POLG MUTATION
A dialogue with Bill Copeland, PhD

Question: What is POLG?
POLG is the name of the gene that codes for a protein that functions as the mitochondrial DNA polymerase, also called DNA polymerase gamma or pol γ for short. Actually the mitochondrial DNA polymerase is made of two proteins, pol γ and a helper protein encoded by the POLG2 gene.

Question: What is the function of POLG?
Pol γ is a DNA polymerase and the function of DNA polymerases is to copy our DNA, such as from mother to daughter cells during DNA replication. DNA polymerases are protein machines (enzymes) that carry out the faithful copying of DNA. DNA is a polymer of nucleotide bases, adenine, guanine, cytosine, and thymine, abbreviated A, G, C, T, respectively. When DNA is copied (from one to two copies), the DNA polymerase reads a template strand of DNA (reading the four bases) and incorporates one of four nucleotide triphosphates (A, G, C, T) opposite in the correct base position in DNA. The DNA polymerase provides the environment for the correct pairing of A with T and G with C and carries out the chemistry to join the nucleotide bases together. There are 16 DNA polymerases in human cells, but only one functions in the mitochondria - pol γ. That means that in the nucleus there is a wide variety of redundancy of function, but not so in the mitochondria. Mitochondrial DNA (also called mtDNA) replication has to rely totally on this one polymerase to copy mtDNA. Also, as the only DNA polymerase in mitochondria, pol γ has to function in DNA repair of mtDNA as well as DNA replication.

Pol γ has an impact in mtDNA diseases in three ways:
1. As the only polymerase in mitochondria, pol γ is responsible for making most all of the germ-line mtDNA mutations (mutations in germ cells that pass on the genetic information to the next generation) and over 80% of somatic mtDNA mutations (mutations anywhere in the body, not necessarily in every cell).
2. The pol γ enzyme is sensitive to antiviral medications (such as AZT or Zidovudine) and patients taking these can develop an induced mitochondrial toxicity.
3. The POLG gene is frequently mutated in many mitochondrial diseases and is considered a major contributor to pediatric and adult onset mitochondrial diseases.

The following is further explanation of the three ways above.
1. Role of pol γ in making mutations.
“IF SOMEONE HAS A POLG MUTATION, ISN’T THAT GOOD ENOUGH TO DIAGNOSIS THEM WITH A MITOCHONDRIAL DISEASE WITHOUT FURTHER TESTS, SUCH AS A BIOPSY?”

Dori and her team cross the finish line at the 8th annual Run Wild for a Cure Walk/Run held in Cleveland, OH. The event raised over $170,000!!

See pages 4–7 for more events and fundraisers!
lack a DNA repair system, called mismatch DNA repair, that corrects errors made by normal DNA replication. In the nucleus, a defect in mismatch repair is responsible for colon cancer. So why can mitochondria get by without a mismatch repair system? Two reasons, 1) because of the high accuracy of pol \( \gamma \), and 2) the high copy number of mtDNA which allows a certain tolerance of DNA mutations. Despite this, mtDNA mutations are made and passed on from mother to child and sometime this can result in disease.

2. Pol \( \gamma \) Sensitivity to antiviral drugs

The pol \( \gamma \) enzyme is particularly sensitive to antiviral drugs used to treat HIV infection. This means that some of the people being treated by antiviral therapy may develop an induced mitochondrial disease. Usually this type of toxicity is reversible, but such a toxicity means that the physicians can’t use a particular drug to treat HIV infection, which presents a problem for antiviral therapy.

3. Mutation in the POLG gene

Mutations in the POLG gene can also cause a mitochondrial disease directly. The first disease mutation was discovered in 2001 by a Belgium research team who found mutations in POLG associated with progressive external ophthalmoplegia, an eye muscle disease that causes paralysis of the external eye muscles. Since that time, clinicians and researchers have been finding POLG mutations to be the genetic cause of a number of mitochondrial diseases including early childhood mtDNA depletion disorders such as Alpers and myocerebrohepatopathy, and mtDNA deletion disorders such as ataxia-neuropathy, MELAS, CMT, and dominant and recessive forms of progressive external ophthalmoplegia. POLG mutations have also been implicated in male infertility, parkinsonism and Leigh’s syndrome. The age of presentation varies along with the symptoms and can present anytime from birth to old age. Mutations in POLG are the most frequent genetic cause of childhood mtDNA depletion disorders.

It has been estimated that nearly 2% of the general population are carriers for a pathogenic (disease-causing) POLG mutation, with the combined presentation of POLG-related disorders from both recessive and dominant mutations to be ~1:10,000. To put this in perspective, if the world population is 6,707,000,000, that means that approximately ~134 million people are carriers of pathogenic POLG mutations and ~670,000 people possibly have a mitochondrial disease as a result of POLG gene mutations.

So what’s our role in all of this? The Mitochondrial DNA Replication group at the NIEHS was the first to discover the human POLG gene in the mid-90s and we have been studying the biochemistry, enzymology, antiviral drug sensitivity, fidelity and consequence of disease mutations ever since. Several years ago we developed a web site database to keep track of all the POLG disease mutations and this has become rather useful to clinicians and researchers. In my biased view, I believe POLG is one of the most important proteins/genes to study mitochondrial disease, since it is responsible for making the majority of mtDNA mutations. And that’s not even considering mutations in the POLG itself.

Question: Is it correct, then, to say that a POLG mutation will create a “secondary” mutation in the mtDNA during the “copying” process? And in addition, even if the POLG protein/gene itself is not mutated, it can still sometimes “mess up” in duplicating the mtDNA, thus creating a mutation?

