!

- **June 21, 2008.** Myles for a Cure was held on the Cedar Valley Nature Trail in Cedar Falls, IA. The walk was held in honor of Myles Eick. Thank you to some of the local supporters including Mix 96.1, Covenant AirCare Helicopter, Sartori Ambulance and Cedar Falls Fire Department.

- **June 28, 2008.** The third annual Save Barnegat Bay 8K and Fitness walk was held in Lavallette, NJ. A portion of the proceeds of this event were donated to the UMDF in honor of the Boyle Family. The 8K race was followed by an awards ceremony and a post-race party sponsored by the Crab’s Claw Inn.
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For more information about a chapter, group or ambassador near you, contact the UMDF office at 888-317-UMDF!
The relationship that Joey and his family have with the Alabaster Firefighters is something truly special. The Paulin family has made well over 25 calls to 911 in the past 4 years. “It is their quick response and knowledge of Joey's illness that plays a part in why our little guy is still with us today,” Roseanna says. Not only do the men of the fire department respond when there is an emergency, they are also there for the family in other ways. They make visits to the hospital and to the Paulin’s home to check on Joey, and even attended his 4th birthday party. “These men have become like family to us,” Roseanna adds.

Roseanna received a phone call from Lt. Ched Burton last month. Lt. Burton informed her that the City of Alabaster was going to name a day after Joey. “I was speechless,” Roseanna said. He then went on to tell her that Joey was also going to be made an Honorary Fireman with the Alabaster Fire Department. “That is when the tears began,” she said. “They were happy tears, though.”

The date had been set. On May 5, 2008, a special reception would be held in Joey’s honor at City Hall. That is the day that Joey, the sweet little dark haired, dark eyed boy, would become a star! The Paulins’ day started off at 4:30 a.m. with an interview at Fox News. The interview had just ended when Roseanna’s phone rang; this time it was NBC News; they too wanted an interview with Alabaster’s Youngest Firefighter. However, Joey had a very important appointment with UAB’s Department of Genetics at 8:30 a.m., so she told NBC News that they would call the minute they were out of the appointment. At 10:00 a.m., Roseanna called NBC only to find out they were already on their way to the fire station. The Paulins made it back to Alabaster and to the interview just in time for the mid-day news.

The rest of the afternoon was filled with calls from local newspapers who wanted interviews.

At 6:30 p.m., Roseanna, Larry, Gionna and Joey arrived at City Hall only to find a “packed house” of friends and family. Among them was Joey’s Youngest Firefighter

A Day in the Life of the Paulin Family

It started with a phone call placed just after 2 a.m. on June 12, 2004. The phone call was made by Roseanna and Larry Paulin to 911 emergency services. Their two month old son, Joey, was experiencing his first seizure; his lips were blue and his eyes had rolled into the back of his head. They had no way of knowing that the responders to that call, the Alabaster Fire and Rescue Service, would become more than just “Joey’s Heroes”, but would become like part of the family.

Joey was born on April 12, 2004. At just 12 hours old, Joey’s eyes began to roll back into his head and he stopped breathing numerous times. After seven days in the NICU, Joey was released to go home. The Paulins were told that Joey had an immature nervous system and that he would be fine in a couple of months. Then on June 12th, the unthinkable happened, Joey had his first seizure. He was rushed to Children’s Hospital where he had another seizure the next day. Joey spent one week in the ICU before he was sent home with a diagnosis of epilepsy.

The next five months of Joey’s life were filled with numerous hospital stays. It was when Joey stopped eating that the doctors decided to perform a muscle biopsy to rule out a mitochondrial defect. In November of 2004, Joey received a diagnosis of Mitochondrial Encephalomyopathy, Complex I Deficiency. The doctors told the Paulins that Joey would not be able to walk, talk, sit or stand, and gave a life expectancy of 4 to 7 years.

Joey is now four years old. He has had surgery to have a g-tube placed in his stomach and has an IV port. He continues to have seizures which have increased in frequency over the past 18 months and have become life threatening. The worst was in September of 2007 when Joey had a seven hour seizure. The doctors did not expect Joey to make it through the night. Joey fought hard and 18 hours later he woke with a smile on his face.

MITO FACTS

• The majority of childhood onset cases are inherited, although there are cases in which the disorder seems to be a random occurrence.

• Because mitochondrial disease is under recognized, there is no way of knowing how many adults in the United States have a mitochondrial disease.

• Patients affected with a mitochondrial disease present with a wide range of symptoms such as a weak heart, seizures, failing kidneys and/or respiratory complications.
The UMDF recently wrapped up our annual symposium “Indy 2008 – Setting the Pace for Mitochondrial Medicine”. This event took place in Indianapolis, Indiana and drew hundreds of families, medical experts, and researchers. Affected individuals or those who came with an affected family member were able to network with mitochondrial disease medical experts and researchers. The medical community was able to listen to information about the very latest mitochondrial research. I want to thank everyone that attended, and I certainly enjoyed visiting with many of you. I will write more about the symposium in our fall issue, but I wanted to talk with you about a couple of memorable items that came up in Indianapolis.

