Please note that we ALWAYS accept donations from Families or Professionals, but remember they are not tax-deductible since we are not an official non-profit organization. If we are able to raise enough funds throughout 2004 then we will definitely resume our printed newsletter and updated Family/Professional Lists in 2005.

Thank you to Dr Barbara Marriage from Abbott Labs for sharing her research on mitochondrial disorders and the use of supplements. Please remember to sign up for the FOD Email List on our website — this is a great forum for ongoing discussion of such topics and questions dealing with practical day-to-day challenges of living with an FOD. Thank you also to our Families that shared their struggles and challenges with us in this issue by way of their stories.

We welcome ALL of your stories and pictures and we will try to either print them in the newsletter or place them on the Family Stories, Newborn Screening, or Love Messages page on our site. We would especially like to encourage families dealing with some of the less common FODs (i.e. HMG, SCHAD, Carnitine Acylcarnitine Translocase, TFP, CPT 1&2 etc.) to share their experiences. We’re also always looking for more low fat recipes, poems, and pictures. Be sure to check our website every now and then as we add new Stories or other special items.

Professionals ~ PLEASE let me know if you’d like to share your knowledge and expertise. We can always use more information and research articles or ongoing FOD studies on our website as well. Additionally, THANK YOU to all the Professionals (researchers, dieticians, counselors etc.) who returned the ‘Professional Questionnaire for FOD Referral Purposes.’ If you haven’t already please complete this one-page questionnaire on our website (Online Forms) so we can update our files, even if you are already listed on the printed Professional List.

The fight to SAVE NEWBORNS continues in the US and abroad in this New Year ~ no matter what disorder your Family is dealing with or which Groups you belong to, get out there and share your experiences to promote expanded newborn screening because...

‘We Are All in This Together!’

Take care...
DLG
June 2004 National Metabolic Conference Information

The National Coalition for PKU & Allied Disorders is pleased to announce we will be hosting our Metabolic Conference at:

Sheraton Detroit
Novi 21111 Haggerty Road
Novi, Michigan 48375
Ph: (248) 349-4000

The conference dates are Friday June 25 and Saturday June 26, 2004. Those traveling to the area should plan on a June 24 arrival and a June 27 departure (or a departure after 5:00 pm on June 26). An agenda of the meeting will be made available at a later date. A brief overview is as follows:

Thursday arrival - Hotel reception Thursday evening.

Friday 8am-5pm Individual conferences for: PKU, HCU, OA, Tyr, FOD

**FOD Speakers so far ~ FOD Speakers so far ~ Dr David Whiteman is our main speaker and will be speaking on "Untangling the Spirals of Metabolic Disease: Primary Diagnoses and Secondary Effects: Implications for Treatment"; Dr Mark Korson from Boston will address neurological and muscle issues; Dr Asamoah from Henry Ford Hospital in Detroit will possibly speak on GI issues (we may have a joint session with the OA Group since GI issues are relevant for them as well); Lynne Wolfe, Metabolic Nurse Practitioner will speak on "Metabolic Co-factors: Vitamins, Minerals and other Supplements"; and I am looking into having Elaina Jurecki, RD, from San Francisco or Dr Barbara Marriage from Abbott Labs speak on nutritional issues. We also may have a speaker very knowledgeable on Educational/Legal issues for Families, but she may speak at the Saturday session with all of our Groups. Dr Roe (our Medical Advisor) will be out of town during these dates so he will not be attending this Conference.**

Friday Evening: Newborn Screening Sessions 7-10 pm

Saturday Conference: PKU, HCU, OAs, Tyr, and FODs will be in one main function room attending presentations applicable to all.

A special session for Michigan families is also being planned. The purpose of the meeting is for Michigan metabolic families to join forces, and create a cohesive support group specifically for Michigan families. Anyone interested in participating in this particular workshop should contact Sandy LaPrad: smlaprad@yahoo.com

Airport: Detroit
Shuttle Transportation: No. We will provide you with the least expensive and easiest alternatives to the hotel from the airport asap. The hotel has contracts with certain companies for this service.

Room Rate: $89.00 per night
Avoid having to stay at an overflow hotel by reserving your room early. Let the hotel know that you will be attending the National Coalition for PKU & Allied Disorders Conference and you will be accommodated with the special rate.

Conference Registration: $50.00 per person. Registration forms will be mailed approximately 3 months prior to the meeting.

*US and Canadian FOD Families and Professionals will receive their registration Forms in the mail as a separate mailing (no printed newsletter in Jan due to low funds). Just so we don't have duplicate listings, if you are already on the FOD Mailing List for newsletters then DO NOT send your address to Trish. I am having Erika run off all our labels for the Registration Form mailings in @ March. If you decide to access the Registration Form on our website (once we get it from Trish) let me know and I can take you off the special March mailing ~ we're trying to conserve funds as much as possible. Overseas' Families/Professionals are welcome at the Conference as well, but we do not have the funds to mail Registration Forms so be sure and check our website.

Due to a number of reasons (too long to list) including liability, day care will NOT be available. Because of medical issues and for other reasons, the OAs will possibly be providing day care for their families. Please refer any questions regarding day care directly to OAA.

No day care will be provided for PKU, HCU, Tyr, or FOD.

Meals: Continental Breakfast and Lunch will be provided. Dinner is on your own.

We look forward to seeing you there!