That is correct. The natural mutation rate for pol \( \gamma \) is one mistake (mutation) for every 500,000 to 1,000,000 DNA bases copied (give or take a few). DNA repair process will then correct most of the mistakes made by DNA polymerases. So, that’s much less than one mutation per mtDNA copy, or about one mutation in every 30 copies. A typical human cell contains anywhere from 1000 to 10,000 copies of mtDNA.

Question: If someone has a POLG mutation, isn’t that good enough to diagnosis them with a mitochondrial disease without further tests, such as a biopsy?

It depends on the mutation. We can certainly predict the outcome of patients with well documented mutations, but many mutations are uncharacterized. Not every mutation in POLG is pathogenic. Most genes carry normal variations that don’t have any outcome and these are usually classified as single nucleotide polymorphisms. They represent mutations that occur in the normal/healthy population at high frequency and don’t usually cause harm. So, since there are so many mutations in POLG, we are not always sure if a particular set of new mutations is pathogenic. That’s why we are doing the research and have also set up our website POLG mutations database (http://tools.niehs.nih.gov/polg/) to help the community in educating them about what is known about each mutation. In short, the knowledge of a POLG mutation is sometimes sufficient, sometime not, to diagnosis an individual.
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.

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THE QUESTION IS...
I have a 24 y/o son with GAI and complex III deficiency. He has been in crisis for a year now. He gets D10 frequently and his ammonia levels stabilize and he comes home. He is only stable for approximately 48 hours then they rise again (198) this week and he is readmitted to the hospital. Can we tell if high ammonia is from GAI or mitochondrial complex III?

RESPONSE FROM GERARD BERRY, MD:
The disorder, GA II, is most certainly linked with hyperammonemia. And the levels of plasma ammonia are more likely to rise in the patient with GA II when the products of fatty acid and amino acid metabolism accumulate in mitochondria as chemical compounds which contain coenzyme A, such as glutaryl-coenzyme A. This usually occurs when the patient has fasted, has ingested too much dietary long-chain fat and/or protein or even just with severe stress. The mitochondrial respiratory chain defects is linked with hyperammonemia deficiency not usually associated with hyperammonemia but it can happen. For example, it may appear in a patient with a Complex III defect when it involves the liver. Under these circumstances, it would be important to find out if the cells in the liver are very sick and undergoing deterioration. For example, it is possible that a respiratory chain defect in the liver may lead to liver failure or cirrhosis. Your physician will be monitoring this problem perhaps with the help of a gastroenterologist. Finally, it is possible that the rare occurrence of both GA II and Complex III deficiencies in liver may change the nature of hyperammonemia due to GA II deficiency, or, more simply, how it tends to appear and then stabilize. In other words, the ammonia might rise even in the absence of severe fasting, dietary lapses and stress. But the encouraging finding in your son is that carbohydrate administration as IV D10 seems to bring the levels back to normal. My suggestion is to work with your physician to try to control the hyperammonemia with dietary cornstarch or other nutrients to augment daily carbohydrate intake, at least as a first step.

THE QUESTION IS...
I have a diagnosis of CPEO with possibilities of Kearns-Sayre Syndrome. Right now I only have eye movement issues. I recently had an incident where I was outside and went to run. I started to move; however both of my legs, especially my left, were reluctant to move and I almost fell. Is that possibly something to do with the Mito Disease?

RESPONSE FROM MICHIO HIRANO, MD:
Although chronic progressive external opthalmoplegia (CPEO) means weakness of eye muscles, limb and oropharyngeal muscles are also affected in many patients with CPEO. Therefore, your difficulty moving your legs may be due to limb muscle weakness, which typically affects proximal (i.e. hip) more than distal (i.e. ankle) muscles. Alternatively, the difficulty moving your legs could be due to cerebellar ataxia (incoordination due to dysfunction of the lower posterior portion of the brain), which is a common feature of Kearns-Sayre Syndrome. Both ataxia and limb muscle weakness could be present. A careful neurological examination by your internist or neurologist may be useful to pinpoint the cause of your trouble moving your legs.

THE QUESTION IS...
Has anybody ever considered alopecia (areata, totalis, and universalis) a mitochondrial disease? I’ve looked for some medical articles but was unable to find any.

RESPONSE FROM SUMIT PARIKH, MD:
This is a very interesting and unique question. Mitochondrial diseases have been associated with changes in hair formation-texture-pattern in some patients - including alopecia, brittle hair, pili torti (kinky hair), hypertrichosis (excess hair), and madarosis (loss of eyelashes). We do not know if these findings are cause-and-effect or simply associations noted by astute clinicians. In addition to this, we know that the mitochondrially-mediated cell death plays a role in chemotherapy leading to hair loss, though specific details are still being researched.

As far as alopecia areata goes, there is no known connection yet to the mitochondria. However, a study in 2002 (J of Dermatologic Science) did find higher than normal levels of antioxidants in scalps of individuals with alopecia areata, along with an increase in reactive oxygen species (ROS). Coenzyme Q, and other supplements are partly used in mitochondrial disease to decrease the burden of ROS that may increase in these conditions. It is plausible that this same mechanism may benefit the individual with alopecia areata if ROS are truly increased in this disease. However, such a theory would not answer the question of whether or not alopecia areata and mitochondrial diseases are connected.

THE QUESTION IS...
My daughter had a muscle biopsy that revealed the following: “Myopathy, with rare necrotic fibers; some fibers show mitochondrial proliferation (ragged blue fibers). Comments: This is an abnormal muscle biopsy which demonstrates mild necrotizing myopathy with no inflammation or fibrosis. Rare fibers with mitochondrial alteration in the form of mitochondrial proliferation are seen.” A frozen piece was then sent off to determine the type of Mito but it was normal. What does that mean? Is it common for the frozen piece to be normal?

