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UNITED MITOCHONDRIAL DISEASE FOUNDATION
MITOCHONDRIAL NEWS

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

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The clinical recognition of mitochondrial disease is often a challenging endeavor. Genetically based, primary mitochondrial dysfunction presents as a heterogeneous group of disorders, which together are now recognized to constitute the most common neurometabolic disorder of childhood.[1] Epidemiologic studies of mitochondrial disease are limited by disease heterogeneity and underdiagnosis. Prevalence figures are less accurate than incidence figures in estimating mitochondrial disease frequency due to the high childhood mortality of these disorders. The preschool incidence was 1 in 11,000 live births in a Swedish study,[2] whereas the incidence of mitochondrial disease presenting by age 16 was 1 in 16,000 live births in an Australian study.[3] The Australian group combined adult prevalence figures with childhood incidence figures to arrive at an estimated minimum "birth prevalence" of 1 in 7,634 live births. Allowing for incomplete ascertainment, a lifetime risk of developing mitochondrial disease of 1 in 5,000 live births is a more probable estimate.[1, 4]

The most common presentation of childhood-onset mitochondrial disease is Leigh syndrome, which is a progressive neurodegenerative disorder that involves developmental regression, brainstem dysfunction, and lactic acidosis, although this classic presentation only accounts for an estimated 18% of all pediatric mitochondrial disease.[5]

Mitochondrial diseases are usually progressive and multisystemic. Typically affected organs are those with a high energy demand, including skeletal and cardiac muscle, endocrine organs, kidney, nonmucosal components of the intestinal tract, retina, and the central nervous system. However, virtually any organ or tissue can be involved. As a general rule, the involvement of 3 or more organ systems without a unifying diagnosis should raise suspicion for mitochondrial disease.

Although effective treatments remain elusive, definitive diagnosis is crucial for permitting appropriate symptom management, as well as accurate prognostic and recurrence-risk counseling. Diagnostic difficulty results not only from the wide spectrum of symptoms and signs that an individual patient may have but also from the absence of a reliable screening or diagnostic biomarker that is both sensitive and specific in all cases of mitochondrial disease. Although primary mitochondrial disease by definition has a genetic etiology, the genetic abnormality may be found in either mitochondrial DNA (mtDNA) or nuclear DNA (nDNA). More than 150 mtDNA athogenic point mutations and 100 mtDNA deletions have been identified in symptomatic patients.[6, 7] However, nDNA mutations account for the majority of mitochondrial disease that

(continued on page 2)

Curtis's Team pictured left at Christopher's Heart Fun Run held in Houston, TX. The event raised over $47,000!

See pages 4-7 for more events and fundraisers!

CONTENTS

Volume 13
Issue 1
Spring 2008

presents in infants and children.[8–10]

A simplified and standardized approach to facilitate the clinical recognition of mitochondrial disease by primary physicians is needed. With this article we aimed to assist the generalist in recognizing the most indicative features of mitochondrial disease, standardize the definitions of primary versus secondary mitochondrial disease, and provide a consensus approach to both empower the generalist to initiate appropriate baseline diagnostic testing and aid with the decision about referral to a specialist center for what, early on, may be a nonspecific presentation.

WHEN TO SUSPECT MITOCHONDRIAL DISEASE

Red Flags

Mitochondrial disease may present with "any symptom in any organ at any age,"[11] but some symptoms and signs truly are more suggestive of a mitochondrial disorder than others. These "red-flag" features warrant the initiation of a baseline diagnostic evaluation for mitochondrial disease. In contrast, there are a multitude of nonspecific symptoms that frequently occur in infants and children with mitochondrial disease but have a broad differential diagnosis, and more often lead to other clear diagnoses. For example, pigmentary retinopathy in a preteenage child may certainly be a feature of primary mitochondrial disease but should also evoke the possibility of juvenile neuronal ceroid lipofuscinosis or another genetic syndrome. Thus, the nonspecific symptoms, particularly if they occur in isolation, do not indicate a mitochondrial problem perse. However, when they are present in combination, the likelihood of a mitochondrial disorder increases, particularly if the nonspecific features involve different organ systems, which should prompt initiation of appropriate baseline diagnostic investigations.

Lactic Acidosis

Lactate, the product of anaerobic glucose metabolism, accumulates when aerobic metabolism is impaired, which causes a shift in the oxidized-to-reduced NAD+/NADH ratio within mitochondria (ie, decrease in the oxidized nicotinamide-adenine dinucleotide/reduced nicotinamide-adenine dinucleotide "redox" ratio). Elevated plasma lactate and/or pyruvate levels may occur in a wide range of conditions. Despite their lack of specificity, an elevated plasma lactate or pyruvate level can be an important marker of mitochondrial disease. Unfortunately, the accurate interpretation of abnormal lactate and pyruvate concentrations is not always straightforward. Spurious plasma lactate elevations commonly occur as a result of physical exercise before collection, a struggling child simulating exercise of that limb, or the use of a tourniquet, which produces venous stasis during collection. Placement of an indwelling butterfly needle or catheter to permit blood-sample collection after the patient has settled for 30 minutes can resolve erroneous lactate elevations due to poor venipuncture technique. Lactate samples are usually collected into fluoride tubes (eg, as used for blood glucose measurements). As an alternative, a blood drop can be measured at bedside with a Food and Drug Administration–approved handheld lactate analyzer. Determination of pyruvate levels can also be a challenge, because they may go up or down depending on how specimens are handled. Proper handling of blood pyruvate requires that the sample be collected in 8% perchlorate, immediately placed on ice, and rapidly analyzed. It is important to pay attention to the timing of the specimen collection in relation to mealtime, because elevated pyruvate levels can occur in the first few hours after a meal in normal individuals. Therefore, elevated plasma alanine levels, when present, may be a useful indicator of long-standing pyruvate accumulation.

Even when plasma levels of lactate and pyruvate are normal, cerebrospinal fluid (CSF) lactate levels may be elevated in patients with mitochondrial disease who have predominant brain manifestations.[12] CSF lactate levels are not influenced by collection technique, but they do rise in association with many other illnesses including seizures, stroke, intracranial infection, inflammation, and malignancy.[13] Some patients with mitochondrial disease may have normal plasma and even normal CSF lactate levels, except during episodes of metabolic decompensation, which may be the only time an increase in lactate and/or pyruvate levels will be found. Finally, the recognition that pronounced lactate and/or pyruvate elevation is not a universal finding in mitochondrial disease demonstrates its limited utility as a diagnostic biomarker. Indeed, some disease phenotypes such as Leigh disease, Kearns-Sayre syndrome, Leber hereditary optic neuropathy, and mitochondrial polymerase γ-associated diseases frequently occur with minimal or no lactate elevation.

Neuroimaging Findings

Although results of brain imaging may be normal in a patient with pure myopathy,[14]

(continued on page 8)

ASK THE MITO DOC℠

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: Greg Enns, MD, ChB, of Stanford University, Stanford.

THE QUESTION IS...