New questions about mitochondrial dysfunctions have been coming up in the press recently. Two examples of this are the side effects of statins, a type of cholesterol drug, and their possible impact on the mitochondria and autism. These stories are continuing to raise the public’s awareness of our issues. We are working on communication strategies that will help us tell our story in a manner that more people can understand. This is critical as we strive for a larger allocation of government research dollars to be made for mitochondrial disease. Our next symposium is scheduled to be held in June 2009 in Washington D.C., to help accomplish this task. Mark your calendars – we will need your help.

I am proud to tell you that through your tireless work and fundraising efforts for the UMDF, our Research Grants Program was able to award more than $1 million to 10 research projects for 2008 that we believe will ultimately help us find a cure for mitochondrial disease. Some of the proposals were quite unique. We all need to celebrate this achievement, because over the past 11 years, the UMDF has provided $6 million in research grants aimed at our mission to find a cure. It is because of your work that the UMDF is the largest, non-governmental contributor of grants focused on mitochondrial disease research. In addition, I am aware of two small pharmaceutical companies that are in the early stages of developing drugs that are targeting the mitochondria. While these drugs are not available yet, I am certain in the next several years we will have new treatment options for our diseases.

Also, I want to tell you about a group of high school students who attended the symposium. Nine young ladies from Sacred Heart Academy in Hamden, Connecticut successfully submitted an abstract for review at the symposium. The abstract was titled “Sequencing the Limulus mitochondrial COX1 gene: Comparison of SNPs to those causing human disease.” According to their submission, the students’ test subject was a horseshoe crab from Long Island Sound (LIS). The study of its COX1 SNPs may reveal important ecological data. Since an American woman has a one in seven risk of developing breast cancer, the students are researching COX1, LIS Limulus, comparing its polymorphisms to those previously identified from inhabitants of the Delaware/Chesapeake Bay, and making a final comparison to cytochrome oxidase mutations implicated in human cancers, specifically breast cancer.

It is very unusual for those who review abstracts to allow a high school project presentation. However, all agreed that we need to encourage young people in their research and help them consider the field of mitochondrial disease research. It was very exciting to have the girls - Francesca A. Carlisle, Meghan C. Crosby, Nicole A. DeLauro, Kelley E. Durham, Alessandra Fiallos, Elena E. Jackson, Erin M. Laskowski, Jennifer A. Limauro and Andrea L. Wagner present their abstract. We would also like to thank Sr. Mary Jane Paolella for her help in guiding the girls in this project and getting them to Indianapolis. Well Done, Ladies!

Energy to all,

W. Dan Wright, UMDF Chairman

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PARTNERS IN PROGRESS - Vol. 13, Issue 2

FROM THE CHAIRMAN

The UMDF recently appointed three new members to the Board. Please help us welcome the new trustees.

John DiCecco is no stranger to the UMDF Board of Trustees. Mr. DiCecco served on the board from 1998-2007. During his time on the board, he held the position of Treasurer and Vice Chairman. Mr. DiCecco was nominated to the board in January of 2008 and will continue the hard work and dedication as he has for the past 10 years. Welcome back Mr. DiCecco!

Rob Platt joined the UMDF Board of Trustees in February of 2008. Although new to the board, he has been a dedicated friend of the UMDF. Mr. Platt’s daughter Carly, 13, was diagnosed in 1999 with a mitochondrial disease. The Platt Family has been involved with many fundraising events for the UMDF. Mr. Platt is the owner and president of Branding Solutions, Inc., an advertising specialty and apparel company located in Cleveland, Ohio. Welcome to the board Mr. Platt!

Dr. Howard Zucker joined the UMDF Board of Trustees in March of 2008. Dr. Zucker brings with him an extensive background in pediatric medicine and has worked for the U.S. Department of Health and Human Services, the National Institutes of Health and the World Health Organization. Dr. Zucker’s experience is an asset to the UMDF and we are all excited to have him with us. Welcome to the board Dr. Zucker!
SUPPORTING OUR CAUSE

MEET THE UMDF SPECIAL EVENTS DEVELOPMENT TEAM

The UMDF Development staff has recently expanded to deal with the increasing need to assist with events occurring nationwide. Our volunteers host so many events across the country and we wanted to provide the assistance necessary to ensure continued successes. Meet our special events team:

Jodie Tabano - Special Events Manager
If you have ever been involved with organizing an event around mitochondrial disease awareness, you most likely know Jodie Tabano. Jodie joined the UMDF staff as the manager of special events in March 2005 and has helped to organize numerous special events across the nation from your walk/runs to golf outings, to virtual tea parties, to galas, to silent auctions, to some very creative fundraising ideas. Jodie is energetic and efficient, with 15 years of development, special events and fundraising experience working for the Muscular Dystrophy Association, La Roche College and the American Lung Association. Jodie hails from Ohio, receiving her BA from Ohio Northern University and her MS from La Roche College. Our development staff is lucky to have Jodie as a member of our team. Jodie coordinates all national event activities, and we hope you have a chance to work with her to organize an event.

Jennifer Belliveau - Special Events Assistant
Jennifer is a 2006 graduate of the University of Notre Dame with a BA in English and a minor in Theology. Prior to joining the staff at UMDF, she did a year of volunteer service with the AmeriCorps VISTA program at the Volunteer Center of Madison County in Huntsville, AL. She joined the UMDF in February 2008. Jennifer is focusing on building our presence and awareness locally in the Pittsburgh community, where our national headquarters is based.