RESPONSE FROM GREG ENNS, MD:
The types of testing that can be performed on frozen compared to fresh muscle biopsy samples are different. In general, more detailed types of studies can be done on a fresh sample. If mitochondrial respiratory chain analysis was performed on the frozen specimen, it is not uncommon for results of such testing to be “normal”, even in the presence of mitochondrial disease. If a definite abnormality is identified on respiratory chain analysis, this may lead to a more precise diagnosis. For example, specific DNA testing may yield an answer in such a case. Without knowing more about your daughter’s clinical presentation and other test findings it is hard to comment further. However, it is always a good idea to be sure she is seen by a specialist who cares for children with mitochondrial disorders.

ASK THE MITO DOC℠ IS A BENEFIT OF THE UMDF “ENERGY” MEMBERSHIP. IF YOU ARE A MEMBER AND WOULD LIKE TO SUBMIT A QUESTION, LOG IN TO THE UMDF WEBSITE USING YOUR USER ID AND PASSWORD. IF YOU WOULD LIKE MORE INFORMATION ON BECOMING A MEMBER OF THE UMDF, EMAIL info@umdf.org.
**CHAP TER ACTIVITIES**

**ATLANTA CHAPTER**
- **April 14, 2009.** The Chili’s at North Point in Alpharetta, GA held its second “Chili’s Gives Back for Mito” night. For every diner that presented their server with a UMDF voucher, Chili’s contributed 10% of their total bill to the UMDF.

**CALIFORNIA CHAPTER**
- **March 7, 2009.** The fourth annual Dinner honoring Brittany Wilkinson was held at Break the Barriers in Fresno, CA. The event featured auctions, raffles, and entertainment. Thank you to all of those who made the night a success.
- **May 17, 2009.** KCAL-FM and Corona Harley Davidson hosted a Poker Run in honor of Taryn Fogel. Proceeds from the event benefited the UMDF through the Taryn Fogel Research Fund.

**CAROLINA FOOTHILLS CHAPTER**
- **April 24-25, 2009.** The fourth annual Caroline’s on My Mind Weekend was held in Spartanburg, SC. Events included a walk/run, golf outing and barbecue/band party. Proceeds from the event benefited the UMDF through the Caroline Virginia Pulliam Mitochondrial Disease Research Fund.

**DELAWARE VALLEY CHAPTER**
- **April 5, 2009.** The Philadelphia Phantoms of the American Hockey League hosted a mitochondrial disease awareness night at their game versus the Norfolk Admirals. Many thanks to all of the Delaware Valley individuals and families who attended. A portion of the ticket sales benefited the UMDF.

**DC/BALTIMORE CHAPTER**
- **May 31, 2009.** Chesapeake Physical & Aquatic Therapy hosted its 6th Annual Bull and Shrimp Roast to benefit the UMDF and the Kennedy Krieger Institute. Guests enjoyed the dinner, auctions and an inspirational speech by Former Baltimore Colt Joe Ehrmann. Thank you for your continued support!

**KANSAS CITY CHAPTER**
- **March 13-14, 2009.** Members of the Kansas City Chapter participated in a give back-night with Buffalo Wild Wings in Johnson County, KS. All proceeds from drink orders were donated to the UMDF on these two days.
- **May 30, 2009.** The sixth annual Mito What? Walk and Family Fun Run was held in Lake Waukomis, MO. The event offered all participants a fun-filled experience with entertainment, refreshments, mitochondrial disease resources, and much, much more! Thank you to all of our participants, donors and race committee members.

**NEW ENGLAND CHAPTER**
- **March 8, 2009.** Bridget Willis hosted a silent auction and gift recycling in her home in honor of her son, Owen. The event raised more than $2000 for life-saving research. Thank you Bridget for your support and dedication!

**NEW YORK METRO CHAPTER**
- **April 25, 2009.** The inaugural OlliePalooza was held in Manhattan in honor of 4-year old Oliver Scheier. The silent auction raised more than $70,000 for the research of mitochondrial disorders. Thank you to all of the participants and donors who made the evening a success.

**OHIO CHAPTER**
- **March 21, 2009.** Elite Fitness in Solon, OH hosted a Spin-a-Thon in which teams of spinners pedaled for four hours in support of Bobby Arnold. The event raised more than $2500 credited to Team Bobby at the Ohio Chapter’s Run Wild for a Cure.
- **April 3, 2009.** The Sixth Annual Guest Bartender Night in honor of Kyle Kobunski to benefit the UMDF was held at the 82nd Street Grill and Pub in North Royalton, Ohio. Tips earned by the guest bartenders were donated to Run Wild for a Cure in support of Team Kobunski.
- **May 9, 2009.** Now in its eighth year, Run Wild for a Cure raised nearly $170,000 for the UMDF at the Cleveland Metroparks Zoo. Participants enjoyed a 5K run, 1-mile walk, games, prizes and much more! Plans are already under way for next year’s race.
- **May 17, 2009.** The Twinsburg Fire Department Local 3630 hosted an All-You-Can-Eat Pancake Breakfast to benefit the UMDF and Relay for Life. Thank you to everyone who came out to enjoy the meal.
UMDF AMBASSADOR PROGRAM REVISED

The United Mitochondrial Disease Foundation Ambassador Program has been revised. Starting on July 1, UMDF Ambassadors will need to renew their commitment annually. Ambassadors will be sent a new Ambassador Guides with helpful tips and ideas for activities they can do as well as a more detailed explanation of what is expected of an Ambassador. We are now asking that Ambassadors complete a minimum of two activities per year. These can be any combination of fundraisers, physician education, awareness activities, advocacy, and compiling a resource guide.