I have a 2 1/2 year old son with a Complex I mitochondrial disorder. A month after his second birthday he started experiencing episodes of vomiting. I do not think that it is cyclic vomiting syndrome, in that he only will vomit once. One day he will vomit without warning, it just pours out of his mouth with no gagging. Then for the next week he has a very poor appetite. He will start to regain his appetite and then he vomits again. When he vomits he becomes pale in color. We have had him to the doctor and they are unsure what it is. They did a blood test and nothing has showed up. Another thing that has been happening which may or may not be related is he gets a rash only on his face. It starts out as a red dot, raised into a white head and then disappears. He is not complaining of any itchiness and we are also unsure of the cause. Could the rash and vomiting be related to his diagnosis?

Another question that I have is that my husband and I would like to have another child, they told us that there was a 25% chance of having another child with mitochondrial disorder. If I start taking the mito supplements prior to and during my pregnancy will it be beneficial in preventing another child with mitochondrial disorder or will it improve the child's outcome, so that they may also have a mild case as my son does?

RESPONSE FROM GREG ENNS, MD, ChB:

I am sorry to hear about your son's episodes of vomiting. Yes, such episodes may occur in children affected by mitochondrial disease, even if a definite cyclic pattern is not present. Rashes may also occur, so it is possible that these symptoms are indeed related to his underlying diagnosis of complex I deficiency. Sometimes children who have mitochondrial disease experience a variety of symptoms related to the function of the autonomic nervous system (which functions to keep our bodies in equilibrium). Such symptoms include gastroesophageal reflux, a rapid heart rate, abnormal body temperature regulation, colonic dysmotility, migraine headaches, and blotchy rashes. In my experience, sometimes treatment with an antihistamine (e.g. cyproheptadine) can help alleviate symptoms in some patients.

From what you say about the recurrence risk you were given, your doctor likely thinks that your son has inherited an autosomal recessive form of mitochondrial disease. In such cases, an affected child inherits an abnormal copy of a gene from each parent. The risk for having another affected child is 25% in autosomal recessive conditions, regardless of taking supplements or medications. In general, siblings who are affected by an autosomal recessive condition are likely to have similar features, although numerous exceptions exist. I think it would be worthwhile to meet with a prenatal genetic counselor to discuss inheritance in more detail. I would not expect taking mitochondrial supplements prenatally to change the outcome, although it is important to take the usual recommended vitamins as prescribed by your obstetrician.

Submitting questions to *Ask the Mito Doc℠* is a benefit of UMDF membership. If you are a member and would like to submit a question, please email your question and user ID and password to askmitodoc@umdf.org. If you would like more information on becoming a member of UMDF, please email us at info@umdf.org.

THE UMDF RESPONDS TO VACCINES, MITOCHONDRIAL DISEASE AND AUTISM

As we are going to press with this newsletter, federal authorities for the first time conceded that there was a connection in one specific case between a child's autistic symptoms and the vaccines she received as an infant. Medical evaluators at the Department of Health and Human Services concluded that the child had been injured by the vaccines and recommended that her family be compensated for the injuries. The panel stated that the child had an underlying mitochondrial disease that was aggravated by the vaccines, causing the autistic like symptoms.

The United Mitochondrial Disease Foundation, in collaboration with our Scientific and Medical Advisory Board stated "There are no scientific studies documenting that childhood vaccinations cause mitochondrial diseases or worsen mitochondrial disease symptoms. In the absence of scientific evidence, the UMDF cannot confirm any association between mitochondrial diseases and vaccines."

Because of the focused attention on mitochondrial disease from this story, Charles A. Mohan, Jr., Executive Director and CEO, was a much sought after person for comment on the issue. Mohan reiterated the SMAB statement, but added that there is very little in the way of any federally funded research into mitochondrial diseases. "Mitochondrial diseases are as prevalent as childhood leukemia; however the National Institutes' of health (NIH) devotes only $11 million a year to research into mitochondrial disorders and only about one-third of that is earmarked for primary mitochondrial disease research. Many scientists believe unmasking the causes of mitochondrial disease may lead to possible cures for Parkinson's, Alzheimer's, heart disease and cancer," Mohan said.

Mohan said everyone should encourage Congress and the NIH to make research into mitochondrial disorders a high priority. "Under funding research for mitochondrial disease is a tragedy for many thousands of children and adults. An increase at the NIH level could reveal one of the mysterious missing links around autism and many other diseases."

In our next newsletter, the UMDF will have expanded information about vaccines and mitochondrial disease from members of our Scientific and Medical Advisory Board.

CHAPTER NOTES

ATLANTA CHAPTER

- ***November 3, 2007.*** Dare to Share, Inc. hosted a "Let's Put a Sparkle in their Eyes" dinner and piano recital with Sergei Pashkevich. Over $10,300 was donated to the UMDF on behalf of the Atlanta Chapter. Thank you Dare to Share for choosing the UMDF as a charitable beneficiary.
- ***March 8, 2008.*** Thanks for helping us score in the fight against mitochondrial disease at the Mito Madness Championship event held at the Peachtree Club in Atlanta, GA. Tip-off began with cocktails, hors d'oeuvres, a silent auction and live music. Special thanks to the home team Bernard and Leslie Reynolds and Chris and Mary Swinn for organizing a great event.
- ***March 17-20, 2008.*** The Beta Club, a community service group at Lanier Middle School hosted an annual awareness fundraiser in Roswell, GA. The students sold UMDF energy bands to help create awareness of mitochondrial disease. The activities were held in honor of Anna Lewis. Thanks for your hard work again this year.
- ***March 29, 2008.*** The Inaugural All Aboard for a Cure Atlanta One Mile Walk and Family Fun Day was held at Thrasher Park in historic Norcross, GA. A special thank you to Lisa Higgins and her crew of volunteers for a job well done!

CAROLINA FOOTHILLS CHAPTER

- ***January 13, 2008.*** Angie Newton and Karen Edwards participated in and finished the Disney Marathon held at Disney World in Orlando, FL. More than $12,655 was raised in honor of Alex Newton for the UMDF. Thank you ladies - you are amazing!

Angie Newton and Karen Edwards

- ***March 14-15, 2008.*** The Third Annual Caroline's On My Mind Weekend was once again a successful event. The weekend began with a golf tournament at the Spartanburg Country Club on Friday afternoon. On Saturday, a 5K Family Walk/Run was held at Duncan Park in the morning. Later that evening, a BBQ/Band Party and Silent Auction was held at the Carolina Country Club Family Pavilion to round out the weekend events. Proceeds benefit the Caroline Virginia Pulliam Mitochondrial Disease Fund (CVPMDF) with a portion being donated to the UMDF.

HOUSTON CHAPTER

- ***February 9, 2008.*** Christopher's Heart Fun Run was held at Sam Houston Park in downtown Houston. Activities included a 5K Run and 1 Mile Walk and children's activities. More than $47,000 was raised to benefit the UMDF. Thank you to the race committee for your hard work.

KANSAS CITY CHAPTER

- ***March 8, 2008.*** Rock for Mito with Ryze was held at the Sheet Metal Workers Hall in Kansas City, MO. The evening included a silent auction and live entertainment from Ryze. Thank you to Deidra Atchley for organizing this family fun event.