Ryan Fisher - Development Assistant
Ryan joined the UMDF in June of 2008 after managing the public relations campaigns of various corporate and non-profit clients. Prior to that, he spent six years on Capitol Hill as the Legislative Director to a member of the U.S. House of Representatives. Ryan has a degree in political science from Miami University in Ohio.

Donald Gielas - Grant Writer
Don brings a wealth of experience to the UMDF after working as a writer and development professional at various Pittsburgh-based institutions. Don has worked in development at the University of Pittsburgh and most recently at Duquesne University, serving the last twelve years as Director of Corporate and Foundation Relations.

Carol Rush, PhD, Director of Development, is also a new addition to the UMDF Development Department and is looking forward to attending her first Brew at the Zoo in Pittsburgh on August 9th. Carol hopes to attend an event or two around the country this year to visit and meet with you.

Please don’t hesitate to contact the Special Events Team to help you get started with hosting a fundraiser or for help in increasing the success of an event that you will be hosting again this year. Let us know how we can help you make your next event a smashing success!

CONTACT THE DEVELOPMENT TEAM
888-317-UMDF (8633)
events@umdf.org

DID YOU KNOW THAT, AS A MEMBER OF THE UMDF, YOU CAN RECEIVE FREE FINANCIAL ADVICE?

The UMDF has a relationship with The Monteverde Group, LTD., wherein members of the UMDF may contact Monteverde for free financial planning advice. Monteverde can assist you in not only planning legacy gifts and bequests to the UMDF, but also personal financial planning as well. For more information contact the UMDF Development Department or Nick Nicholson at The Monteverde Group, LTD. directly:

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Andy Garrison, 27 years old with Leigh’s Disease
**2008 UMDF RESEARCH GRANT RECIPIENTS**

- **$150,000** Brendan James Battersby, PhD, University of Helsinki. Project title: “Identifying genetic modifiers of tissue-specific mitochondrial DNA segregation.”

- **$128,563** Stuart Smith, PhD, DSc, Children’s Hospital & Research Institute at Oakland. Project title: “Utilization of knockout mouse models to elucidate the importance of the de novo mitochondrial fatty acid synthesis pathway in mitochondrial function.”

- **$125,000** Leo Joseph Pallanck, PhD, University of Washington. Project title: “The role of the PINK1/Parkin pathway in mitochondrial integrity.”

- **$116,428** Bridget Elizabeth Bax, PhD, St. George’s University of London. Project title: “Evaluation of the efficacy and safety of erythrocyte encapsulated thymidine phosphorylase therapy in two patients with mitochondrial neurogastrointestinal encephalomyopathy.”


- **$100,000** Deepa Vinay Dabir, PhD, University of California-Los Angeles. Project title: “Study of redox regulated pathways in the mitochondrion.”

- **$99,998** Timothy E. Shutt, PhD, Yale University School of Medicine. Project title: “Selective alteration of mitochondrial gene expression via modulation of the dual-function h-mtTFB1 and B2 factors as a potential therapy for mitochondrial diseases.”

- **$99,998** Sion L. Williams, PhD, University of Miami. Project title: “Evaluation of novel zinc finger nucleases as a means to target m.3243 A>G in vivo.”

- **$99,990** Rebeca Acin-Perez, PhD, Weill Medical College, Cornell University. Project title: “OXPHOS modulation by mitochondrial protein phosphorylation in mtDNA mutant cells.”

- **$98,300** Elizabeth Anne Amiott, PhD, University of Utah. Project title: “Mitochondrial Fusion Defects in Neurological Disease.”

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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.
ATLANTA CHAPTER
- 2008. Riverside Design Group, Inc’s Plates with Purpose created seven customized plates to benefit non-profit partners. The UMDF is among one of the partners and will receive 15% from its customized plate sales. The UMDF plate design is a lady bug, in loving memory of Morgan Reynolds, who loved lady bugs very much. Plates can be purchased at www.plateswithpurpose.com. For more information contact info@riversidedesigns.com.

- September 22, 2008. The third annual Kovalcik & Geraghty Wealth Partners Charity Golf Outing will be held at Worthington Country Club in Columbus, OH. Registration and lunch begin at 11:00 a.m. with a shotgun start at noon. For more information, contact Lorrie Stickel at lorrie.stickel@raymondjames.com. Register or donate online at www.umdf.org/KGWP.

- October 5, 2008. The eighth annual Olivia Steele Memorial Golf Outing will be held at Kyber Run Golf Course in Johnstown, OH. The outing is brought to you by Performance Site Co., Shelly Material and the Steele Family. Golf sponsorships are available. Individual greens fees are $70 per person or $280 per team. For more information, contact JR Steele at 614-206-6237 or email jr.steele@performancesitecompany.com.

CHICAGO CHAPTER
- September 14, 2008. The second annual UMDF Chicago Area Chapter 5K Walk and Family Fun Day will be held along scenic Lake Michigan at Lincoln Park in Chicago, IL. For more information, contact Vicki at vternberg@niag.com, call 847-441-8261 or go to www.umdf.org/chicagowalk.

- Summer, 2008. New Image Cut-a-thons will be held at the Lake County Captains Games in East Lake, OH again this baseball season! Barbers Paul and Frank will be giving haircuts for a donation to the UMDF to those who attend games on: July 25th and August 22nd. This event is held in honor of Jonathan Kucaric.