An Ambassador is a local liaison in a community that does not have enough UMDF members for the formation of a mito group or chapter. They respond to inquiries made by affected individuals and interested parties and provide them with information regarding doctors and other resources. Offering a sympathetic ear for people seeking support and connecting them with the national UMDF office is a major part of the position. If there is not currently a UMDF representative in your area and you are interested in becoming an Ambassador, contact us at info@umdf.org.

OTHER NOTABLE EVENTS

• March 20-21, 2009. Preston’s March for Energy was held in Virginia Beach, Virginia in conjunction with the Shamrock Sportsfest marathon. Building on past successes, the march raised more than $40,000 for the UMDF in honor of Preston Buenaga.

• March 27, 2009. Jude’s Fais Do-Do for Mito was held at the American Legion in Sulphur, LA. The Cajun-style dance gala in honor of Jude Manley featured a silent auction and raised more than $30,000 for the UMDF.

• April 11, 2009. The St. Louis Mito Group held their sixth annual Mito What? Run and Family Fun Day at the Our Lady of the Snows in Belleville, IL. You can view race results and photos at www.umdf.org/stlouisirace. Thank you to all of our supporters who helped to raise more than $10,000 for research and awareness.

• April 18, 2009. The first ever Mito Mania dinner raised over $18,000 toward research for a cure. The event was held in Cincinnati, OH at St. Antoninus Church and featured auction items, basket raffles and a 50/50.

• April 25, 2009. The fifth annual Race for Riley raised more than $1400 for the UMDF through a silent auction. The event was held in honor of Riley Peek of Atlanta, GA. Many thanks to all of the donors and participants.

• May 9, 2009. Matthew Dudgeon Memorial Fund held its fifth annual 7K Run and 9th Annual Walk and Dinner Dance. Partial proceeds from this day of fun benefited the UMDF.

• May 30, 2009. The fourth annual Golf Tournament honoring Brittany Wilkinson and benefiting the UMDF was held at the Madera Golf Course in Madera, California. Many thanks to all of the golfers and sponsors for a wonderful day on the links!
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A triathlon is a multi-sport endurance event consisting of swimming, cycling and running in immediate succession over various distances. A triathlete is one who competes for the fastest overall course completion time, including timed transitions between the individual swim, bike and run components. The life of a triathlete is intense, training seven days a week, multiple hours a day.

Sherri McLauchlin is a triathlete. Sherri McLauchlin has a mitochondrial disease.

For many years, Sherri had problems with her kidneys, chronic constipation, suffered from migraines, constant fatigue and muscle cramps. She saw specialists for each symptom and they could not find an obvious cause after numerous tests and procedures. It was not until Sherri started to compete in the triathlons, and had muscle cramps so severe that she knew something was seriously wrong. The muscle cramping would start just twenty minutes into her training and many times would stop her dead in her tracks.

Sherri recalls the start of a viscous cycle, “I would cramp at night, get no sleep, be tired all day, train for the triathlon and get even more cramping.” After many tests and constant bugging, Sherri’s primary care doctor sent her to a neurologist. More tests were done including a muscle biopsy; while there were some abnormalities, they still could not figure out what was wrong.

That is when Sherri set out on her own, medical files in hand; she sought out a doctor who specialized in muscles and found Dr. Allan Weiss of St. Petersburg, Florida. After meeting with Dr. Weiss for 15 minutes, he had an idea of what was wrong and ordered another muscle biopsy. The results came back in a few weeks with a diagnosis of Mitochondrial Myopathy. “What the heck is that and how can we fix it,” was the first thing that Sherri remembers saying. She was told that she would have to slow down and stop participating in triathlons. That was not something that Sherri was ready to give up. “If I can finish, I am doing them,” was what she told her doctors. “I was not about to give up on something I loved, even if I was weaker and more tired after each event.”

It was during training with an open water swim in the Gulf of Mexico that Sherri finally understood what her doctors had been telling her. She was swimming with a group of swimmers when she started to have leg cramps. “I knew I had to head to shore,” Sherri explains. “I made it to the sandbar and tried to work the cramps out.” She then decided to try to make it to the shore, without realizing how deep the water would get again. “I then began to cramp from the waist down and could barely swim at all,” she said. “I then began to yell from my friends who had passed me up in the water.” Her swimming companions came to her rescue and pulled her to safety. Sherri finished her final triathlon a week later and retired from doing them.

Sherri still exercises six days a week, but not at the extent she was used to doing. “It is very frustrating to have a desire to be very active but my body can not keep up.” Sherri says. “I guess I am addicted to exercise but sometimes it is not good for me.” She still swims, bikes, and runs, just not all together. “I have to wear a swim belt” her swim belt will inflate when she can not swim, “and can only swim in our pool.” Sherri has a wonderful support team consisting of her husband, two daughters and her brother. “They have always been there cheering me on.” Sherri’s husband and brother would be at each event in their bright orange shirts so that she could spot them in the crowds of people. “I could always hear my husband cheering, even when I was in the water swimming.”
This is a very busy and exciting time of the year for members of the United Mitochondrial Disease Foundation because our annual symposium is about to get underway in Washington D.C. On June 24, 2009, Mitochondrial Medicine: 2009 Capitol Hill will kick off at the Sheraton in Tyson’s Corner with the arrival of more than 250 members of the scientific and medical community.