Trish Mullaley
Mom to Julie & Jenn (cpku)
President, National Coalition for PKU & Allied Disorders
P. O. Box 1244
Mansfield, MA 02048
Toll Free (877) 996-2723
Coalition4PKUAD@aol.com
My husband William was frantically driving me to the hospital at 4 am. He wasn't just nervous because he was about to be a father. He was trying to get us to the hospital in one piece. Caleb Patrick was born at 5:20 pm during a really horrible tropical storm. My son blended into the world as the storm blasted by us. **I will always remember September 14, 2001. That is the day I became a first time mother. It was the happiest day of my life!**

My son was finally here. He made it through all of my infections, severe preeclampsia (I am waiting to find out if it was FLOP or HELPP) and having the umbilical cord around his neck twice. I was the proudest mother ever. **I had a fighter for a son.** I was only able to hold Caleb for a minute before he was taken to NICU. Caleb was placed in the NICU because he was born at 35 weeks and his blood sugar had to be monitored because I had Gestational Diabetes. I was so afraid to hold him for the first time. I was afraid that if I touched him that he would get sick. So all I did was lift up his blue knitted hat to see if he had hair. **I told him that "I love you and I will see you soon."**

I was on bed rest for the next two days so I wasn't able to visit Caleb in the nursery. The nurses did however sneak Caleb in for a thirty-minute visit. That was the only time I was really able to be Caleb's mommy. I didn't have any staff watching me or listening to what I was telling my son. During that visit I told Caleb all of my hopes for him. I looked at every inch of his body. And yes I counted his fingers and toes. They were all accounted for so I was happy and at peace. Every day William would come and visit us. I would sometimes get jealous. William would come in and say hi to me and give me a kiss then say, "I gotta go see my boy." Now, I am jealous because William had more time with our son. **I am thankful that Caleb received so much love from his father. Caleb knows that his parents love him so much and will never stop loving him.**

I was released on my fourth day. That is when I found out that Caleb wouldn't be coming home with us. I was then informed that he was being kept because he was a lazy po (by mouth) feeder. I wasn't even informed of this by his doctor. I was informed of this when I went to the nursery to feed Caleb. I was always left in the dark by the first hospital. That night William and I attended Caleb's 6 pm feeding. The nurse in charge of Caleb that night told us that **Caleb was floppy earlier so she tested his sugar.** It was at 20. **She sent the blood to the lab and it was actually under 20.** She said Caleb received some sugar water in a bottle and that he perked right up. Both my husband and I asked if they are still monitoring his sugar. We were told that it was not necessary. It was just an episode for my having Gestational Diabetes. I went on to explain to the nurse that I have a family history of Diabetes. She still said that there was no need.

That night Caleb took 20cc of formula. Over the week William and I would make our daily visits to drop off breast milk and to feed our son. During these visits we were not allowed to hold him until it was time to feed him, then we would have to leave right when he was finished. **Needless to say, we never really got to bond with our son. I will never forgive the hospital staff for that.**

I was allowed to change Caleb's diaper a few times. I loved it. I was actually allowed to take care of my child. **During this time Caleb became a lazier po feeder according to the doctors.** Caleb was put on an NG tube on his fifth day of life. Caleb would start off on a bottle for 20 minutes, and then what formula was left would be given by NG. On Caleb's 10th day of life everything was looking bright. We were present for his 9 pm feeding. Caleb took 29cc in 10 minutes. I made a comment to my husband, "Watch, Caleb will be home this Friday or next Friday by the latest." I said this on a Tuesday night.

**The following afternoon at 1:30 I received a call from the NICU telling me to come to the hospital. I didn't hear any concern in the doctor's voice. I asked if Caleb was being released. I was told that **Caleb had gotten worse.** Of course I said "What?" I was then informed that during Caleb's noon feeding he became floppy in the nurse's arms and that he was given oxygen. I said "How is he right now?" They said, "He's still floppy, cold and pasty white." I called William home and we arrived an hour later. **I have never seen a baby so white.**

We sat caressing our son until 6:30 pm. **That is when I ran out of the nursery while they were bagging Caleb.** I came back after he was stable to tell him that I loved him and to fight for mommy. I wanted to wrap my baby in a blanket and just hold him. He was so cold. During this time I was informed that he was being transferred to a level three hospital. Caleb was transferred at 9 pm. We were informed that **we would not be able to see Caleb until morning.** I kissed Caleb goodbye and told him that I loved him.

The transport team went one way while we went another. As we were heading to the parking lot the transfer team were coming out a different door. A little boy was walking in front of us, he said to his parents "Awe, look at that cute little baby," **I broke down ~ praying to God, "Please don't let this be the last time that I see my son."** William and I sat in our car until the ambulance pulled away. I cry whenever I hear sirens.

**Caleb survived the transport.** I called that night at midnight to see how he was doing. I was told that he is hanging in there. And that he was receiving a blood transfusion due to anemia. I could hear my baby crying. At this point whenever someone would touch Caleb he would cry. He knew that another needle was coming. I asked if he was going to make it through the night. I was told yes and that I can visit him tomorrow. I was up all night just staring at the clock waiting for it to turn to 9:30 am.

We arrived at 10:30 to visit Caleb. The doctor in-charge of Caleb that day spoke to us. **We were informed that Caleb might have an inborn disorder that affects the heart. We were then told not to expect Caleb to survive the night.**

I kept praying that God would spare my son and if it was not meant to be to please let my mom make it in time from Michigan so that she can hold his grandson. William's parents flew in from Texas. They were able to welcome their first grandchild into the world and they were able to be present when he passed.

During our first visit Caleb opened his eyes three times. It was if he was telling me, "Hey mom I am still here." **That was the last time I saw Caleb's beautiful blue eyes.** I also noticed that his umbilical cord came off. I made the nurse find it. William and I were waiting for the day that it would fall off. Will used it as an excuse for not changing his diaper. The excuse was now gone. The cord and a lock of my son's hair are the only physical reminders I have. The second visit that we were allowed that day I just kept touching him. I kissed any part of him that didn't have an IV in it. Caleb had six total. And he was on a respirator. By the third visit the IVs all started failing on him. A central line was ordered, but the surgeon never showed. At 8:30pm I had my son Baptized. It was something I felt I had to do. I wanted my son to be able to enter Heaven to be with my father. This way I knew he would be in good hands. Everyone in the NICU sang YES JESUS LOVES CALEB.
Caleb Patrick, TFP… cont’d

Visiting hours were over at 10:30. I was kicked out at 10:45. Caleb arrested at 11:00 pm. They were able to revive him. I was told at 11:30 that this happened. I told the doctor that if it happens again that I want to be there. "I was there when he came into this world ~ I will be there when he leaves."