MINNEAPOLIS / ST. PAUL CHAPTER

- ***November 12, 2007.*** A "Girls Night Out" event was held at the Fun Sister Boutique in Minneapolis, MN. Fun Sisters opened their doors for a unique shopping experience to benefit the UMDF. Over $600 was raised that evening. Thank you to Fun Sisters Boutique for your support!
- ***December 12, 2007.*** The ONION sponsored a fun holiday event "Brewing for a Cause" to benefit the Minneapolis/St. Paul Chapter of the UMDF. "Brewing for a Cause", raising close to $5,000 was held at PICOSA. Thank you to the ONION for choosing UMDF as your charity of choice.

NEW ENGLAND CHAPTER

- ***March 2, 2008.*** The Willis family held their Annual Post-Holiday Gift Recycling Party again this year. The event was held in honor of Owen Willis and included a silent auction, refreshments and tons of fun. A $10 donation and a "re-give" item for the auction were requested from all attendees. Special thanks to the Willis family for your continued support.

OHIO CHAPTER

- ***December 9, 2007.*** The Strongsville Skating Club in Strongsville, OH was host to the figure skating show "The Nutcracker at Iceland USA". Bill Hodges of the Ohio Chapter was on hand to accept a $2,150 check in honor of Malia Kovalcik. Thank you to the Strongsville Skating Club for selecting UMDF as your charity this year!

Victoria Stanbridge with the Strongsville Skating Club and Bill Hodges

THE UMDF WELCOMES THE CAROLINA FOOTHILLS CHAPTER

The Carolina Foothills Chapter becomes the fourteenth chapter of the United Mitochondrial Disease Foundation. The Carolina Foothills Chapter will serve Northeast Georgia, Greater Charlotte North Carolina area and the Upstate of South Carolina. UMDF CEO/Executive Director, Chuck Mohan and Kara Strittmatter, Director of Member Services, traveled to South Carolina in February for the induction of the chapter. Allison Rogers will be serving as the Chapter President and was presented with a gavel from Mr. Mohan. Joining Allison on the board is Donna Pulliam as Vice President, and Donna Vinson as Secretary. Other members on the board include Christy Koury, Nancy Higginson and Elizabeth Connelly.

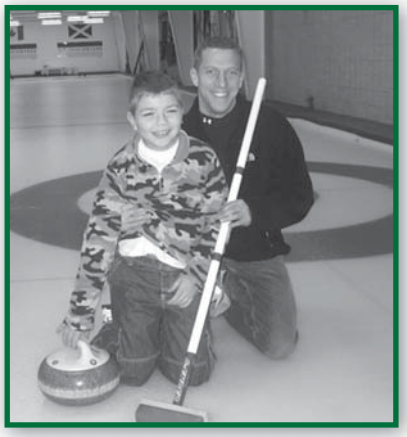
The chapter has mapped out exciting plans for the upcoming fiscal year (2008-2009) – including, but not limited to, the following: building and diversifying its board; identifying and applying for two local grants; increasing awareness of Mitochondrial Disease and UMDF with local physicians and general public; and promoting education through Grand Rounds in Charlotte as well as planning Family/Allied Health educational meetings in two separate locations.

(L to R) Allison Rogers, Chuck Mohan, Donna Vinson and Donna Pulliam

To increase awareness, the chapter will establish Mitochondrial Disease Awareness Weeks for the third week of September in both South Carolina and North Carolina – and promote events that celebrate the week. The chapter will also continue supporting current annual events benefiting UMDF such as the Caroline's On My Mind weekend, Goobers/UMDF golf outing and other local events.

OTHER NOTABLE EVENTS

- ***February 2, 2008.*** The Fifth Annual Curl-O-Rama Charity Curing Tournament was held at the National Capital Curling Center in Laurel, MD. The event presented by Chesapeake Physical & Aquatic Therapy drew 30 teams with 124 curlers including 20 children, 4 of whom are affected with a mitochondrial disease. Events included Funds for Friends at the Green Turtle, a silent auction, raffle and the learn to curl tournament. Thank you to Chesapeake Physical & Aquatic Therapy for your continued support in honor of Peter Lubelczyk.



Peter Lubelczyk and Jared Goldstein

- ***February 4-11, 2008.*** The National Honor Society at Minnechaug Regional High School in Wilbraham, MA took part in their Third Annual Themed Basket Raffle Fundraiser. Thank you to all of the students for your hard work!
- ***February 16, 2008.*** Stan Hickson participated in the Myrtle Beach Marathon in honor of his son, William. To date, $8,970 has been raised for the UMDF through this event with donations continuing to come in. Congratulations Stan for your great achievement! We knew you could do it!
- ***March 1, 2008.*** The Fifth Annual Bet on Baylee Casino Night honoring Baylee Thompson was held at the Roseville Community Center in Roseville, OH. Event activities included a Texas Hold'em Tournament, an amazing auction and entertainment by Boneyard Romance. Thank you to A.J. Hawk for your support with auction items and PSA announcements! You're the best!
- ***March 1, 2008.*** The Fourth Annual Kindbom Wine Tasting and Theme Basket Raffle was held at the Lakeside Clubhouse in Medford, NJ. The event was held in honor of Rachel Kindbom with proceeds benefiting the UMDF through the Angelray Research Fund. Thank you to the Kindbom family for your continued support!
- ***March 14-16, 2008.*** The Fourth Annual Writers at the Beach: Seaglass 2008, a three-day writers conference, was held in Rehoboth Beach, DE. The annual event was held in memory of Sam and Zachary Juhlmann. For a recap on the weekend, go to www.writersatthebeach.com.
- ***March 14-15, 2008.*** Preston's March for Energy was held during the 2008 Shamrock Marathon in Virginia Beach, VA. Preston's March was held on March 15th with a celebratory reception for all participants in the evening. A special thanks to Jerry and Amy Frostick - this year they opened up 100 charity spots for the half marathon for $150 each - and they sold out within 4 days! For detailed information, go to www.shamrockmarathon.com.
- ***March 22, 2008.*** The Annual Brittany Wilkinson Dinner and Auction was held at Break the Barriers in Fresno, CA again this year. A great time was had by all and a special thanks goes out to the Wilkinson family and friends for hosting another fantastic event in honor of Brittany. All proceeds from the event benefit the UMDF through the Brittany Wilkinson Research Fund.
- ***March 29, 2008.*** Mito Jam at Club Aurora was held in Taunton, MA. The concert featured entertainment from Whitman, MA native Steve Lanza, Jr. and friends; Richmond, VA's John Moossa and Easton, MA recording artist Dave Spagone. The Richmond, VA and South Eastern, MA musicians joined forces for this family friendly concert to raise awareness that will hopefully result in a cure for mitochondrial disease.

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For more information about a chapter, group or ambassador near you, contact the UMDF office at 888-317-UMDF!