This is the eleventh annual conference organized by UMDF which brings together clinical and basic science researchers that share an interest in mitochondria. Participants come from many fields, including biochemistry, genetics, neurosciences, cardiology, cancer, diabetes, nephrology, hematology, pediatrics, and aging research. The meeting is considered to be the largest gatherings in the nation of members of the scientific and medical community who dedicate their lives to mitochondrial medicine and research.

While we are still getting registrations, we are pleased to tell you that the numbers of those attending from the scientific and medical community are up significantly from last year.

On June 25, 2009, our family meetings begin with a host of valuable informational sessions. There will be sessions that specifically address the needs of adults, teens and children. As always, our popular session, “Ask the Mito Doc,” will be held this year. One of the very unique things about our symposium is that we mix doctors, researchers and patients all together at one event. This mixing allows each group to gain insights that would not be otherwise possible at just a research event. It is also at our symposium that we will announce the 2009 UMDF Grant Award Recipients, as well as the winners of our LEAP and Heartstrings Award and the Stanley A. Davis Award.

Sandwiched in between the scientific and family sessions is our first “Day on the Hill”. More than 250 people have registered to participate in our advocacy effort. By the busload, we will travel to Capitol Hill for meetings that have been scheduled with members of Congress and the U.S. Senate. It is an opportunity for our community to “be part of the cure” as we educate elected officials about mitochondrial disease and ask for their support for additional research funding. For those who missed the sign up deadline or just can’t make it to Washington to help us this year, watch your email. We will have a way for you to participate in advocacy from home.

This will be the tenth symposium that I have attended. I have had fun and increased my knowledge at each and every event. I have also made some great friends. If you are registered to attend, we look forward to seeing you again. But if you are unable to make it, please check the website periodically for updates, pictures and news from Mitochondrial Medicine 2009: Capitol Hill.

Energy to all,

W. Dan Wright, UMDF Chairman

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**2009 UMDF RESEARCH GRANT RECIPIENTS**

- **$130,000**  
  Christoph Handschin, PhD, University of Basel, Switzerland  
  *Project title: “Mitochondrial dysfunction, exercise intolerance and myopathy in skeletal muscle-specific PGC-1α-deficient mice.”*  
  Dr. Handschin will investigate a gene that controls the formation and activity of mitochondria in skeletal muscle. This is important, because mice that possess a mutated form of this gene experience muscle dysfunction similar to that associated with mitochondrial diseases in humans.

- **$110,000**  
  Michael P. Murphy, PhD, Medical Research Council, Dunn Human Nutrition Unit, Cambridge, U.K.  
  *Project title: “Development of a Novel Mass Spectrometric Approach to Measure Mitochondrial Oxidative Damage In Vivo.”*  
  Dr. Murphy will develop a method for measuring in living organisms the extent to which their mitochondria have been damaged by oxygen free radicals. This is significant because it could ultimately be used to monitor ongoing changes in the function of mitochondria of mitochondrial disease patients and aid in assessing the effectiveness of potential therapies.

- **$81,857**  
  Patrick H. O'Farrell, PhD, University of California-San Francisco  
  *Project title: “Selecting for Transformation with Mitochondrial DNA”*  
  Dr. O’Farrell will develop a method for introducing DNA into the mitochondria of fruit flies. This important research will produce reliable animal models for the study of a variety of mitochondrial diseases and could also help guide attempts to repair the mitochondrial genome in humans.

- **$90,804**  
  Sarika Srivastava, PhD, Harvard Medical School  
  *Project title: “Investigating the Rescue of Mitochondrial Dysfunction by SIRT1 and Calorie Restriction.”*  
  Dr. Srivastava will study the activity of an important regulator of mitochondrial energy metabolism in mice. This is significant because finding ways to enhance the activity of this gene could point to therapies for increasing energy production by mitochondria.
Charitable giving and planning for you and your family’s future may not be uppermost on your mind as you face the day-to-day worries in these tough economic times. But they should be.

That’s why the United Mitochondrial Disease Foundation wants to remind you of its “Taking Control” initiative designed to help our members and donors develop, coordinate, and implement the necessary planning, strategies, and financial instruments to ensure that your financial goals are met.

Nick Nicholson, a senior financial advisor with The Monteverde Group, is available to provide free, no obligation, expert assistance coupled with complete confidentiality to all UMDF members and potential donors. The UMDF has been a long-time partner with Nick and The Monteverde Group. There is no better time to discover how planned giving can be BOTH an integral part of your family’s financial planning and tax management as well as an effective way to support the UMDF and its mission.

For a free consultation, all you need to do is contact Don Gielas in the UMDF Development Office at 888-317-UMDF or email don@umdf.org. He will have Nick contact you to discuss any financial issues dealing with charitable donations and tax management. Even if you currently have a financial advisor, Nick can provide a second opinion on your financial situation just to ensure your financial comfort and stability. You have nothing to lose because the service is free with no obligation.

As you think about evaluating your financial situation with Nick, consider these 13 important ways that charitable giving through a planned gift can help you and the UMDF:

1. **Give yourself a tax deduction** by giving a charitable contribution
2. **Give yourself an income stream** through a gift annuity that can provide a generous, non-fluctuating stream of income—for life!
3. **Give your children a college education** through planned giving techniques that can fund your child’s education with tax advantaged accumulation.
4. **Give your heirs a larger inheritance** through charitable giving options that offer many effective options for tax effective estate planning, including removing tax liability.
5. **Give yourself a retirement plan** through a charitable remainder trust that is tax exempt, funded by contributions that are partially tax deductible, and even provides life insurance that can make the plan self-completing.
6. **Give yourself a business exit strategy** and help your family, partners, or employees plan for business succession. Planned giving techniques can help eliminate large taxes on capital gains or reduce estate taxes that challenge business transfers to the next generation.
7. **Give yourself increased cash flow** because charitable giving provides tax deductions and can diminish taxes on capital gains of highly appreciated assets.
8. **Give your children protection** with a properly drafted trust to protect their inheritance or preserve their benefits eligibility with a special needs trust.
9. **Give your investment portfolio a boost** with a charitable remainder trust that will help you better manage your investments without tax concerns.
10. **Give a memorial gift** on behalf of a loved one or friend that will acknowledge their memory and help UMDF continue the fight towards a cure.
11. **Give your heirs a purpose and a mission** in life by setting up a private foundation, a supporting organization or donor-advised funds that help them appreciate the value of money and exhibit compassion for others by participating in grant-making to UMDF and other important causes.
12. **Give yourself control** by simply sending your tax dollars to the government for them to control or taking charge by using charitable devices to support the causes and efforts dear to you…like UMDF.
13. **Give hope, energy and life** by using charitable giving techniques to make tax deductible contributions to UMDF to advance research, educational initiatives and support for affected individuals and their families.

Call UMDF today and take control of your finances and future.

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Four year old Jude Manley, diagnosed with a mitochondrial disease
While we would like to list ALL of our donors individually, please understand that it would be very difficult to do so with 5,906 donors of $1-$99 (THANK YOU!), 1,632 donors of $100-$249 (THANK YOU!) and 341 donors of $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 January 1, 2009, and June 30, 2009. If your name or company’s name is incorrect or not listed, please contact info@umdf.org.

BENEFACTOR $10,000+
Corporations, Organizations and Foundations:
Bio Enhance Technologies Inc.
Josie Mazoo Children's Charities
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Appreciating those who have helped along the way!

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The Zachary L. Friedman Foundation

Individuals:
Jim & Wendy Abrams
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- Brian Zweig
- Rick Zystra
UPCOMING EVENTS

ATLANTA CHAPTER
• **September 18, 2009.** The Atlanta Braves will host a mitochondrial disease awareness night at Turner Field. A portion of the tickets sales will be donated to the UMDF.
• **September 26, 2009.** The third annual Music for Megan in Alpharetta, GA will feature Atlanta’s favorite “high-energy, hard-kicking” band - Banks & Shane! Children will enjoy pony rides, inflatables, a rock climbing wall and much, much more! Buy your tickets today at www.musicformegan.com.

• **August 15, 2009.** Baylee Bringin’ the Bands, in honor of Baylee Thompson, will feature the area’s best talent in Cannelville, OH. The event will benefit the UMDF.
• **September 21, 2009.** The fourth annual KWGP Charity Golf Outing will be at the Worthington Hills Country Club in honor of Ellie Kovalcik. Register your foursome today at www.umdf.org/kgwpgolf.

CHICAGO CHAPTER
• **September 20, 2009.** Join hundreds of individuals and families at the third annual Chicago Walk and Family Fun Day at Pottawatomie Park in St. Charles, IL. Register at www.umdf.org/chicagowalk to enjoy a day of fun in support of funding for mitochondrial disease research.

DELWARE VALLEY CHAPTER
• **September 12, 2009.** Come celebrate a decade of success at the 10th annual Go for Mito Walk/Run at the Philadelphia Museum of Art. Join a team, start one of your own, or register as an individual at www.umdf.org/goformito.

NEW ENGLAND CHAPTER
• **October 10, 2009.** The second annual Mito Walk and Family Fun Day will be held at the LEGO Campus in Enfield, CT. Register at www.mitowhat.org to enjoy a day of family-friendly fun! Come to view many of the intricate designs by LEGO's master builders and try your hand at a model of your own.

MINNEAPOLIS-ST. PAUL CHAPTER
• **September 19, 2009.** The fourth annual Mito 5K Walk/Run will be held on at Thomas Lake Park in Eagan, MN. The walk raised more than $17,000 last year. Help us build on that success by registering today at www.umdf.org/minnesotarace.

PITTSBURGH MITO GROUP
• **August 2, 2009.** The Beauty First Salon in McIntyre Square is holding a Cut-a-Thon in honor of Aubrey Austin. A portion of the proceeds from each salon service will be donated to the UMDF.
• **August 15, 2009.** Now in its third year, Brew at the Zoo promises to be bigger and better than ever. Buy your tickets at www.TheZooBrew.com, then come to the Pittsburgh Zoo & PPG Aquarium to sample beers from more than 50 local, national and home brewers.

• **August 27, 2009.** The Pittsburgh-based restaurant, Casbah, a member of the innovative Big Burrito Group, will host a special dining event to benefit the UMDF. Casbah donates 100 percent of proceeds to the UMDF from this special dinner. We are limited to 50 seats, so make your reservation today! Visit www.umdf.org/casbah today.
• **September 26, 2009.** The third annual Sip and Savor the Moment for a Cure is a wine and chocolate tasting gala in honor of Hannah Pallas. Buy your tickets at www.SipSavorCure.com, and then come to the La Casa Narcisi Winery in Gibsonia, PA for an evening of auctions items and great fun!