I asked if I could visit with Caleb. I was told no and that they were trying to stabilize him. Around this time a nurse from the first hospital came to be with us. Caleb was known as their little Mr. Grabow. He touched so many in such a short time. We were talking when the doctor informed us that Caleb stop breathing. I ran in back. My son was already blue. I had them stop what they were doing. I said let my son go in peace.

I ran in back. My son was already blue. I had them stop what they were doing. I said let my son go in peace.

They unhooked the last two IV's and wrapped him in a blanket and placed my baby in my arms. Caleb took his last two breaths in my arms at 12:45 am. It was Caleb's 14th day of life.

Afterwards I gave my son his first bath and the nurse from the first hospital combed his hair. She was the first one to do it and I wanted her to have the honor of being the last. I put my son in a diaper and dressed him for his next journey into God's hands.

The following day was Friday. The day I thought my son would be home by for sure. It was the day I had to plan my son's funeral. The day I had to pick out his urn. I finally brought Caleb home on October 9, 2001.

No mother should have to put her child's urn in a crib. I kept telling Caleb I couldn't wait until he was home sleeping in his own bed. I will be crying for the rest of my life.

I am just grateful that I was Caleb's mommy for fourteen days. Caleb we love you BIGGIE MUCH.

Love,
Mommy and Daddy
Shelly and William Grabow
boo1974bear@netzero.com

Update to Caleb’s Story ~ Birth of Caden and Noah (TFP)

(Caden and Noah’s pictures are in the Kids Korner Section—Caleb’s Story and Picture are online)

Since telling our story we have become proud parents to Caden Nathaniel Grabow on 12/31/02 and Noah Riley Grabow on 11/18/03. Both Caden and Noah were tested for TFP via Supplemental Newborn Screening after birth. Caden does not have TFP, but he might be a carrier for his father’s mutation. However, he does have Multi-Cystic Dysplastic Kidney Disease. On September 23, 2003 his left kidney was removed. Caden is in great health. He’s a normal eleven-month-old who enjoys talking, crawling, walking and most importantly getting into everything. Noah on the other hand tested positive for TFP. He was placed on a protocol for TFP. He is gaining weight and maintaining his blood sugars. He will be released from the hospital sometime this week after a one-month stay in the NICU. [Note from Shelly’s Dec 18th email: Noah was released from the hospital last night!]

Yesterday was one of the best days in my life. It feels so great to have him home even though I had no sleep. I lost count after the 20th time I went to check on him. I am so nervous and so very happy at the same time. I think Dad is doing better than me. He slept like a baby all night long. It must be nice! Caden still has no idea that he is a big brother. Noah is doing great with the change of environment. I can’t believe that he turned one month today.

We are hopeful that we will be able to continue to raise him here on Earth. I have to admit that my days are shadowed by the fear of the unknown. After Caleb passed away I realized for the first time how precious life is and how short it can be. We have had to learn to make changes in our life. We can no longer plan things. We have to take things day by day. We never know what may or may not happen. I keep being told that Noah is doing great but not to forget that things can change in an instant. All that we can do is love and care for our children and hope that we are doing the job right. I am so scared of failure but when it comes to being a caregiver and parent to our children I cannot afford to fail especially in Noah’s case. It would cost him his life. I just thank God everyday for my family and FODSupport. I know that I am not alone. I have somewhere to turn when I have questions…and a place where I can talk about my fears. Thank you for being there for us.

Sincerely,
Shelly Grabow

FOD Family Questionnaire

If you do NOT see your name on past Family List, it is because I (Deb) never received the FOD Family Questionnaire that I sent you in the Family Packet when you first registered with us. If you would like to be listed for networking purposes, please go to ‘Online Forms’ on our website (www.fodsupport.org) and print out the Questionnaire. Then SIGN it and DATE it so I have your permission to list you. Please mail it to me via the regular mail (see page 1 of this issue for address) so we can list you in the next List Update.
Hello, my name is Curt and I have been a member of the FOD Group for about 4 months now and really enjoy reading your postings. Well let me tell you a little about myself. I'm 36 and live in the Adirondack Region of upstate New York...although I was born and raised in North Carolina. I was a Sautée Chef for most of my life, well at least 16 years worth. I had to "retire" in 1998, two years after my diagnoses of CPT 2 and fiber atrophy 2. That was probably the hardest thing to accept...cooking was more of a passion than a career for me. I've always been an outdoor type person from the time I can remember, always climbing trees and such. My health was what I had thought to be great growing up. As a kid I was always very active, boxing from the ages of 12-18, hiking/climbing...just doing the things that kids do. Don't get me wrong, I had my complaints of aches and pains...but they were always contributed to "Growing Pains"... don't you hate that?

I spent the better part of my life thinking that I was crazy and I'm sure I'm not the only one. Talk about emotions going haywire...I can tell you stories there. I mean when no one will listen, listen with not only their ears, but hearts as well, you have a tendency to get a tad bit angry. "Am I crazy?" you ask yourself, thinking if this Doctor of 30 years can't find anything wrong, well...perhaps it is in my head. I still pushed on with the way I was living my life, doing the same things that I've always done, but a price was to be had, and it hurt like hell too. I still try and enjoy the same activities that I have always had a passion for, but I've learned to take it easy and go slower if I need to.

Having both a neuromuscular and metabolic disease my symptoms vary: I get severe leg cramps as well as having my sides cramp up, kind of takes my breath away for a moment if you know what I mean. I also experienced "Rhabdomyolysis" a few different times, but never felt it was serious enough to go to the hospital. At times I find it hard to muster up the energy to do even the simplest of tasks...just no energy.

As for my neuromuscular disease, well the symptoms are slightly different. I still cramp up at times, but I'm also losing muscle cells...which makes it a little harder to do some things. My gait isn't the best...I'm like a "Weeble Wobble, but don't fall down." Thank God my fiancée is a Registered Nurse. I am currently taking a creatine supplement, trying to eat the proper foods, and avoiding too much exercise. I guess if there was a reason for me to have these diseases I'd have to say to make me more compassionate, to understand others...after all we're all human. As strange as this may sound...I'd never want to be cured (normal). I now see life through a whole new set of eyes, and I'm happy with what I now see.