UNITED MITOCHONDRIAL DISEASE FOUNDATION
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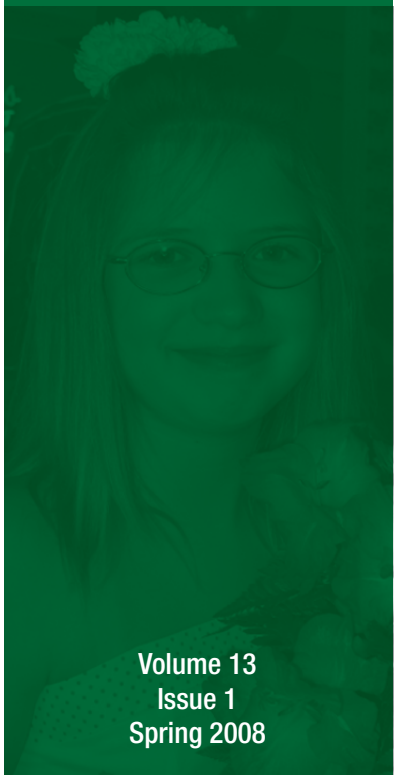
PARTNERS IN PROGRESS

MITO FACTS

- A "red flag" for mitochondrial disease is when a child or adult has more than 3 organ systems with problems.
- Confirmation of diagnosis of mitochondrial disease is usually possible through various means, including blood and urine tests, brain MRI, muscle and skin biopsies.
- In a person with mitochondrial disease, the mitochondria are failing and cannot convert food and oxygen into life-sustaining energy.



Volume 13
Issue 1
Spring 2008

DETERMINATION REPLACES ENERGY

A Day in the Life of the Richards Family

Krystena Richards is a UMDF Ambassador, a mother of three beautiful boys (with a fourth on the way), a caring wife, an aspiring photographer and she is living every day with a mitochondrial disorder.

As Krystena was growing up, she never really felt "good". She was sick all the time and battled with extreme leg and stomach pains that left her screaming in pain. At the age of 8, she was diagnosed with gastritis of the stomach and was put on a grease-free diet, which seemed to help with the stomach pains.

Krystena suffered from many symptoms throughout her early adult life, such as, extreme fatigue, one-sided numbness, ptosis of one eye, unstable blood pressure, neurological symptoms and finally a metabolic stroke. After her stroke, the battery of tests started; the testing began with "common tests" for multiple sclerosis, lyme disease, other autoimmune and infectious disease, all of which left her without answers.

Those answers did not come until after the birth of her second son, Caden. Krystena's doctors suspected that her illness was related to the host of problems that Caden had been experiencing and that the stress of taking care of a sick child was the reason behind her symptoms. After a long road, Caden finally received a blood test indicating that he had a rare mitochondrial disease. "This was the turning point in our lives" Krystena says. The geneticist they took Caden to see informed the Richards that Krystena's illness was likely related to their son's. Krystena eventually has tested and her test came back positive for a mitochondrial disease.

When Krystena was 25 years old, she finally had some answers. Krystena and her son Caden were diagnosed with a variant form of MADD/GAII, which is an autosomal recessive mitochondrial disease. After diagnosis, Krystena and Caden started taking medication and supplements and finally began to see improvements.

Krystena and her husband Mike live life to the fullest with their three boys, Warren, 13, Caden, 9 and Carson, 4, who was also diagnosed with MADD/GII shortly after birth. "We take advantage of each and every precious day we have together," Krystena said.

Krystena is currently expecting their fourth son! From what the doctors have told her, she was not able to have any more children due to her low hormone levels and polycystic ovarian syndrome. "I am elated and worried at the same time," she said. "Our baby has a high chance of inheriting this disease; I cannot say that his life will not be worth living, even if he is affected." The pregnancy has been very hard on Krystena and she has been in and out of the hospital with complications and is unable to maintain her own body weight. However, the baby seems to be doing well and is already in the 66th percentile for his weight at just 22.5 weeks gestation.

"I WILL NEVER EMBRACE MITOCHONDRIAL DISEASE AS IT HAS TAKEN SO MUCH FROM OUR FAMILY BUT, I DO STILL STRIVE TO FIND THE POSITIVES IN LIFE," KRYSTENA SAID. "I CHOOSE TO LOOK AT LIFE WITH MY EYES WIDE OPEN TO ALL OF THE FACTUAL POSITIVES."

Krystena made a very easy decision in 2006 to become a UMDF Ambassador in Louisville, Kentucky. "I want nothing more than to help other people looking for support," Krystena said. "Living with mitochondrial disease, whether it be yourself or your child that is affected, can be a lonely place without a support group and other resources."

FROM THE CHAIRMAN

Since this is my first "Chairman's Message," please allow me to express thanks to the Board of Trustees for placing their faith in me as I continue to serve the United Mitochondrial Disease Foundation in this new capacity. I am honored to accept the position although this new role comes with a mix of sadness and excitement. I am the Chairman only because our previous chairman, Stan Davis, passed away. Stan was a tremendous friend both to me and the UMDF. He had a personal stake in our goals and worked tirelessly to promote the UMDF mission to find a cure. I learned much from Stan when I was Vice Chairman, and he will be tough to replace, but I will strive to do my best.

I would also like to take this opportunity to inform you about our new Executive Board. Sharon Shaw, who was serving as Secretary, is now our Vice Chairman. Marty Lyman joins the Executive Board as our Secretary and Richard Kubach, Jr. will remain as Treasurer. Also, all of us on the Board of Trustees would like to welcome back a long-term friend to the board. John DiCecco, a previous Board Chairman and trustee accepted another term on the Board. I can truly tell you that we have a very active board and each of us is committed to bringing their unique skills to benefit the UMDF.

In preparing this note, I have reviewed many of the past UMDF newsletters. They helped me reflect on the fact that our membership continues to grow as do the fundraising and awareness activities held by our chapters, groups and ambassadors. It is through your hard work and dedication that we continue to move forward and you all deserve the credit and our thanks. Visit our newly designed website for more information on how you can be involved.

Who would have thought just a few years ago that the United Mitochondrial Disease Foundation would be the largest, non-governmental funding source for research towards a cure for mitochondrial disease? I encourage you to read fellow trustee Rick Leach's article about advocacy. Rick tells us how the hiring of Porter Novelli will help us reach the goal of attracting more funding into primary mitochondrial disease research.

Our annual symposium continues to be the premier national event that brings together medical experts, researchers and clinicians along with affected families in an effort to network our resources to answer questions, offer support and ultimately find a cure. Our return to Indianapolis for our next symposium, "INDY 2008: Setting the Pace in Mitochondrial Medicine" is just a few months away. Scientific and clinical meetings will be held at the Hyatt Regency Hotel in Indianapolis, IN on June 25-28. Family meetings are scheduled June 27 and 28.

As you may know, my family has been on the mitochondrial road for almost 25 years now. We know and have experienced the decade long search for a diagnosis, the lack of physicians' knowledge of the disease, the day-to-day problems of our daughter's (Kelsey) fight with the disease and the longing for a cure. I have attended all but one of the symposiums since UMDF was founded and have witnessed the evolution of this organization. I see great growth and a very positive change. I feel very strongly that we have reached a milestone in this organization and in the awareness of the disease. We have a long way to go but I sense an intense excitement and enthusiasm because the future is bright for this cause.