- August 1, 2008. The Kraig family in Ashtabula, OH is holding a benefit drawing for two ready to build property lots with utilities in Roming Shores (corner of Pierpont Road and Sugar Creek Drive). The tickets are $25 each or two for $45. A portion of the proceeds will benefit the UMDF. For more information, contact Jeff Kraig at 440-222-7450 or email jkraig@yahoo.com. Winner is responsible for property taxes and association fees.

CLEVELAND CHAPTER
- Summer, 2008. “Masterpieces” is available for purchase at her CD titled “Louann Carnahan, Piano Virtuoso.” To create awareness about “mito” and raise funds for research, her CD titled “Louann Carnahan, Piano Masterpieces” is available for purchase at www.beatmito.com. For more information contact Louann at lcarnahan@kcumb.edu.

- August 1, 2008. The Kraig family in Ashtabula, OH is holding a benefit drawing for two ready to build property lots with utilities in Roming Shores (corner of Pierpont Road and Sugar Creek Drive). The tickets are $25 each or two for $45. A portion of the proceeds will benefit the UMDF. For more information, contact Jeff Kraig at 440-567-1267 or email jeffkraig@yahoo.com. Winner is responsible for property taxes and association fees.

DELWARE VALLEY CHAPTER
- July 26, 2008. The fifth annual Brew at the Zoo will be held this year at the Philadelphia Zoo in Philadelphia, PA. This event will feature more than 20 national, local and home brewers in the unique setting of the Philadelphia Zoo. For more information go to www.umdf.org/batzphiladelphia or contact Jennifer Belliveau at 888-317-UMDF or events@umdf.org.

- September 13, 2008. The ninth annual Go for Mito! 5K Run 1 Mile Walk will be held at the Philadelphia Museum of Art in Philadelphia, PA. If you are interested in volunteering or serving on the race committee, contact Therese at nivrag53@aol.com or call 215-281-9228. Register or donate online at www.umdf.org/goformito.

- September 27, 2008. Jamming with Jamie and Friends will be held in conjunction with the New Jersey Mitochondrial Disease Awareness Week events. If you are interested in volunteering or helping with the event committee, contact Laurel at dalsmith@aol.com or call 609-714-0091. To purchase tickets or donate online, go to www.umdf.org/jammingwithjamie.

- October 4, 2008. An Outdoor Family Fun Fest event will be held at the Cannstatter Zoo in Philadelphia, PA. For more information, contact Tinna Donnelly at 267-240-2393 or email tinna211@comcast.net.
While we would like to list ALL of our donors individually, please understand that it would be very difficult to do so with 5,567 names that donated $1-$99 (THANK YOU!), 1,420 names that donated $100-$249 (THANK YOU!) and 303 names that donated $250-$499 (THANK YOU!). Please know that we have not forgotten and we deeply appreciate each and every individual, organization and corporation that has generously donated money to support our mission. We know who you are, and we thank you! 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 July 1, 2007, and Dec. 31, 2007. If your name or company’s name is incorrect or not listed, please contact info@umdf.org.

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- Grantham, Mayo, Van Otterloo House of Blues
- Kelley Management Consulting

Individuals:
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- Sharon & Christopher Schleif
- Robert Sheridan
- Harry & Roslyn Weinrauch

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- March of Dimes
- Sage Foundation
- Stratham Vanderbilt Womens Club

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Equivalent Pharmaceutical Industries Corp.
Giant Eagle Market District
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Michael & Laura Weinrauch
Richard Weisberg
Gregory Winfield
Scott Winston
Susan & Fred Winther
Kurt & Angela Woelffer
John Yahres
Brian Yamaato
Julie Yannes
Tina Zadrozny
Daniel & Joyce Zallanta
GREATER JACKSON MISSISSIPPI MITO GROUP

• September 20, 2008. Jude’s Fais DO DO for Mito will be held at the Jubilee Center in Sulphur, LA starting at 6:30 p.m. Activities include dinner, dancing, silent auction and raffles. Tickets are $25 in advance or $30 at the door. We are honored to have Mary Kay Koening, MD as our honorary chair for this event. She will be on hand to meet our families and friends in the community. For more information or to purchase a ticket online go to www.judgesmitojourney.com.

WESTERN PA MITO GROUP

• August 9, 2008. The second annual Pittsburgh Brew at the Zoo will be held at the Pittsburgh Zoo & PPG Aquarium in Pittsburgh, PA. This fun fundraiser will feature more than 20 national, local and home brewers in the unique setting of the Pittsburgh Zoo. Along with great beer, the event will also have food, live music, door prizes and more. For more information go to www.umdf.org/batzpittsburgh, contact Jennifer Belliveau at 888-317-UMDF or email events@umdf.org.

• September 13, 2008. The second annual Sip and Savor the Moment for a Cure will be held at the LaCasa Narcisi Winery in Gibsonia, PA. The event is held in honor of Hannah Pallas. The evening will begin at 7:00 p.m and will include wine tasting, chocolate fountain, chocolate confections, appetizers, live entertainment, a silent auction and basket raffles. For more information or to order tickets go to www.umdf.org/sipandsavor.

AROUND THE COUNTRY

• October 11, 2008. The second annual Kure for Kat will be held at South Lakes Park in Denton, TX. If you are interested in volunteering or serving on the race committee contact Maggie Dickens at kureforkat@yahoo.com or call 940-382-2434. Register or donate online at www.umdf.org/kureforkat. Kure for Kat is held in honor of Katherine Dickens and proceeds benefit the UMDF through the Katherine Dickens UMDF Research Fund.