Thank you for allowing me to ramble on.

Curt Vose
New York
www.bearphotoimages.com
curt@bearphotoimages.com

New FOD related articles (full articles in pdf will soon be posted on our website on the Medical Information page/Medical Articles):


• Spiekerkoetter U, Khuchua Z, Yue Z, Bennett MJ, Strauss AW. Mitochondrial trifunctional protein deficiency due to either a or b-subunit mutations is one uniform disease because mutations in either subunit have equal effects on TFP complex stability. Pediatr Res, in press.

Look inside the cell of a human body and you’ll find a control center, the nucleus of the cell. Inside the nucleus are the set of instructions that tell the body what proteins need to be made, and how to make them. Proteins are important to the body, and have many roles, including enzyme activities.

Inside the human nucleus there are 46 chromosomes. Chromosomes are “packages” of DNA (deoxyribonucleic acid). DNA serves as the set of instructions the body utilizes to make proteins. Looking more closely at a chromosome, one will find genes. Genes are the individual instructions that result in the production of specific proteins.

Humans have 23 pairs of chromosomes (46 chromosomes total). Chromosomes are inherited from your parents. Therefore, 1 member of each chromosome pair is from your mother, and the other member of the pair is from your father. Each of us should then have 22 pairs of “autosomes” and 1 pair of “sex chromosomes”. Sex chromosomes determine gender: XX = females, XY = males. Autosomes are simply not sex chromosomes. For example, a woman inherits 22 autosomes and an X chromosome from her mother AND 22 autosomes and another X chromosome from her father. A man inherits 22 autosomes and an X from his mother AND 22 autosomes and a Y chromosome from his father. This simply means that we inherit ½ of our DNA from our mother, and ½ from our father.

When chromosomes are studied, the autosomes are numbered from 1 – 22 and each chromosome is matched to its “partner”. So, looking at a picture of your chromosomes (a karyotype) looks something like a bunch of worms paired up and numbered plus a couple labeled with X’s and/or Y. The karyotype allows us to view the structure of the chromosome, and to verify that the set is complete in number. Genes however, are too small to see by looking at them with the unaided eye and require more specialized testing.

Changes can occur in genes that change the specific set of instructions normally spelled out in the DNA. These changes can result in disease. Both the normal set of instructions and a set of instructions with changes (called mutations) are inherited. There are several types of inheritance, but this article will focus on autosomal recessive inheritance since most FOD disorders are inherited in this manner. Remember that autosomal means not a sex chromosome, therefore autosomal means one of the chromosomes numbered 1 – 22. So what does recessive mean? Recessive inheritance means that a person must inherit two copies of the gene that causes disease in order to be affected with that disease. This means that each member of the chromosome pair has a copy of the disease gene. If a person has one copy of the disease gene on one chromosome, and no other copy of the disease gene, the person is a carrier and not affected with the disease. For example:

Let’s assume that mom and dad have a complete set of chromosomes, and let’s ignore the other chromosomes except for the imaginary chromosome R. R represents a “normal” chromosome, and “r” represents its “partner” but with the disease causing gene. The following possibilities exist for this couple’s children:

Rr (carrier mom) RR (dad)
RR (mom) OR Rr (carrier dad)
50% RR (not carrier, not affected)
50% Rr (carrier)

Rr (carrier mom) Rr (carrier dad)
25% RR (not affected, not carrier)
50% Rr (carrier) 25% rr (affected)

Note that these risk figures are for each pregnancy. Having one affected child does not decrease your risk for having another affected child with the next pregnancy. Additionally, there could be more than one gene that can cause a single disease. In that case, the genes inherited from the parents do not have to be identical. This couple can pass on two different genes known to cause the same disease, and the child be affected with that disease. The risk to their children is the same as illustrated above.
Mitochondria, specialized compartments present in every cell of the body (except red blood cells), produce 90% of the energy needed to support growth and sustain life. Mitochondrial disorders result from a decrease in the ability of the mitochondria to make energy in the form of adenosine 5' triphosphate (ATP). There are over 40 known types of mitochondrial disorders. These disorders often affect tissues that have a high-energy demand, such as the heart, brain, eyes, and skeletal muscles. The symptoms may range from isolated eye problems to severe developmental delay with muscle weakness. Because of the diverse symptoms, there is limited information on effective treatment therapies. The goal of cofactor treatment is to increase ATP production and to reduce the buildup of toxic metabolites. The use of supplemental vitamins and cofactors is largely unproven and their use in mitochondrial disorders remains controversial. Coenzyme Q₁₀ is the most widely used supplement in the treatment of oxidative phosphorylation (OXPHOS) disorders.

What is CoQ₁₀?

CoQ₁₀, or ubiquinone, is a natural substance found in our body that helps transfer electrons in the respiratory chain. This process is part of ATP synthesis, or energy production. CoQ₁₀ also acts as an antioxidant and helps protect cells against oxidative damage. CoQ levels in the body are maintained by the body's own production of CoQ and from dietary sources, mainly of animal origin.

Is it Effective?

CoQ₁₀ has been reported to have a beneficial effect on clinical outcome and biochemical measures in a variety of mitochondrial disorders. The positive effects have included a reduction of serum lactate and pyruvate (Abe et al. 1991b; Bresolin et al. 1988c; Goda et al. 1987b; Nishikawa et al. 1989b; Ogashahara et al. 1985b; Yamamoto et al. 1987b), improvement in cardiac conduction defects and eye movements (Goda et al. 1987a), reduced muscle weakness (Abe et al. 1991a; Ihara et al. 1989b; Yamamoto et al. 1987a), and improved exercise tolerance (Bresolin et al. 1988b; Goda et al. 1987a), improved oxygen utilization during exercise (Abe et al. 1999), decreased peripheral nerve damage (Ihara et al. 1989a), improvement in neurological function (Bresolin et al. 1988a), increased respiratory chain activity, and acceleration of post-exercise recovery (Bendahan et al. 1992; Nishikawa et al. 1989a). Most reports regarding treatment have been case studies or anecdotal reports with limited numbers of patients, various treatment periods, and CoQ₁₀ dosages ranging from 30 to 300 mg/day.