Energy to all,

W. Dan Wright

REMEMBERING FORMER UMDF CHAIRMAN - STAN DAVIS

I never heard of this before, what is a mitochondrial disease?" "Tell me what I can do to help my granddaughter. I'll do whatever it takes." And with that Stan Davis made a commitment to become part of the cure rather than a victim of the disease, he joined the quest toward a cure!

Stan contacted and engaged his family, friends, neighbors and business associates educating them and involving them in his quest. They sponsored, supported and participated in his annual golf outing, the largest and most successful golf outing supporting the UMDF mission.

Stanley, in 2001, accepted a seat on the UMDF Board of Trustees where his business skills positioned him well for the office of Treasurer. As Treasurer and member of the UMDF Investment Committee he helped guide the positive placement of UMDF investments helping insure support for growth of UMDF's Research Grant Program.

Stanley continued his involvement in UMDF by serving as Vice Chairman and most recently as UMDF's 2007 Chairman. During his tenure UMDF increased its Chapters and groups to over 60, membership increased by 25% and this year we will issue our largest research grant award pushing our cumulative total to over $6,000,000.

The accomplishments and achievements have not come without tremendous costs. While many continue with daily routines many lives continue moment to moment consisting of abstract routines which, out of necessity, have been adopted as normal.

Life continues to show us the futility of set plans. We don't plan lives, we live them and deal with them the best way we can.

Stan Davis will not be remembered only by his words, but by his kind deeds. His Life will not be measured by the breaths he took, but by the moments his actions took our breath away.

Stanley Davis
August 12, 1942 – January 19, 2008

There is a purpose for everything that happens. The timetables and schedules that rule our life are of our creation. God's calendar and schedules are His and do not consist of years, months, weeks, days or the promise of tomorrows, but consists of a lifetime and a promise of eternity. The lifetime is finite with a beginning and an end while eternity knows no time. God does not wear a watch.

Perhaps Stan's purpose was to help us understand that our response to this life must never be measured by time or tomorrows but by doing and today's. Perhaps we are to understand that God's time may not always have a tomorrow to love, to hold, to care, to smile, or to apologize. Tomorrow's time belongs to God and God does not wear a watch.

Thank you Stanley Davis for being part of the "quest towards a cure."

Great thoughts speak only to the thoughtful mind, But great actions speak to all mankind. - Emily P. Bissell

SUPPORTING OUR CAUSE

BI-LO CHARITY CLASSIC

For the last two years, the BI-LO Charity Classic has selected UMDF as one of the many charities it supports through its golf tournament. BI-LO generously donated $5,000 in 2006 and 2007 to support medical research into mitochondrial disease.

The BI-LO Charity Classic, held every June in Greenville, South Carolina, is the largest, 1-day golf tournament in the country.

Over the past 24 years, the tournament raised more than $39 million for non-profit organizations in BI-LO's four-state area of South Carolina, North Carolina, Georgia and Tennessee. UMDF's Carolina Foothills Chapter is in the heart of BI-LO's giving area.

In 2007, the event raised over $5.5 million 2007 and benefited over 500 charities. The grants are distributed every November during Bi-Lo's "10 Days of Giving." Organizations that are eligible include those that help children, deal with hunger or are involved with education.

More information on the golf tournament can be found at www.bi-locharityclassic.com.

UMDF is deeply grateful for Bi-Lo's support of our mission. Thank you!

WHAT THE DEVELOPMENT DEPARTMENT CAN DO FOR YOU

- Help write requests and gather any necessary attachments for organizations that might donate to your event
- Help approach potential donors and prepare sponsor materials
- Assist in putting together requests for workplace giving programs
- Follow up on a lead to a charitable foundation that might donate to UMDF

WHAT YOU CAN DO FOR THE DEVELOPMENT DEPARTMENT

- Inquire at your workplace about programs that match gifts or volunteer hours
- Write UMDF on your workplace giving pledge card (i.e. United Way or Community Health Charities)
- Let us know if you hear of any grant opportunities in your area

RESEARCH SPOTLIGHT - Haya Lorberboum-Galski, PhD

In 2006, the UMDF awarded a grant in the amount of $115,000 to Haya Lorberboum-Galski, PhD, of Hebrew University of Jerusalem in Israel to investigate the possibility that enzyme replacement therapy can be used to treat LAD deficiency.

The many different medical conditions that are placed under the category "mitochondrial disease" have in common the disruption of one or more components of the energy "machinery" of the mitochondria. This machinery is made up of a number of enzymes that are crucial to the production of the energy molecule ATP. If even one of these enzymes is defective or absent, then ATP production may be greatly reduced and toxic metabolites may be accumulated, with serious consequences for organs such as the brain and skeletal muscle that depend upon large amounts of ATP for normal development and activity.

Lipoamide dehydrogenase (LAD) is an important component of three enzyme complexes in mitochondria and LAD deficiency is an inherited disease that disrupts normal mitochondrial function.

Prof. Loberboum-Galski and her colleagues at the Department of Cellular Biochemistry and Human Genetics at Hebrew University are conducting research to see if enzyme replacement therapy can be used to treat LAD deficiency. They are developing methods for placing the normally functioning enzyme into cultured cells of mitochondrial disease patients and also into the mitochondria of laboratory mice that are afflicted with the disease. This is a promising approach because there are currently no cures for mitochondrial disease and such research could lead to therapies that would correct its fundamental causes.

UNITED MITOCHONDRIAL DISEASE FOUNDATION®
HOPE. ENERGY. LIFE.

UPCOMING EVENTS

ATLANTA CHAPTER

- ***June 7, 2008.*** An outdoor movie night and family fun activities at Joint Venture Park at Dave's Creek in Cumming, GA. For more information, contact Nadine Mancuso at pnm711@bellsouth.net.
- ***September 27, 2008.*** Second Annual Music for Megan Charity Benefit Concert at Wills Equestrian Park in Alpharetta, GA. Go to www.meganshope.org for more information.
- ***October 6, 2008.*** Fifth Annual Fore-A-Cure Golf Tournament at The Standard Club in Duluth, GA. For information, contact Chris Swinn at atlchapter@umdf.org.

HOUSTON CHAPTER

- ***October 15, 2008.*** First annual Golf Tournament at the Forest Hills Course at the Clubs of Kingwood. For more information or to join the Golf Committee, contact Tom Zyroll at houstonchapter@umdf.org

INDIANA CHAPTER

- ***April 5, 2008.*** Indiana Ice and UMDF night of family entertainment at Conseco Fieldhouse. $5 from each ticket purchased on UMDF's behalf will be donated to the foundation. For information and to purchase tickets, contact Matt Duncan at 317-925-4423 ext. 218.

KANSAS CITY CHAPTER

- ***June 21, 2008.*** Kansas City Mito What? Family Fun Run in Lake Waukomis, MO. For information go to www.umdf.org/kcmitowhat, or contact Theresa Edwards at 816-419-8836 or tkedwards@kc.rr.com.

NEW YORK METRO CHAPTER

- ***May 10, 2008.*** Eighth Annual Matthew Dudgeon Memorial Walk and 7K Run at the Chester Train Station in Chester, NY. Followed by a Dinner Dance at in the evening. For information, contact Gina Dudgeon at gdudgeon@mw.k12.ny.us or visit www.themattyfund.org.