(continued from page P1)

neurologist, some of his nurses and of course, the men of the Alabaster Fire and Rescue Service. “They were in their dress blues,” Roseanna said. “They looked very handsome.”

Alabaster’s Mayor, David Frings, presented Joey and the Paulin Family with a proclamation declaring April 12 as “Joey Paulin Day” in the City of Alabaster and also made him an official honorary firefighter with the City of Alabaster. The fire department also presented Joey with an authentic fireman’s hat and badge. This honor has made Joey Paulin Alabama’s Youngest Firefighter!

Joey getting a fireman’s haircut from FF Josh!
• Megan Cochran of Georgia celebrated her second birthday this year! The Cochran Family sent invitations to Megan’s Party and made it a letter writing campaign for the UMDF. They raised an outstanding $1,804.11 in honor of young Megan! Happy Birthday Megan!

• Shiloh Elementary School hosted a Pennies for Jenny coins drive in honor of Jenny Schnitzler. The students raised $2,222 for the UMDF! Way to go!

• Akerman, Link & Sartory held a dress down day in honor of Michael Link and the Southern Florida Mito Group! They raised $200 for the UMDF.

• Derek and Sara Swanson of Arizona recently celebrated their fifth birthday! In honor of their birthday, $200 was donated to the UMDF. Happy Birthday Derek and Sara!

• The employees of the John Deere Waterloo Operations in Waterloo, IA participated in the seventh annual Festival of Trees. Employees purchased and then decorated a tree. The winners were chosen by votes, in the form of donations, for their favorite tree. Fifty-six trees were decorated and donations were shared with various charities. Over $370 was donated to the UMDF in honor of Myles Eick.

• The 2007-2008 Waukesha Lady Wolverine Junior Varsity Basketball team held a “Free Throw for Friends” fundraiser as part of their community outreach program. Players collected donations for the number of free throws they made during the conference scheduled portion of the season. The ladies made 122 free throws in 14 games and raised $1,379 for the UMDF in memory of Zachary Juhlmann. Great job, ladies!

• The Custer Family of Bluffton, SC participated in a local event called “MayFest”. They had a UMDF table with special HOPE.ENERGY.LIFE bracelets that they sold along with ice cold water. The table raised almost $500 for the UMDF in honor of Olivia Custer.

• The sixth annual Hefferon Post-Holiday Gift Recycling Auction was held in McLean, VA in honor of David Hefferon. The event raised $6,309.50 for the UMDF!

• In lieu of company Christmas gifts to their employees, Georgia EMC made a donation of $1,000 to the UMDF. The generous gift was made in memory of Jake Lyons and Morgan Reynolds.

• Isabel Taylor organized a bake sale at Ralya Elementary School in East Lansing, MI in honor of her brother Simon. With some help from her friends, Isabel raised over $529 for the UMDF! Way to go, girls!

• Judy Dockert of Milbank, SD retired from Otter Tail Power Company this year. In lieu of a formal retirement observance, a donation of $430 was sent to the UMDF in her name.

• Westley Clapp of Boise, ID celebrated his third birthday this year! To help celebrate, the family asked that in lieu of toys for his birthday that a donation be made to the UMDF. Over $300 was donated in honor of Westley. Happy Birthday, Westley!

• Some special friends of Lilly Grossman had a birthday party in Boston, MA and in lieu of gifts, they asked that donations be made in honor of Lilly. The girls raised $280 for the UMDF. A special thank you to these young girls for their thoughtfulness!

• The Anderson Family in memory of Andrew & in honor of Peter Lulebzyk

A special thank you to all of these families and to the families who are still participating in Coins for a CureSM and Tea for MitoSM! For information about how you can become involved, contact Jennifer at 888-317-UMDF or jennifer@umdf.org.
should always be made when symptoms and signs strongly suggest mitochondrial disease, patients appear potentially unstable with classic metabolic disease features, lactic acidosis is present in blood or CSF, a maternal inheritance pattern is observed, or abnormalities are identified through baseline diagnostic evaluation. Referral by a primary care physician is also prudent when more elaborate testing is needed, such as provocative testing or muscle biopsy with investigation of electrontransport chain enzymes. If a biochemical diagnosis has been made but the molecular basis remains unknown, additional genetic testing and counseling should be coordinated by a specialist.

Mitochondrial disease is clearly not a single entity but, rather, a heterogeneous disorder of energy dysfunction caused by hundreds of different nuclear and mitochondrial gene mutations, and other defects. Thus, there exists no currently accepted gene-based diagnostic algo-rithm that is useful for all patients or pursued by all metabolic specialists. Testing for nDNA mutations can be performed on any tissue, including blood. However, most diagnostic nDNA gene testing should not be performed a priori but, rather, guided by the clinical picture, tissue-specific signs, and biochemical findings in a given patient. In contrast, testing for mtDNA mutations is frequently most informative when performed on a muscle biopsy specimen, although urinary sediment and buccal cells may also be useful specimens. It is important to recognize that dietary advice should always be given in a specialized setting. In addition, although there are only a few available treatment options for mitochondrial disease, they are best offered by clinicians with experience in these disorders.