Several short-term studies have shown variable results with CoQ₁₀ treatment. Two trials in patients with muscular dystrophies and neurogenic atrophies showed an improvement in cardiac function and physical performance (Folkers & Simonsen 1995). In another short-term study in patients with mitochondrial encephalomyopathies, a trend of effectiveness of CoQ₁₀ was noted by improved muscle endurance, decreased fatigability of daily activities, and decreased serum lactate and pyruvate levels, but statistical significance was only noted in global muscle strength (Chen et al. 1997). In a study with CoQ₁₀ supplementation in patients with mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS), there was an improvement in neuromuscular symptoms, but no difference in fasting glucose or glycemic control (Andersen et al. 1997).

Longer-term studies of six months duration have evaluated the effectiveness of CoQ₁₀ treatment. Phosphorous magnetic resonance spectroscopy (³¹PMRS) was utilized to study the effect of CoQ₁₀ treatment on brain and skeletal muscle mitochondrial function. Baseline brain and skeletal muscle metabolism was compared to 36 age-matched healthy controls. Mitochondrial function in both brain and muscle was reduced by 25% and 29%, respectively, compared to controls. Treatment with CoQ₁₀ statistically improved phosphorylation potential and calculated ATP synthesis in both brain and skeletal muscle in all patients studied (Barbiroli et al. 1997).

In a multi-center trial of CoQ₁₀ in mitochondrial cytopathies, a 25% decrease in post-exercise lactate levels was observed in over one-third of the patients (Bresolin et al. 1990). A bicycle ergometry study in patients with mitochondrial encephalomyopathies treated with CoQ₁₀ showed no change in metabolic parameters after three months of treatment, but after six months of treatment, a decrease in lactate/pyruvate ratios at rest and in association with exercise was noted in approximately half of the patients (Chan et al. 1998). It is unclear why some patients respond and others with the same clinical phenotype and biochemical defect do not show any beneficial effects.

Continued on Page 8
The longest-term study examined supplementation of CoQ$_{10}$ in patients with the 3243 MELAS mutation. The group of the ME-LAS patients with diabetes experienced an increased insulin secretory response and improved lactate response after exercise with the CoQ$_{10}$ treatment. In addition, there was no progression of hearing loss (a common symptom in this disorder). The CoQ$_{10}$ treatment did not affect the insulin secretory response of the patients with impaired or normal glucose tolerance (Suzuki et al. 1998).

While most cases of decreased CoQ$_{10}$ levels are secondary to other causes, primary muscle CoQ$_{10}$ deficiency has been documented (Musumeci et al. 2001b; Rotig et al. 2000; Sobreira et al. 1997). In patients with muscle CoQ$_{10}$ deficiency, CoQ$_{10}$ administration resulted in dramatic improvements in strength, seizure control, muscle weakness, and ability to walk (Musumeci et al. 2001a).

Availability and Use of CoQ$_{10}$

Commercially prepared CoQ$_{10}$ supplements are available as powder-filled, hard shell capsules; oil-based suspensions in a soft gel capsule; emulsions in a soft gel capsule and liquid. There are limited reports on the bioavailability or absorption of CoQ in these preparations. CoQ can be purchased from a variety of sources, including the local drug store. Animal and human studies have demonstrated that approximately 2 to 10% of the dose administered is taken up into the blood (Weber 2001; Zhang et al. 1995). Dosages of 90 to 150 mg/day of CoQ$_{10}$ have been shown to increase plasma concentrations by 180% (Kaikkonen et al 1997). The benefit of CoQ$_{10}$ supplementation in mitochondrial disorders is unclear. Many patients report improvement in clinical symptoms, and side effects from large pharmaceutical dosages are extremely rare. It is hoped that CoQ$_{10}$ may enhance enzyme function and result in an increase in energy production. CoQ's role as an antioxidant may help slow the progression of the disease. Dosage in the treatment of mitochondrial disorders varies but clinicians currently recommend 4 to 15 mg/kg/day to determine the effectiveness in an individual patient (Gold & Cohen 2001). Remember, consult your physician before starting any treatment. Adapted from: Marriage B et al. Nutritional cofactor treatment in mitochondrial disorders. JADA 2003:103:1029-1038. References for this article are posted on www.fodsupport.org. Medical Information page, scroll down to Related Informational Articles

Q: What fats do MCADers need to avoid?

A: Fats are made up of long chains of carbon atoms. They are referred to as very long, long, medium and short chains depending on the number of those carbon atoms. Very long fats >20 or more carbon atoms, long 18-14, medium 12-4, short <4. Each chain length has specific dehydrogenase enzyme that cut off 2 carbons at a time when functioning normally. Hence, MCAD = Medium Chain Acyl CoA Dehydrogenase deficiency. In that disorder fats with carbon lengths between 12 and 4 cannot be broken down and toxins build up. Aside from MCT oil, most medium chain fats in our diets come from the normal breakdown of very long and long chain fats that are found in meats and dairy products. So we really cannot completely avoid eating these, and we would not want to because fats are critical to the normal development of our nervous system and hormone synthesis. So we eat wisely - lean, low fat meats and fish, veggies, fruits etc. Avoid saturated and trans fats as much as possible. Avoid too many simple carbohydrates that create obesity - instead use complex carbohydrates. There are some websites that do list fat chain lengths (i.e., www.nutritiondata.com). The bottom line is though that you need to monitor the saturated fats and foods with lots of long and very long chain fats.

Lynne Wolfe
Metabolic NP
LAWPNP@aol.com
question & answer ... cont’d

Q: I have a daughter who was diagnosed with MCAD. Has anyone been told that their child carries two different mutations and that because of the two different mutations the MCAD is ‘mild?’ We still have her on a 3 to 4 hour feeding schedule, but I’m wondering why does having the two different mutations make a difference?