OHIO CHAPTER

- ***April 11, 2008.*** Fifth Annual Guest Bartender Night at 82nd Street Grill and Pub in North Royalton, OH in honor of Kyle Kobunski. For information, contact John Kobunski at jkobunski@aol.com.
- ***May 10, 2008.*** Seventh Annual Run Wild for a Cure 5K Run and 1 Mile Walk at the Cleveland Metroparks Zoo. For information, visit www.runwildforacure.org.

AROUND THE COUNTRY

- ***April 19, 2008.*** Fifth Annual Mito What? Family Fun Day at the Shrine of Our Lady of the Snows in Belleville, IL. For information, visit www.umdf.org/stlouisrace, or contact Marsha Hohe at 618-233-6919 or Joni Schnitzler at 618-624-0216.
- ***April 19, 2008.*** Emmyfest 2008 for Mitochondrial Awareness Charity Benefit Concert at the Capital Ale House in Richmond, VA in honor of Emily Jurek. For information, visit www.umdf.org/emmyfest or email jmbmoose@hotmail.com.
- ***May 2, 2008.*** Third Annual Josie Mazzo Children's Charities Golf Tournament at Greystone Golf Club in Dickson, TN. For information, visist www.josiemazzo.org or contact Mendy Mazzo at mendy.mazzo@skanskusa.com.
- ***May 4, 2008.*** Stephanie Watkins and friends will participate in the Frederick Running Festival in honor of Stephanie's son Noah. For information and to sponsor Team Noah visit www.umdf.org/teamnoah.
- ***May 10-17, 2008.*** Eighth Annual Kites for Kristen at St. Daniel the Prophet School. Activites include a kite decorating contest with prizes and a dress down day at the school. Proceeds to benefit the UMDF through the Kristen Charleston Research Fund.
- ***May 31, 2008.*** Second Annual Greater Mito Open Golf at Old Highlander Golf Course in memory of Zachary & Sam Juhlmann. For information, contact David Dobke at 262-853-4045 or ddobke@wi.rr.com.
- ***June 7, 2008.*** Sixth Annual Pittsburgh "One Step Closer to a Cure" 5K Run and 1 Mile Walk at the North Park Boathouse in Allison Park, PA. For information, visit www.umdf.org/pittsburghrace or contact info@umdf.org.
- ***Summer, 2008.*** The DC Area Mito Group is in the process of launching a cookbook fundraiser that will be available this summer. Submit your favorite recipes! For information, contact Margaret DeLacy at rijac970@yahoo.com or Harriet Ulrich at hsulrich@verizon.net.

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.

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GIFTS FROM THE HEART

- Ed Mansell of North Olmstead, OH celebrated his birthday this year with a very special party. In lieu of gifts, family and friends were asked to make a donation to the UMDF. Over $1,230 was raised in honor of Ed's daughter Robin Bryson.
- A BBQ Cookoff was held at the Houston Livestock Show and Rodeo in Houston, TX. The Cut Above cooking team raised $250 in memory of Christopher Schindler and HLSR friend Henry Lehnoff.
- The faculty and staff at Spring-Ford High School in Royersford, PA held a Jeans Day and raised $306 in honor of Maddie Connaughton.
- The students at Trinity South Elementary School in Washington, PA pulled together to collect pop tabs and raised $75 in honor of Ellie Kovalcik.
- Connor Clifton of Pembroke Pines, FL celebrated his third birthday and over $100 in donations were sent in honor of Connor.
- An Alternative Gift Market was held at the Franklin United Methodist Church in Franklin, TN and raised $465 in honor of Andy Garrison.

Don, Andy and Nancy Garrison

- The Nathan Mowrer Annual Playground event was held on August 18 in Pottstown, PA. The event raised funds for a playground, and donated a portion to the UMDF in memory of Nathan Mowrer.
- The Knights of Columbus of Chatham, IL held a Texas Hold'em Tournament and raised $250 in honor of David Bauer.
- The members of the Colonial Football Officials Association of Western CT raised $183 in honor of Jenny Schnitzler.

Ball Ground Elementary School Student Council, the school raised over $3,100 with a Coins for a Cure campaign.

- A Fall Festival was held on November 3 at the Heartland Kids Pediatric Clinic in Orange Park, FL. Cotton Candy was sold and over $102 was donated to the UMDF.
- The Renn Wealth Management Group, Inc of Atlanta, GA donated $1,000 to the UMDF in honor of clients and friends of the organization.
- Guests of Rachel Langer's Bat Mitzvah donated $4,893 in memory of Rachel's sister, Sarah Langer.
- A dress down day was held at the Lands' End Inlet Store in Rochester, NY and raised $135 in honor of Kevin Honan.
- The Art Beins Karate Center in Freehold, NJ raised $1,335 in honor of Nicholas Nunno.
- A Beef and Beer Fundraiser was held in New Jersey in honor of Ryan Donnelly. The amazing event raised $4,000 for the UMDF.
- Dr. Martin Markowitz of Chagrin Falls, OH celebrated his 70th birthday this year! Friends and family generously donated $95 on his behalf.
- A Honda Goldwing Anniversary Party was held in Saginaw, MI in memory of Jake Kubczak. The event raised $301 for the UMDF.
- The Nova Scotia Club in Trenton, MI held a basket raffle and raised $250 for the UMDF in honor of Brian MacDougall, Samantha Bommarito and in memory of Sean Clark.
- A Recess fundraiser was held at Ellie Bube's school in Seattle, WA. The event raised $35 in honor of Ellie's brother Jacob Bube.
- A Quinnipac fundraiser was held in Hamden, CT in memory of Elijah Bonney. The event raised $167 for the UMDF.
- Tyson's Foods in Pine Bluff, AR held a Halloween Dress Down Day in honor of Jude Manley and contributed $958 to the UMDF.

ONGOING FUNDRAISERS

COINS FOR A CURE℠

The following families participated in the Coins for a Cure℠ campaign and sent in funds raised in the months of October, November and December:

- The Norrod Family
  in honor of Daniel Norrod
- The Blake Family
  in honor of Jude Manley Research Fund
- The Wehling Family
  in honor of Gail Wehling
- The Olenderski Family
  in honor of Walter Olenderski
- The Polo Club
  in memory of Stan Davis
- CulinArt, Inc
  in honor of Michelle Mohan
- Ball Ground Elementary
  in honor of Colton Freeman

TEA FOR MITO℠

The following family participated in Tea for Mito℠ and sent in funds raised in the months of October, November and December.