ROLE OF THE PRIMARY PRACTITIONER IN THE DIAGNOSTIC PROCESS

The relative scarcity of metabolism specialists worldwide underscores the value of aving the primary physician assist, when possible, in the preliminary stages of the diagnostic evaluation by initiating appropriate baseline diagnostic testing. This cadre of testing is particularly useful to perform for children with “vague” presentations when the primary practitioner may be uncertain if there is sufficient evidence to warrant a metabolic referral. Similarly, normal results from baseline testing may lessen concern that a mitochondrial diagnosis is being missed. Of course, if symptoms or signs persist, worsen, or remain unexplained, consultation by a metabolic specialist may still be indicated.

CONCLUSIONS

The unique nature of the symbiotic and semiautonomous physiology of mitochondrial biology gives rise to a wide range of human mitochondrial disease. What were once regarded as a few rare diseases to be described at grand rounds or as case reports in journals are now commonly recognized disorders that are seen daily in a broad array of patient clinics. It is hoped that greater familiarity among primary care physicians with the protean but real manifestations of mitochondrial disease will facilitate the proper diagnosis and management of this growing cohort of diseases that present across all specialties.

REFERENCES

40. Lucke T, Hoppner W, Schmidt E, Illinger S, Das AM. Fabry disease: reduced activities of respiratory chain enzymes with decreased levels of energy-rich phosphates in fibroblasts. Mol Genet Metab. 2004;82:93–97
Dr. Melissa Nixon, DVM, of Nevada City, CA, was recently honored at the Tower of Hope’s Lighting the Path Gala in Manhattan, NY. Melissa was presented with the 2008 “Beacon of Light Award,” acknowledging her commitment to being an active volunteer in veterinary disaster relief for the past 20 years. She is a caregiver to her husband, Ken, who is a disabled veteran and is a partner to her beloved service dog, Thor.

Melissa has mitochondrial myopathy. She continued her practice of veterinary medicine for years with the aid of wheelchairs and crutches. In August of 2007, she decided to retire from her beloved profession. Just two months later, Thor qualified as a working service dog and immediately began his partnership with Melissa.

Melissa’s volunteer work with veterinary disaster relief has since been moved from the front line to her computer. She is a volunteer for Veterinary Information Network, sits on the CARES committee for the state of California and has worked for various disaster response entities. This coming June, Melissa will be participating live in a round table presentation at the National Conference on Animals in Disasters in Sacramento, CA.

Along with the UMDF, the Tower of Hope Foundation is near and dear to Melissa’s heart. Knowing how much she relies on Thor, she knows that the work the Tower of Hope does will help many others in need.

The Tower of Hope is a non-profit organization that strives to empower people who are living with a disability, a chronic illness, or have been in an abusive relationship to live happier and more independent lives through service and assistance animals.

“Think how much it would mean to be able to stand and walk, because of a dog’s assistance, instead of being wheelchair bound.” Melissa said in her acceptance speech, “In case you can’t picture that - I will tell you from personal experience - it means the world.”

Louann Carnahan is a 25-year-old medical student, an accomplished pianist and suffers from a mitochondrial disease called MERRF Syndrome (Myoclonic Epilepsy associated with Ragged Red Fibers). The disease causes Louann to have seizures and muscle weakness that get progressively worse over time.

At just 5 years old, Louann began to study classical piano and continued until she graduated from high school. When she was 11 years old, she started recording her songs and completed a classical music CD by the age of 17. Around the same time, Louann experienced her first grand-mal seizure and was diagnosed with Juvenile Myoclonic Epilepsy.

During college, Louann took a break from classical music and started playing the piano at her church. In her first year of medical school, she knew that classical music was a part of her soul and she began to play again. “It was an outlet for the intense rigors and pressures of the first two years of medical school.” Louann says of her classical music. “I learned these songs as I pushed through a seizure incident.”

In January of 2008, Louann was diagnosed with MERRF after seeing a muscle specialist because of her increasing muscle weakness. Louann passed her Step 1 Medical Board Exams in July of 2007, and on the advice of her neurologist decided to take off the 2007-2008 school year, her third year of medical school, to let her body rest.

While Louann is taking a break from school, she has not taken a break from her music. “Playing the piano seems to be an outlet to channel her emotions,” Louann’s mother Caroline says. “They flow through her fingers and into the keyboard with passion and skill.”

Louann has decided to “Beat MITO” and make a fundraiser out of her classical CD, entitled “Louann Carnahan, Piano Masterpieces”. All proceeds from the CD will benefit the UMDF to support research and education for mitochondrial disease. You can find out more information and hear a sampling of her music at www.beatmito.com. Information is also available at www.umdf.org in the calendar of events. Copies of the CD are now for sale, and order forms are available at both websites. Louann’s photo is featured on the front cover of this issue of Mitochondrial News!

**ONE WOMAN’S DECISION TO “BEAT MITO”**

**CALLING ALL MITO ADULTS!**

**Mitochondrial News Needs YOU!** Please consider submitting an article on your experiences with a specific topic that would be of interest to other mito adults. If you want to contribute, please contact UMDF at info@umdf.org or 888-317-UMDF. We look forward to hearing from you!
A CHAMPION IN THE UNITED STATES CONGRESS - BY RICK LEACH, UMDF TRUSTEE AND CHAIRMAN OF GOVERNMENTAL AFFAIRS COMMITTEE

A primary focus of our national advocacy effort is to build support among Members of the U.S. Congress for increased research into mitochondrial disease. We need to identify “champions” in the U.S. Congress who will play a leadership role in promoting our research agenda.