A: Many people with recessive genetic conditions (needing mutations in both copies of the gene to have the condition) have two different mutations. There are many places on a gene where a mutation can occur and two different mutations only reflect the parents’ individual genetic backgrounds. In the case of MCAD, it happens that the A985G mutation is by far the most common, and about 80% of patients have TWO copies of this mutation. So your daughter is not considered ‘mild’ because she has two different mutations, but because we know something about the nature of those mutations individually. The T199C is a newly discovered mutation that was only detected when states started screening for MCAD at birth. That meant that all these kids were diagnosed with MCAD before they ever had the opportunity to get sick and be diagnosed that way. Many of the kids diagnosed by newborn screening might NEVER have become ill from their MCAD, or only have been diagnosed much later in life during a very severe viral illness. The T199C mutation has not been seen in a child who was diagnosed by her symptoms, but is showing up a lot in kids diagnosed by newborn screening and so that implies that it is a mild mutation. Studies of the mutation and what it does to the enzyme show that it only mildly disrupts the way the enzyme works and so the enzyme works pretty well under normal conditions. If anyone is interested in reading a scientific journal article about MCAD mutations, below is an abstract from Andresen et al. American Journal of Human Genetics, 68, 1408-1418. With the exception of the T199C mutation, the best guide to whether your child is mild or not is her clinical picture and not her mutations. That is, how long can he/she generally fast before getting hypoglycemic, has she/he experienced normal childhood illnesses without needing special support, or did a cold or sinus infection put her into the hospital? Does she need cornstarch at night or a nighttime snack to avoid hypoglycemia in the morning or can she make it through without trouble? These are questions I’d encourage you to discuss with your metabolic team and are better indicators of how mild or severe she might be. Also, if your child was diagnosed years ago, they might not have identified the second mutation. The A985G mutation is so variable. Case in point: we just diagnosed a little girl with MCAD who is homozygous (has two copies of the same mutation) for A985G. She didn’t get hypoglycemic until she had an operation and had been fasting and then vomiting for 36 hours! So you really can’t make any predictions by using the mutations alone. I encourage all of you to discuss your questions with your metabolic team and a genetic counselor (if he/she is not part of the team already). You cannot depend on the mutations alone to predict how your child will do. This is not a black and white issue in terms of what is mild and severe, and having a label of ‘mild’ or ‘severe’ isn't necessarily helpful either. There's so much variability with MCAD (and other FODs); there are obviously other factors, but they’re not clearly understood. But we don't need to understand exactly what makes one child more sensitive than another to treat that child appropriately. I think it's best to realize that everybody who has children with MCAD (or other FOD) have that common bond over the diagnosis, but beyond that, each child is an individual. As with any medical condition, there are common ways to treat, but management really needs to be individualized. So talk to your doctors and nutritionists about your children and ask about the thinking behind their management decisions so you'll have a good sense of your team's approach to care.

Lisa
Genetic Counselor

A: (additional comments from Lynne, Metabolic NP)

This is a perfect time to agree with Lisa’s comments above. FODs, OAs, and many other biochemical disorders really cannot be assigned a mild to severe label based on any DNA mutations alone - we have to look at how each individual reacts. In Genetics, there are two concepts that are critical. Phenotype = what the patient looks like physically, biochemically etc., AND Genotype = what the genes actually are. Most of the FODs have been studied to look for matches between Phenotype and Genotype because IF the Genotype really did dictate the Phenotype, we could manage disorders easily. That is not the case at all. There really is not any Phenotype/Genotype matches in any of these disorders. As Lisa said, each child is an individual and will present with symptoms of their FOD depending on their other Genetic predispositions, their environment, etc. We are all still learning about these disorders and how genes dictate our normal body functions. It will be a while until we can say for sure what the real impact is of all the genetic mutations we are still in the process of discovering. The difficulty is that humans have normal variations in genes called Polymorphisms and they occur in a predictable incidence in the normal population. When that is all that is found in a patient who has a disease, and it is unlikely that a common polymorphism is causing the disease but nothing else has turned up, it is occasionally labeled "mild" for want of any better explanation. However, it is more likely that either the patient has a "private" mutation (unique to their family) or a mutation that has not been previously discovered. We have LOTS to learn about Genetic disorders.
NBS Update

Be sure to visit our website (In the News page) for the current articles on NBS efforts across the country. More states are getting on board (albeit slowly!) so check http://genes-r-us.uthscsa.edu/resources/newborn/screenstatus.htm every now and then to update yourselves on what your state is adding to their NBS panel of tests.

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Attention All Canadian Families

My name is Tammy Clark. Some of you may already be familiar with our daughter Jenna's tragic story of undiagnosed Medium Chain Acyl CoA Dehydrogenase Deficiency (MCAD). Please visit her site at http://www3.sympatico.ca/tammy_roger/Jennastory.htm. I am posting this message specifically to Canadian families to request your support in establishing a Canadian chapter of Save Babies Through Screening. The main goal of the group would be to raise public awareness of the need for the Canadian government to take some initiative to guarantee access to expanded newborn screening for all Canadian babies. Currently not all Canadian babies are receiving the same standard of care when it comes to newborn screening. For example babies born in Ontario are being screened for two disorders and babies born in Saskatchewan are being screened for up to 30 disorders. Babies are currently being screened for MCAD in BC, Saskatchewan and Nova Scotia. It saddens me that babies are not having the same opportunities at a healthy start in this country.

We are supposed to have "universal" health care in this country. The most important component to this initiative is all of you out there right now reading this message. I realize that people get busy with their lives, caring for your families, work etc., but if we the families affected by this situation do not speak up, children such as our Jenna will continue to die needlessly. Together we can make a difference!!

Please e-mail me directly at canadasavebabies@yahoo.ca. I would really appreciate any support (whether it be just sharing your child's story, or signing a petition etc.) your family could offer with this initiative.

Sincerely,
Tammy Clark

Pharmaceutical Update

Sigma-Tau Pharmaceuticals, Inc., makers of Carnitor®, can be reached at 1-800-447-0169 or on their website http://www.sigmataupharma.com/. Their Carnitor® website at www.carnitor.com helps to Educate and Empower Providers and Consumers by providing important information about carnitine deficiency and supplementation with Carnitor®.