- The Simon Family
  in honor of Ainsley Higgins

A special thank you to all of these families and to the families who are still participating in Coins for a Cure℠ and Tea for Mito℠!

most patients with mitochondrial disease with central nervous system involvement do show MRI abnormalities.[15,16] A nonspecific, delayed myelination pattern may be seen early in the course of the disease.[14,16,17] In addition, certain MRI findings are highly sensitive and specific for mitochondrial disease. The most common specific MRI finding is a symmetrical signal abnormality of deep gray matter, which is seen as a high signal on T2-weighted and fluidattenuated inversion-recovery (FLAIR) MRI with a low T1-weighted signal. Any deep structure can be involved, with the character of the lesion being either patchy or homogeneous.[18] Leigh disease is the prototype mitochondrial disease in which imaging findings may show involvement of the brainstem, diencephalon, basal ganglia, and cerebellum, although symmetric basal ganglia lesions are the most common finding. mtDNA-deletion disorders often involve cerebral and cerebellar atrophy with bilateral thalamic and basal ganglia lesions.[18–20] In contrast, the imaging landmark of mitochondrial myopathy, encephalopathy lactic acidosis, and stroke-like episodes (MELAS) is infarct-like lesions that may appear only transiently and are not confined to vascular territories.[21–23] Obtaining diffusion-weighted MRI sequences during a stroke is critical to the diagnostic workup, because lesions show an increased diffusion coefficient in mitochondrial disease but a significantly reduced diffusion coefficient in acute ischemic stroke.[24–26]

Brain proton (^{1}H) magnetic resonance spectroscopy (MRS) is a newer modality that can be obtained at the time of brain MRI to noninvasively measure CSF and brain lactate levels to aid in the diagnosis and monitoring of mitochondrial disease.[14,21,27,28] However, proton MRS abnormalities are usually only present in patients with central nervous system involvement rather than patients with "pure" myopathy. As with all diagnostic testing for possible mitochondrial disease, no single imaging test accurately defines all patients (see www.mitosoc.org and select "diagnosis toolkit" for a more extensive discussion of typical MRI and MRS findings in specific mitochondrial diseases).

The Older Child and Young Adult

Mitochondrial disease may present at any age. The symptomatic presentation of mitochondrial disease in older patients differs from that seen in infants and young children. The general rule that "the more severe the metabolic disorder, the earlier it presents in life" generally applies to mitochondrial disease. Later-onset primary (genetic) mitochondrial diseases tend to follow a chronic course, although exceptions abound. Patients may report general good health until they develop insidious signs of chronic disease or neurologic symptoms. Isolated myopathic and/or cardiomyopathy presentations, frequently with exercise intolerance, are common in teenagers and young adults. The diagnosis of fibromyalgia or chronic fatigue syndrome may be considered before that of mitochondrial disease. In contrast, a rapidly progressive disease course may be seen with sudden regression, often in association with a physiologic stressor such as a viral illness or bacterial infection, other severe illness, pregnancy and delivery, or surgery. Regression may manifest as nonvascular stroke, ophthalmoplegia, visual decline, mental status changes, an array of new neurologic complaints, or worsening exercise tolerance and fatigue. It is important to recognize that the first episode of metabolic stroke in MELAS, or metabolic encephalopathy in Leigh disease, may be fatal at any age.

Part II of Mitochondrial Disease: A Practical Approach for Primary Care Physicians will be continued in Mitochondrial News Volume 13 Issue 2 coming out in June 2008.

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PROMISE OF PEACE By Suzanne Cizek Moore

The gift came in the way of a wishbone. Silver, and strung on red thread. The card said, 'Put this on and make a wish. When the necklace breaks, your wish will come true.' My thought was it wouldn't last long. It looked so fragile. My wish was easy.

I am 48 and have a rare mitochondrial disease. It is chronic and progressive. I will die from it.

Despite the disease, I've lived a fairly normal life. For the past 14 years, I have lived in a condo in Palma Ceia, FL. Six years ago, I married my husband, Rick, and soon after the symptoms worsened. I lost more weight and strength. My eyes became completely paralyzed and I was declared legally blind. My hearing diminished, and I can only walk short distances with a cane.

I was a skinny kid. I remember mom worrying about me. Every school morning, she would line up seven sandwiches on the counter. Two each for me and my brother, one each for my three sisters. But I never ate much, and everyone wanted me to eat. Little did we know it was part of a disease. At 15, a routine physical led to a diagnosis of a mitochondrial disease, an amazing catch that long ago. It was close, but not the correct diagnosis. That didn't come until two years ago.

My disease is called MNGIE, short for Mitochondrial Neuro Gastro-Intestinal Encephalomyopathy. Less than 200 cases have been diagnosed worldwide. It's a multi-system disorder. The digestive system is especially affected, everything from swallowing to eliminating. This means I am unable to eat normal amounts of food. Eventually, I will probably starve.

I spend my days trying to get down 1000 calories. I drink a dietary supplement mixed with fortified milk, a vitamin cocktail, a little yogurt or cream of wheat, and what I call 'Thanksgiving in a cup'. This last consists of chicken, carrots, onion, potato, celery and garlic, simmered in chicken broth and pureed. Meeting my nutritional quota each day is a huge effort. I am 5 feet 7 inches and weigh 94 pounds. One day at a time, I tell myself. This is my job.

Last August, I decided I needed a break. I wanted to be rescued. With admission papers in one hand and my husband holding the other, I moved into a private hospital room. Rick slept in a cot and my bed was, well, not my bed. No one seemed to know who I was and why I was there.

I was to have a feeding tube inserted, but after repeated attempts, this failed. After two weeks with little nutrition, a severe loss of weight and energy, and with my health deteriorating rapidly, it became apparent I would have to save myself. If I was going to live any longer, I would have to abandon the hospital and take my chances at home.

I was sure my death was near. I called my family to come. It was as if I was cramming for a test and I wanted to get an A in life. I could have been a better person, not so judgmental. I know the closer I get to death, the better student I become.

I am tired. I don't fear death, but I do fear the process of dying. It's so scary. I'm losing control. I'm losing my looks. I keep thinking, I wish I could go out and sweep the front porch or the back patio. What I would give to do a load of wash. Just little things. I'm at the mercy of others.

Now I'm a hospice patient. I hate the connotation. There's no 'Get well soon'. But, the support from hospice is invaluable. Some friends struggle with the 'D' word. They do what I describe as ducking. Avoiding phone calls. Promising visits 'tomorrow.'

I am making plans for my memorial service. I guess this is one good thing about my situation. I can do that. While most people might not want to think about their memorial service, I find comfort in knowing that I have control over something. My family is thankful to have my wishes met. I want to have bubbles and butterflies. There is a quaint courtyard with a flower garden where the butterflies can fly free. All my friends will be there, I want to be remembered by their words.

Black and white photos of my family cover my living room wall. My earliest memories are of my dad snapping pictures of us. Every day I talk to my sister, Ann Starr. As the oldest child, she is the captain of the family. My mother tells me I can call any time, day or night. It's hard for my family to watch me grow weaker. Especially for my parents. This is not the natural order.

I have been nourished with love and support. If that were the cure, I would be healed. The reality is, even with all the comfort and care, I am in this alone. Every waking minute I fight to stay alive, but death is solitary.

The silver wishbone came from a friend in Miami. Fifteen weeks later, I still wear it. The string is as fragile as I am, but it has not broken. I didn't think either of us would last this long. For now I try to appreciate each day and look forward to getting my wish: peace, peace beyond understanding.

AACT MISSION

To ensure equal representation and service of the affected adult community to the affected pediatric community -- and to better represent, serve and assist adults with mitochondrial disease.