Representative Jim McDermott (D-WA) is one of our champions. He is currently serving his ninth term in the U.S. House of Representatives, representing Washington State’s 7th Congressional District, which includes Seattle and parts of neighboring communities. He is also a physician with a long-standing interest in health issues.

Representative McDermott is a champion for those affected by mitochondrial disease and their families. He has met with families impacted by mitochondrial disease in his congressional district and in Washington, DC. He was instrumental in helping us to determine how much money NIH had allocated to research into mitochondrial disease. This information is critical to building our case about the need for more research funding. He also led the effort in the U.S. House of Representatives to include language within the last appropriations bill that funds NIH to urge NIH to allocate more funds for research into mitochondrial disease. He has also expressed strong support for a mitochondrial disease Roadmap Initiative within NIH. A Roadmap Initiative would enhance collaboration among many of the Institutes at NIH in the area of mitochondrial disease research. Representative McDermott has also worked with families in the Seattle area and supported the efforts of Seattle Children’s Hospital.

Over the years of his public service, Representative McDermott has consistently been a leader on health issues. In the mid 1980s, well before HIV/AIDS was commonly understood, he was the first elected official to call attention to the need for better understanding of this disease and its potentially far-reaching impact. Similarly, he was the first Member of Congress to speak about the potential impact of stem cell research and to advocate for increased support. In the same visionary way that he has taken up these causes, Representative Jim McDermott has now stepped up to champion our cause.

The challenge to our chapters and members throughout the country is to engage other champions in the United States Congress that can work with our good friend Jim McDermott.

MITOCHONDRIAL DISEASE AWARENESS WEEK

With the help of some amazing volunteers, a Mitochondrial Disease Awareness Week has been established in the following states:

PERMANENT

ALABAMA
GEORGIA
MICHIGAN
NEW JERSEY
WISCONSIN

ONE TIME

MASSACHUSETTS
OHIO
PENNSYLVANIA
WASHINGTON
FRESNO, CALIFORNIA
CLOVIS CITY, CALIFORNIA

PENDING

LOUISIANNA
MISSISSIPPI
SOUTH CAROLINA
TENNESSEE

MITOCHONDRIAL DISEASE AWARENESS WEEK
SEPTEMBER 21-27, 2008

WE NEED YOUR HELP TO DESIGNATE A NATIONWIDE MITOCHONDRIAL DISEASE AWARENESS WEEK

WHAT CAN YOU DO?
Contact your local Senator or Representative today!
Start to plan an awareness activity during Awareness Week!

NOT SURE WHAT TO DO?
Log on to www.umdf.org to download the Awareness Week “Toolkit”, and learn how to get started with sample letters, a sample bill template and letter writing tips!

QUESTIONS?
Contact Cliff Gorski at 888-317-UMDF or email cliff@umdf.org
Tissue and Organ Donation for Research

The United Mitochondrial Disease Foundation occasionally receives requests for information regarding facilities that accept tissue and organ donations for research purposes. Some of our mitochondrial disease experts have provided us with the following brain and tissue bank sites:

National Disease Research Interchange
The NDRI is a non-profit organization that has been providing human tissues, cells, and organs to scientific research for over 25 years. They are currently in need of musculoskeletal system tissues, cardiovascular system tissues, integumentary system tissues, nervous system tissues and eye tissues. If you would like to learn more about NDRI, you can visit their website at www.ndriresource.org/donor_programs/tissue_banks/112/ or call 800-222-NDRI (6374).

Brain and Tissue Bank for Developmental Disorders (MD)
The Brain and Tissue Bank for Development Disorders in Boston, MA was established to advance the research of developmental disorders by providing human tissue to research. For more information, visit their website at http://medschool.umd.edu/btb/gintro.html, call 410-706-1755 or call toll-free 800-847-1539.

Brain and Tissue Bank for Developmental Disorders (FL)
The Brain and Tissue Bank for Development Disorders in Miami, FL collects samples from patients with a variety of genetic and developmental disorders, patients with related disorders, and age-matched controls. The material is used by investigators studying disease that affect normal brain development. For more information, visit their website at http://pathology.med.miami.edu/btb or call 800-59BRAIN.

Autism Tissue Program
The Autism Tissue Program is a brain tissue donation program. They do post-mortem home visits to establish autism diagnosis and have a number of projects that are on going. For more information, visit their website at www.memoriesofhope.org and www.atpportal.org or call 877-333-0999.

Solace Nutrition Offers Cyto-Q

Solace Nutrition recently announced the launch of Cyto-Q, the first and only liposomal encapsulated Coenzyme Q10 Medical Food for the dietary management of mitochondrial cytopathies. Solace Nutrition partnered with Tischon to develop Cyto-Q. Cyto-Q is to be used under medical supervision in the dietary management of mitochondrial disorders, where patients are not capable of producing adequate levels of Coenzyme Q10.

The ubiquinol form of Coenzyme Q10, which is used in Cyto-Q, dominates most human tissue. Mark De Fries, CEO of Solace Nutrition commented that “clinical studies demonstrated that ubiquinol is the most effective suppressant of free radicals and also increases mitochondrial energy output. While Coenzyme Q10 exists in both ubiquinol and ubiquinone forms, the ubiquinol form has been shown to be vastly more bio-available than conventional ubiquinone, commonly sold as an OTC supplement”.