Pampered Chef ‘Fundraiser’

I would like to undertake a fundraiser for the FOD Support Group. My sister, Sheri Merrill (Kristen, MCAD) ran the fundraiser last year. I have been an Independent Pampered Chef Consultant in Vermont for one year now. Pampered Chef offers a product fundraiser that has a limited amount of products that range in price of $4.50 to $14.00. Forty percent of sales will be donated to the FOD Support Group. This will NOT be tax deductible. The FOD Support Group does NOT have tax-exempt status so you will not be able to deduct this from your taxes. Here is how it will work. Email me directly if you are interested. If you would like to participate I will mail you the product fundraiser order form. You will then need to collect a minimum of five orders or $100.00 in sales. Mail the order form back to me with payment in FULL (if people write checks have them make them out to Debbie Fagnant). I will then send in the order to Pampered Chef and it will be mailed to you directly and you will separate the orders and deliver them. I will mail you the receipts to be sent out with the customers’ orders. Once I send in the order I will send a check to the FOD Support Group for the funds raised. I will also send a letter stating who raised the funds for the Group. I think this is a great opportunity to give back to the FOD Support Group. If anyone is interested please e-mail me at dpfagnant@prodigy.net. If you would like to see the Pampered Chef products you can visit their web site at www.Pamperedchef.com. Thank you and I hope to hear from you soon. P.S. Pampered Chef products make great gifts too!

Debbie Fagnant
dpfagnant@prodigy.net
Please remember these families in your thoughts and prayers throughout the year

Toni and Mark Cline
Kasie - Birth June 6, 1990 Death March 10, 1991

Sandy and Jon Cooper

Martin and Kathy Davis
Mary Katherine - Birth June 27, 1996 Death Nov 7, 1996

David and Amy Deshais

Doug and June Evenhouse
Marie - Birth Dec 15, 1985 Death Nov 19, 1986

Andrea and Phillip Franklin
Brandi - Birth Dec 2, 1986 Death Jan 1988

Lance and Dawn Goldsmith

Deb and Dan Gould
Kristen - Birth Oct 6, 1983 Death July 21, 1985

Shelly and William Grabow

Brandis Greichunos
Madison Burchette - Birth March 8, 2001 Death March 24, 2002

Jeannette and Keith Guillory
Dominique - Birth Jan 21, 1997 Death Jan 23, 1997

Nicole and Chris Gulinello
Alec - Birth Feb 21, 2001 Death Aug 24, 2001

Michael and Nicole Gumiela
Michael - Born March 28, 1998 Death April 4, 1999

Carol and John Hall
Sarah - Birth June 8, 1998 Death July 30, 2000

Robin and Vince Haygood
Ben - Birth Feb 19, 1998 Death Aug 8, 2000

Ralph and Angie Hedrick
Chelsea - Birth Jan 11, 1995 Death Apr 3, 1996

Nikki and Toby Hiatt
Reece - Birth Aug 1998 Death April 18, 1999

Pauline and Bill Hill
Amy and Matthew Hoffman

Brad and Kim Holmes

Debbie and Dave Houk
Lauren - Birth May 4, 1988 Death Dec 15, 1989

Robert and Dixie Howard
Cody - Birth July 30, 1987 Death Dec 26, 1992

Stephanie and Doug Huber
Jace - Birth March 8, 2000  Death Feb 14, 2001

Meredith and Neil Hughes
Claire - Birth Sept 1, 1986 Death June 23, 1997

Karen and Steve Tmhoff
Michael - Birth July 25, 1991  Death July 8, 2002

Brian and Patricia Karhu

Vickie and Burnell Keller
Paul - Birth Mar 31, 1993  Death Sept 20, 1993
Annie - Birth Nov 26, 1998  Death April 22, 1999

Diane and Mickey Kennedy
Marie - Birth Dec 1, 1989  Death Oct 5, 1991

Andy and Temple Ketch
Nancy - Birth Feb 8, 1989  Death July 20, 1990

Robert Knoff
Teresa - Birth Nov 7, 1994  Death June 29, 1995

Sondra Koehn

Jamie and Tom Lazzaro

Lisa and Pete Leonard
Devin - Birth July 18, 1997  Death July 19, 1997

Mary Lingle
Candice - Birth Feb 21, 1991  Death Nov 8, 1993

Darlene and Larry Lopez
Marissa - Death Feb, 1999

Heather and Phillip Marsella

Ron and Paula Matthews
Daniel - Birth May 19, 1981  Death Jan 12, 1982

Randy and Misty McDonald

Christine and Mark McFarland

Linelle and Matt Meadows
Cole - Birth Mar 21, 1999  Death Oct 18, 1999

Elvira Melendres
Katherine - Birth Mar 6, 2000  Death May 3, 2000

Lori and Jeff Michaud

Simone and Michael Miller

Mike and Sheryl Mulhall
Justin - Birth April 22, 1990  Death April 22, 1990

Verna Parker

Diana and Kevin Patterson

Steve Bruski and Liz Pease
Caitlin - Birth July 10, 1989  Death May 10, 1996

Albert and Arleen Phang
Andrew - Birth Dec 7, 1989  Death April 17, 1991
Alexander - Birth Dec 3, 1994  Death Feb 8, 1995

Jennifer and Jason Pierson
Alexander - Birth June 1, 1995  Death June 3, 1995

Stephanie and Andrew Plaisted
Drew - Birth May 7, 1997  Death Dec 27, 2000

John and Sally Reichelder
Zachary - Birth March 24, 1997  Death March 27, 1997

Tanya and Pat Robitaille
Richard - (stillborn) June 24, 1993
Rachel - Born August 13, 1995  Death December 29, 1995

Brian and Cherryl Rosenberger

Janice and Steve Rowland

Litzy Sanz de Solis and Jesus Solis Sanchez
Jesus - Birth Sept, 14, 1996  Death March 16, 1998

Jackie Shears
Welcome to new babies!