AACT QUESTIONNAIRE

The Adult Advisory Council Team needs your input! All adults (18 years of age and older) who are affected with a mitochondrial disease are asked to complete an online questionnaire. The questionnaire can be accessed by going to the "For Adults" section of our website at www.umdf.org. Preliminary results will be available online; however the questionnaire will be ongoing. If you do not have access to a computer, and would like a copy of the questionnaire to complete, please contact info@umdf.org.

ADVOCACY NEWS

BY RICK LEACH, UMDF TRUSTEE AND CHAIRMAN OF GOVERNMENTAL AFFAIRS COMMITTEE

It should come as no surprise to you that the United Mitochondrial Disease Foundation is the leading non-governmental contributor of grants focused on mitochondrial disease research. In the past decade, the UMDF has funded more than $5 million towards our mission of funding the research to find a cure. What you might find surprising is that the National Institutes of Health (NIH) has an annual budget of $29 billion. $11 million of that amount is spent on various research projects dealing with mitochondrial dysfunction. NIH funds about $3.6 million annually on primary mitochondrial disease research. This is the research that could lead to a cure.

The goal of our advocacy effort is to significantly increase governmental funding for research into mitochondrial disease. We have already taken steps in furtherance of this goal. For example, we worked with Members of the U.S. Congress to urge NIH to support additional mitochondrial research (the U.S. Congress allocates the funds for NIH). Congress included language with the 2008 NIH funding bill that "encourages NIH to intensify its research efforts into primary mitochondrial disease…" This is an important step in the longer-term process of building a broad base of support for more research among key policy-makers.

We have also taken steps to expand the UMDF's advocacy capacity by securing the services of a national public relations firm (generously supported by an individual donor). Specifically, the firm will help us develop an effective way to discuss mitochondrial disease and the potentially far-reaching implications of research. There have been numerous studies demonstrating that mitochondrial dysfunction is implicated in other diseases such as Parkinson's, Alzheimer's, diabetes, and Gulf War Syndrome. Intensified research into mitochondrial disease could help to further understand these other conditions.

The public relations firm will also prepare the range of materials needed to effectively communicate with policy-makers and other targeted audiences. They will also develop an advocacy component for our website. They will engage in an extensive media outreach effort to ensure that more stories related to mitochondrial disease appear in print and electronic media and via the internet. This will help raise our profile and increase public understanding of the importance of research. In addition, we are working with the firm to develop a Tool Kit for use by the UMDF members and chapters that will help you reach out to your elected officials. We will present the Tool Kit and provide an update on our advocacy effort at the meeting in Indianapolis.

In selecting the public relations firm, the UMDF board of trustees issued a request for proposals to leading firms in Washington, D.C. Five proposals were reviewed by the board. In December 2007, several board members and UMDF staff met in Washington D.C. with the two finalists. Following final presentations by these firms and further consideration by the board members and staff, the firm of Porter Novelli was selected.

We've just started this journey, but it will be one in which we will need you to participate. We look forward to seeing you at the UMDF Symposium.

MEMBER RESOURCES

THE UMDF JOINS FORCES WITH CARE PAGES

The United Mitochondrial Disease Foundation now offers patients, their families and friends access to CarePages.com, healthcare's largest social network to provide the emotional support needed for healing and well-being.

CarePages.com enables those involved in a health event to create private web sites for sharing health updates, photos and supportive messages. CarePages.com also has resources to help people cope with the emotional challenges of a health event, including inspiring stories, tools to meet others in similar situations, and practical tips and advice on "what to do" and "what to say" during difficult and stressful situations.

"CarePages helps the UMDF to continue to allow families to share their personal journeys," said Chuck Mohan, Jr., Executive Director and CEO. "Setting up a personal page is easy. Just click on the link under resources on our web site or visit www.carepages.com/umdf and follow the easy directions".

CarePages.com is secure and compliant with all HIPAA requirements. It ensures that patients, families and healthcare providers are protected online through strict privacy policies, password-protection and visitor management tools for Care Page Managers.

CarePages℠
connect, share...care.

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!

The United Mitochondrial Disease Foundation, The Mitochondrial Medicine Society, The Mitochondria Research Society, Mitochondrial Physiology Society and The Cleveland Clinic

Present

INDY 2008

Setting the Pace In Mitochondrial Medicine

Hyatt Regency Indianapolis • Indianapolis, Indiana
Scientific Program June 25-28, 2008 • Family Program June 27-28, 2008

RESERVE YOUR HOTEL ROOM NOW TO RECEIVE THE UMDF SPECIAL RATE!

Registration brochures are being mailed soon, and online registration is available at www.umdf.org!

Cleveland Clinic

Mitochondrial Physiology MiP

The Mitochondria Research Society MRS www.mitoresearch.org

The Mitochondrial Medicine Society MMS www.mitosoc.org

ANNOUNCEMENTS

HEARTSTRINGS℠ AND LEAP℠ AWARDS

The UMDF wants to recognize our champions and heroes, and we need your help! Nominations are open for the 2008 UMDF Heartstrings℠ and LEAP℠ awards. You can download and print the appropriate forms from the UMDF website, or you can request the forms by mail or fax by contacting us at info@umdf.org or at 888-317-UMDF. Submissions must be received or postmarked by April 25, 2008.

UMDF.ORG GETS A NEW LOOK

The UMDF is pleased to announce the launch of our new website this past February! If you haven't already done so, we encourage you to visit www.umdf.org to see the new features. The website is divided into two sides. The first includes information for "Patients, Families and General Public." The second has information for "Scientists, Physicians and Healthcare Professionals." The new website will be a work in progress, and we encourage your input. If you have a comment, suggestion or question regarding the website, please contact us at info@umdf.org.

NEW UMDF STAFF MEMBER

The national office is very pleased to add another new face to our professional office staff. We would like to introduce you to Scott Precopia, who is our first full-time IT Manager.

Scott joined the UMDF in January. He will be responsible for all of the computer-related issues within the UMDF. Before coming to the UMDF, Scott worked as NOC Manager for a Pittsburgh-based, professional services IT company. Prior to that, he owned two businesses where he honed his technical skills and gained a vast knowledge of the IT industry, business and technology.

MITOCHONDRIAL DISEASE AWARENESS WEEK IN GEORGIA

On February 5, 2008, the Georgia General Assembly adopted HR1188, which designates the third week of September as Mitochondrial Disease Awareness Week. Georgia becomes the third state to adopt this designation, joining New Jersey and Wisconsin. In 2007, Massachusetts, Ohio, Washington and Fresno and Clovis City in California had received a one time proclamation designating the week of September 16-22, 2007 as Mitochondrial Disease Awareness Week. Pennsylvania had also received a one time proclamation in 2007. The Chapters of the UMDF are busy trying to get a permanent bill passed in their state for the third week of September. If you are interested in learning how you can get a permanent bill passed in your state, contact info@umdf.org or visit www.umdf.org to download the "Mitochondrial Disease Awareness Week" toolkit.

SUBMISSION DEADLINE FOR ISSUE 2 IS APRIL 30, 2008!

UNITED MITOCHONDRIAL DISEASE FOUNDATION®

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