Until now, patients could only purchase Coenzyme Q10 in the ubiquinone form, which is less bio-available. These products are classified as dietary supplements and purchased at retail stores. Unfortunately, patients were burdened with paying 100% of the cost of cost with no support from medical insurance.

The launch of Cyto-Q offers relief to patients by meeting their needs. Cyto-Q is available in easy to consume pineapple-orange flavored liquipaks. Each Cyto-Q liquipak guarantees to deliver 80 mg of the ubiquinol form of Coenzyme Q10. With the classification of a medical food, Cyto-Q could be covered by some insurance companies and government agencies.

For more information, please visit www.solacenutrition.com. Cyto-Q can be ordered online or by calling 888-876-5223. The price is $65.00 per box and each box contains 30 liquipaks.

Phase 3 Trial of Coenzyme Q10 in Mitochondrial Diseases

Eligibility: Children age 12 months to 17 years with biochemical or genetic proof of a mitochondrial disease. There are factors that could exclude some children with a mitochondrial disorder from participating. Ask the study coordinator if your child is eligible to participate.

Study Information: The study will help provide information to determine if Coenzyme Q10 (CoQ10) is a safe and effective treatment for children with mitochondrial diseases. There are no cures for mitochondrial diseases at this time. Treatments have sought to help reduce the symptoms or delay the progression of the disease. CoQ10 is a mitochondrial cofactor and antioxidant that is a part of the process by which food and oxygen are converted into energy by cells.

Your participation is important because: It will contribute to medical knowledge about mitochondrial diseases, help determine the benefit of CoQ10 in treating mitochondrial diseases, and help future clinical research trials look for specific treatments for people with mitochondrial deficiencies.

Potential Benefits: Developmental evaluation of your child, evaluation of his/her motor skills, objective evaluation of effectiveness of CoQ10.

Visits: Total of 5, one every 3 months (for about 1-2 days), at one of the participating centers.

Medication: CoQ10 every day for 6 months and placebo every day for 6 months. In addition, each child will be given a nutritional cocktail (vitamins) and vitamin daily. Your child will be allowed to continue taking all medications necessary for his/her health. You will be asked to keep a daily medication diary of the study medications during the course of the trial.

Tests: Medical history, physical exam, neuromuscular exam, six minute walk test (if able), blood and urine tests, speaking with a dietician about your child’s diet, completing developmental, sleep, behavior and quality of life questionnaires.

Participating Centers:
- General Clinical Research Center, University of Florida, Gainesville, FL
- William T. Dahms Clinical Research Unit, University Hospitals Case Medical Center, Cleveland, OH
- Cincinnati Children’s Hospital Medical Center, Cincinnati, OH
- Hospital for Sick Children, University of Toronto, Ontario, Canada

For more information contact: Tracie L. Kurtz, Senior RN Specialist Study Coordinator at: University of Florida
JH Miller Health Science Center, PO Box 100226, Gainesville, FL 32610
Email: Tracie.Kurtz@medicine.ufl.edu
**ANNOUNCEMENTS**

**UMDF MERCHANDISE**

UMDF merchandise is a great way to raise awareness and to support the UMDF and all those affected with mitochondrial diseases. The UMDF has recently secured a new merchandise fulfillment company to run the online Logoshop. Accurate Advertising, located in Pittsburgh, PA, has partnered with the UMDF to handle UMDF merchandise. The UMDF Logoshop is scheduled to open on July 1, 2008. Be sure to check www.umdf.org for more information!

**THE UMDF HOLIDAY AND FOUR SEASONS ART CONTEST WINNERS ANNOUNCED!**

The winner of the second annual UMDF Holiday Card Art Contest is 11-year-old Kelsee Pollak from Pittsburgh, PA! Kelsee’s work is in honor of her cousin, Hannah Pallas, and is titled “Next Stop…Cure!” because she believes we are always one step closer to a cure. Before becoming involved with the UMDF, Kelsee felt there was no help to find a cure. Now that she and her family are involved with the UMDF, they have hope that we will reach a cure. As the winner of the Holiday Card Art Contest, Kelsee was presented with a Chuck-E-Cheese party pack gift certificate.

The first UMDF Four Season Greeting Card contest winner is 16-year-old David Charleston of Chicago, IL. David’s artwork entitled “Time” is dedicated to his sister, Kristen, who was diagnosed with MELAS when she was five years old. Although Kristen can no longer participate in these activities, David holds his memories of their time together through the seasons close to his heart. David was presented with a Pizza Hut and Applebees gift certificate package.

Congratulations to both winners!! The winners were chosen by Jamie, an art student from The Art Institute of Pittsburgh. Thank you for your time!

This upcoming fall both greeting cards will be available for purchase to send to your family and friends during the holiday season. Proceeds from card purchases will help the UMDF work toward a cure and sending the greeting cards will help to generate awareness of mitochondrial diseases.

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**IN THIS ISSUE:**

- **Practical Approach for Primary Care Physicians - Part II**
  Read page 1!

- **Concerned about having your child vaccinated?**
  See page 3!

- **What’s new in my area?**
  Check out pages 4-7!

- **Mitochondrial Disease Awareness Week**
  Find out more on page 10!