- Mandy Myram and Enrique Cabrera welcomed Lara (sister to Joe, SCAD) into the world on May 15, 2003. She weighed 3.4kg (almost 7½ lbs) and was 49cm (19½ in).
- Marjorie and Walter Vukelich are pleased to announce the birth of Daniel Paul (brother of Nickolas, MCAD). He was born on June 20, 2003, at 10:48pm and was 7lbs 4oz, and 20 ½ in long.
- Matthew Baer (brother of Nicholas, SCAD) came into the world on July 17, 2003. Jackie and Mike are the proud parents.
- Noah Grabow, TFP (brother to Caleb, undiagnosed TFP, and Caden, unaffected) announced himself to the world 8 weeks early on November 18th at 10:32pm. He weighed in at 2lbs 14 oz and his parents, Shelly and Will, say he’s holding his own and growing every day!
- Liz and Frank Harnos (parents to John and Steven, both MCAD) called to tell me Will (MCAD) was born on Feb 23, 2003 and was 9lbs 13oz – it was great to talk with you!
- Betsy and Eric Furler and son, Henry (unclassified FOD) welcomed Sam on December 17, 2003. He weighed 6lbs 10oz and was 20” long.

Condolences

We were very saddened to hear of the death of Grace Hoffman (SCAD) on October 25, 2003. Grace’s parents, Amy and Matthew, said that her death was unrelated to her SCAD. Our deepest thoughts and prayers go out to the Hoffman Family at this time of saddened hearts.

Deb’s Updated Email Address
Please update your address book –
Main address is deb@fodsupport.org
Backup address at fodgroup@triad.rr.com

VLCAD Email Network

Gina (Brett, VLCAD) is looking to start an FOD subgroup for VLCAD families. If you are interested in networking with other VLCAD families around the world by e-mail, then please e-mail her at mjb3@frontiernet.net. She may also be reached by phone at (845) 928-9574.

‘Love builds bridges where there are none’
~ R.H. Delaney
**SNAP’s Medical Insurance Empowerment Program** will help you understand your policy and problem-solve your insurance nightmares! 1-888-310-9889 or [www.snapinfo.org](http://www.snapinfo.org) for more information.


Reviewed by Robert Naseef, PhD on [www.specialfamilies.com](http://www.specialfamilies.com) (this ‘snippet’ is shortened by Deb) ~

‘Looking for a good book about inclusion for children with a diagnosis within the autism spectrum? Look no further. A book dedicated to guiding the teaching of students with autism in the inclusive classroom is long overdue. In her book, she provides ready-to-use strategies for including students with autism in both primary and secondary school classrooms. First-person accounts of students who have autism give readers insight into the experience of having autism and show educators how to adapt classrooms to support student participation in class work, school routines, social activities and more.’

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**Tupperware ‘Fundraiser’**

I am pleased to offer a Tupperware Fundraiser to support the FOD Family Support Group! I have recently become a Tupperware Consultant. This fundraiser will be much like the Pampered Chef fundraiser that I offered last year (that my sister is offering this year) except 50% of sales will be donated to the FOD Support Group. This will not be tax deductible. If you are interested in a Tupperware Fundraiser for the FOD Support Group please e-mail me directly at Shermerr@juno.com.

I will send you the catalog and order forms to you and you will collect the orders. Then the orders and payment in full will be mailed back to me and I will process the orders. Shipping charges are 10% of the order product total and then charge your sales tax rate for your zip code/state. I cannot process the orders without this. For those of you that have sent me orders already I will process them as I receive them.

Orders will be sent by Fed Ex to your house and you will distribute the orders. Not all Tupperware products are available in the Fundraiser. Each person collecting orders will be considered a separate fundraiser, but all money raised will be sent to the FOD Support Group. Or if you don't want to collect orders you can place a single order and 50% will be donated to the FOD Support Group (at the fundraiser price). If you have any questions, please email me directly.

Sherri Merrill (Kristen 6, MCAD and Jamie 4, Carrier)
Shermerr@juno.com
For more information on products offered visit [www.Tupperware.com](http://www.Tupperware.com)
Caden and Noah (TFP) Grabow (brothers of Caleb, Undiagnosed TFP)

Martina Weyand (MCAD)

Morgan Jones (GA 2)

Drew Bryan (VLCAD)

Maggie Dozier 14 (MCAD)  
Climbing the NC mountains

Candace and Reatha Boyd (LCHAD)

Clark Family From Canada  
Tammy, Roger, Jasmine, and Justin~not pictured, Jenna, undiagnosed MCAD, but always in our hearts.
Family & Professional Donations


**Professional Donations:** Sigma-Tau Pharmaceuticals, Inc. (makers of Carnitor®)

We greatly appreciate donations to help with postage and copying fees. **Checks can be made payable to FOD FAMILY SUPPORT GROUP.** Because we are not officially a non-profit organization, donations are not tax deductible.

Reminders

**Families** - Please send **TYPED** stories by June 1, 2004. To be listed on the FAMILY LIST, please return the SIGNED Family Questionnaire or hand-write your information as seen on the current Family List and sign and date it. Continue to spread the word about FODs and the need for screening -- it will SAVE LIVES!

**Professionals** - Please let us know about your research and/or clinical work with FOD Families. Send articles by June 1, 2004. Also, please return to Deb the **Professional Questionnaire** even if you are already listed on the printed Professional List.

Communicate With Us

Please **ADD** me to your mailing list

Family  Professional  **(please circle one)**

Name/Address or Address Correction **(circle one)**


Please **REMOVE** me from your mailing list:

Name/Address:


Please include ideas for future issues or your questions

*The purpose of life is a life of purpose*

~ Robert Byrne ~

Thank You

Thank you to Erika Wallace - erikawallacepa@yahoo.com (Mailing Lists), Mary Lingle - Mcartwrite@aol.com (Web Page) and Brian Gould - briangould@triad.rr.com (newsletter) for all your hard work.

Special thanks to Sigma-Tau Pharmaceuticals, Inc. for their continued financial support.

The views expressed in the FOD Communication Network Newsletter do not necessarily represent the views of our Advisors or all of our members. Before trying anything new with your child or yourself in regard to treatment, please discuss matters with your doctor or specialist.

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