AROUND THE COUNTRY
• **August 1, 2009.** The second annual Run 4 Raley is a great family-friendly activity held in Philo, IL. Gather your friends, family and co-workers to start a walking team at www.umdf.org/run4raley. There are no special skills needed to participate; simply come out and enjoy the day while supporting those affected by mitochondrial disease.
• **September 13, 2009.** The Amanda’s Journey Remembrance Walk will be held at the Crossings of Colonie in Loudonville, NY. The event is held in memory of Amanda Perrotta and benefits the UMDF, as well as the Albany Medical Center.
**GIFTS FROM THE HEART**

- The family of Caden Jarvis held a Coins for a Cure campaign in Schaghticoke, NY and raised $370 in his honor.
- Cathy Osland, of Cumming, GA held a letter-writing campaign in honor of their granddaughter, Danielle Magerfleisch. In total, Cathy raised nearly $400.
- Michelle McGuire organized a live music night in Alpharetta, GA. The event showcased local talent and raised $500 for the UMDF.
- Jackson Culley celebrated his 2nd birthday on March 21st. His family held a birthday party celebration and in lieu of gifts, donated $500 to the UMDF in honor of Jackson. Thank you to the Culley family for your support and Happy Birthday Jackson!
- In lieu of gifts, Mindy Lopez asked for donations for her 11th birthday. Mindy donated $170 to the UMDF in honor of Christopher Santos.
- Kivian Johnson held a bake sale to raise awareness of mitochondrial disease. Kivian was able raise $85 to be donated to the UMDF. Thank you Kivian for your hard work!
- Jennifer Cook held a Coins for a Cure campaign at the JC Penney Salon in Rock Springs, WY. More than $350 was donated to the UMDF in honor of Logan Palmer.
- Cloverdale Elementary School held a Jump Rope for Mito Event in honor of Andrew. Students from grades K-2 collected pledges and donations for the UMDF.
- The Shiloh School in Shiloh, IL conducted a Pennies for Jenny campaign in honor of Jenny Schnitzler. In total, the Shiloh School collected more than $2,600 for the UMDF.
- The Spring Ford Area School District held a “Dress Down Day.” Students and faculty raised more than $300 for research toward a cure.

**MEMBER RESOURCES**

**Band-Aides and Blackboards**

Band-Aides and Blackboards is a site for children and teens about growing up with medical problems. The site is designed and arranged by Joan Fleitas, EdD, RN, of Lehman College. Band-Aides and Blackboards is broken into three interactive sections for kids, teens, and adults (including teachers.) To see what Band-Aides and Blackboards has to offer, visit http://www.lehman.cuny.edu/faculty/jfleitas/bandaides/.

**Eli’s Angels**

Eli’s Angels is a non-profit organization that sends gifts to children, 10 years of age or younger, who are affected with Leigh’s Disease, a mitochondrial disorder, or a metabolic disorder. Eli’s Angels will send these gifts to children who live within the United States (excluding Hawaii and Alaska.) You can register your child online at http://home.comcast.net/~elis_angels/. For more information, contact Eli’s Angels at 1-800-497-5219 or email ekurtz03@comcast.net.

**Hugs and Hope for Sick Children**

Hugs and Hope is a non-profit organization that recruits volunteers to send “happy mail” to sick children to brighten their day. You can volunteer to send cheery cards to these children or you can register your child to receive “happy mail” from their volunteers. To learn more about Hugs and Hope or to sign up, visit http://www.hugsandhope.org or email ejupdates@yahoo.com.

**Songs of Love**

The Songs of Love Foundation is a nonprofit organization that provides personalized songs to children and teens that are facing medical, physical, and emotional challenges. The foundation produces these personalized songs, that are never duplicated, for your child FREE of charge! A patient’s parent, legal guardian, or hospital staff member can request a song. Requesting a song is very easy to do! You can request a profile sheet by calling the foundation at 1-800-960-SONG(7664). You can also visit www.songsoflove.org to fill out the form online or you could download and print the profile sheet.
DAY ON THE HILL
On Thursday, June 25, 2009, the United Mitochondrial Disease Foundation will hold its first Day on the Hill advocacy effort. More than 250 people have signed up to be part of our advocacy action day. It is a unique opportunity for those who are participating to educate our elected officials about mitochondrial disease and the effect it has on individuals and their family. We will also have a specific request for our elected officials.

The Day on the Hill will begin early on June 24, 2009. Those who confirmed that they are participating will be bussed to Capitol Hill where they will meet with their representatives in the United States Congress and Senate. Currently, meetings are scheduled with 89 members of Congress and 61 members of the Senate.

The meetings follow a year long advocacy program in which UMDF members, family and friends were encouraged to sign up and utilize the ‘Advocacy Action Center’ on the UMDF website. The ‘Advocacy Action Center’ allowed those who signed up the ability to send a message to their elected official supporting a letter that was signed by more than 70 members of the scientific and medical community to President Barack Obama. More than 3500 letters were sent from the UMDF action center to representatives in 48 of the 50 states.

The largest contingent of UMDF members participating in the Day on the Hill are coming from the state of Georgia where 31 people registered for the event. We also have representation from California, Washington, Oregon, Arizona, Texas, Oklahoma, Kansas, Minnesota, Iowa, Wisconsin, Illinois, Indiana, Ohio, Pennsylvania, Michigan, New York, Massachusetts, Maine, New Hampshire, Connecticut, Maryland, Virginia, Louisiana, Mississippi, South Carolina, Florida and Tennessee.

While the deadline to participate has passed, the UMDF will be offering a way for those who are not attending to participate. We will be calling on you to support the efforts of those who are advocating in Washington, DC on June 24th by sending an email and making phone calls to elected officials, so watch for an email from the UMDF with instructions.

MITOCHONDRIAL DISEASE AWARENESS WEEK

The third week of September (20th–26th) will be Mitochondrial Disease Awareness Week! We need your help to designate an awareness week in your state! Now is the time to start contacting your local Senator or Representative. The UMDF has created an Awareness Week Toolkit (available at www.umdf.org) to help you along the way! Sample letters, a sample bill template and letter writing tips, the toolkit has everything you need to get started. Along with contacting your senator and representative, we also urge you to hold an awareness week activity. Let the UMDF know your plans and we will be listing all Mitochondrial Disease Awareness Week activities in our next newsletter (July 31, 2009 deadline.)

We are excited to add two states to the growing list of permanent proclamations designating the third week of September “Mitochondrial Disease Awareness Week.” In March of 2009, the state of Tennessee became the sixth state with a permanent resolution. Thanks to the hard work and dedication of the Middle Tennessee Chapter, the East Tennessee Mito Group and the Memphis Area Mito Group, Tennessee will now recognize Mitochondrial Disease Awareness Week every year. We applaud you in your great success!

Through the efforts of the UMDF California Chapter, the state of California became the seventh state with a permanent resolution. ACR25 was introduced by Assemblyman Jeff Miller and approved by the state legislature in April. This permanent resolution was made possible by the hard work and effort of UMDF Ambassador EJ Fogel of Corona, CA, and Youth Ambassador Brittany Wilkinson of Clovis, CA.

California and Tennessee join the ranks of Alabama, Georgia, Michigan, New Jersey, and Wisconsin in holding permanent resolutions. The state of Ohio has declared a one-time resolution for 2009. Copies of all of these resolutions are available at www.umdf.org. Just click on ‘Find a Local UMDF Chapter, Group or Ambassador’ and click on each state to view the resolution. If you know of a resolution that has passed in your state, please let us know!
My name is Gail Wehling and I was born and raised in Aurora, Illinois. I am the proud fourth child of Dolores and Thomas Wehling, a former prominent and long-time local business owner. In the fall of 1977, neither I nor my family could possibly predict how suddenly and unexpectedly my young life was about to change, forever.

As I was preparing to begin my junior year of high school, I was looking at my new school ID card and noticed that my eyelids looked droopy. My mom agreed and called a neighbor who, in turn, referred me to one of the leading ophthalmologists in not only the Chicagoland area, but the nation and world as well. Following my initial appointment began many long and difficult trips into Chicago over many days, weeks and months for extensive tests and examinations. After many exhausting months, I was diagnosed with Chronic Progressive External Ophthalmoplegia (CPEO). CPEO is a form of Kearns Sayre syndrome (KSS) that primarily affects my eyes and vision, but now affects my heart and all my muscles. As I have aged, the disease has also progressed. All parts of my eyes are now affected which limits my ability to function well on a daily basis. In addition, my heart and systemic muscle weakness, fatigue, and overall lack of energy (which my body cannot properly produce for it to function normally) limits what I can do every day.

Because of these new conditions and increased limitations, I recently moved back in with my parents and I now receive Social Security Disability (SSD). I am the first in my family to receive SSD and it was one of the hardest and most difficult decisions I had to make, but due to the progression of my disease, I simply cannot work anymore. The physical and financial toll has been difficult and painful. I used to be an independent and self-sustaining woman, but now this disease has taken most of that away from me.

However, despite the numerous surgeries, new symptoms and challenges, I can honestly say that this disease has been a positive influence in my life. It has given me three “gifts” - a greater appreciation, a deep gratitude, and clarity in my life. I have a greater appreciation for all that I have. I have a deep gratitude for everyone in my life beginning with my mom and dad, siblings, nieces and nephews, friends, pastor/parish and doctors. Simply, I would not be where I am today without their support, care and love.

And, this disease has given me clarity in my life I do not believe I would have if not for the disease.

Lastly, I am also very fortunate. Fortune to have recognized the problem; to have acted upon it; to have been referred to one of the leading ophthalmologists who happened to be local; to have received a “quick” diagnosis of a disease that is familiar and recognizable (in the ophthalmology field); to have all my doctors who continually problem-solve and give me outstanding care; and to have the United Mitochondrial Disease Foundation (UMDF) that provides all of us with mitochondrial disease with help and assistance, support, care and compassion.

The UMDF staff, and its associated doctors and researchers are dedicated and compassionate. They work hard every day to find cures and treatments for mitochondrial disease, and provide support to all affected individuals and families. I first came into contact with UMDF in 2000. Because they were so helpful and supportive of me, I was inspired to form the first Mitochondrial Support Group in the Chicago area in 2003. Currently, I am co-chair of the UMDF Chicago Area Chapter Support Committee and am also co-chair of the UMDF Adult Advisory Council (AACT) - the mission of which is to represent and serve the unique needs of the adult mitochondrial community.

Every day, I am deeply thankful and grateful to all in my life, especially the UMDF - their work and mission is now my life work as well.
UMDF MEMBERSHIP LEVELS
The United Mitochondrial Disease Foundation is excited to announce a new level of membership designed to help individuals promote education, awareness and support. The new membership level that will be added to the HOPE and ENERGY levels is our LIFE membership. The LIFE membership is a $250 two-year membership and will allow the member to help support a fellow member of the UMDF. Below are the additional benefits available to the LIFE member. (Please note: LIFE membership is a two-year membership.)

LIFE Membership ($250 - two-years)
All benefits included in the HOPE and ENERGY levels plus:
(go to www.umdf.org for a complete listing of HOPE and ENERGY level benefits)
- Hard Copy Resource Guide (upon request)
- 20% Discount on Merchandise
- Choice of UMDF Category Merchandise (subject to availability)
- Complimentary Set of Symposium Videos
- $100 of your LIFE membership will help support a family to attend the UMDF Symposium or a selected UMDF research project
- Enables you to provide a free, one year ENERGY Membership for the individual of your choice

To become a UMDF member and to learn more about the member benefits, visit www.umdf.org or call 888-317-8633.

SUBMISSION DEADLINE FOR VOLUME 14 ISSUE 3 IS JULY 31, 2009!