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This has been an extremely busy summer and I apologize for being so late on our July newsletter – but at least we had enough funds to print this issue and update the Family and Professional Lists! We not only had our National Metabolic Conference in June, but our family has been busy moving across the country (Dan will commute from Michigan State University for 2 years) and within the city of Greensboro (we sold our house and moved to an apartment) – so please note the ADDRESS CHANGE for our Group in the header above.

Our National Metabolic Conference near Detroit, MI on June 25-26th was a HUGE success and I want to thank ALL of our Families that attended – we had @ 45 for our daylong sessions on that Friday and Saturday. It was because of our Family and Professional donations that we were able to participate once again in this Conference that was sponsored by the National Coalition for PKU & Allied Disorders – THANKS Trish Mullaley for inviting us! 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.

...continued...

Our Families had the opportunity to meet several of our FOD Professionals, many of whom we have been corresponding with via email and phone for years, to learn from them and ask them questions about dealing with various FOD issues in an open forum. Along with their prepared presentations (you can view their power point slides on our website), I felt this part of the meeting was the most valuable for Families. It was also a great time to network with other Families dealing with a similar disorder. Start thinking about our next one in @18 mos – topics, speakers, and location!

We also had sessions on Newborn Screening, legal and educational issues related to having special needs, formula legislation, and a CALL FOR SUPPORT AND FUNDING from corporations, foundations, and other granting agencies connected with FODs (and other metabolic disorders) to not only support research but to invest in education and awareness (establish grants for this purpose) – WE NEED MORE TRAINED METABOLIC PHYSICIANS FOR OUR FAMILIES – and this fact was reinforced by Dr Mark Korson’s presentation discussing his METABOLIC OUTREACH PROPOSAL in the Boston/New England area and hopefully nationally at some point in the future. It’s great that research facilities are getting funding but we also need to train new and future Drs to help treat our Families – many of you have already run into difficulty finding an FOD TRAINED physician (or other metabolic professional, ie metabolic nutritionists etc.). So if you’d like to help in this effort, try contacting corporations or foundations that have available funds for grants and ask them to HELP SAVE OUR CHILDREN by creating innovative funding opportunities that directly support efforts to educate and train new physicians. We will provide more detailed information about Dr. Korson’s final proposal in future editions of our newsletter.

Thank you to Dr Marsha Fearing for sharing her research on FODs and Pregnancy ~ it’s important information for ALL the FODs and Professionals ~ PLEASE let me know if you’d like to share your knowledge and expertise.

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. Please let us know if you also treat ADULT FODers!

Whether you’re a Family or a Professional, we are all striving to create awareness, education, screening and diagnosis, treatment, and research ~ by being on the same team...

‘We Are All in This Together!’

Take care...      DLG

The FOD Communication Network Newsletter was created by and is currently edited by Deb and Dan Gould, 1559 New Garden Rd Apt# 2E, Greensboro, NC 27410 (phone) 336-547-8682 http://www.fodsupport.org email: deb@fodsupport.org  (AIM: fodgroup)  Medical Advisor: Dr. Charles Roe; charlesr@baylorhealth.edu

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**Family Stories - Bruna’s Story, Unclassified FOD**

Bruna was born on 22 July and at her birth we suspected that something was wrong because she couldn’t open her eyes. We saw our daughter’s eyes for the 1st time 15 days after birth.

I tried to breast feed but failed in my attempt. She was a very hard baby to feed. She lost weight week after week. Her first hospitalization was in September. She was dehydrated. They did a lot of tests but they didn’t find anything so she came home. Then in December she went again to the hospital and stayed in the NICU because she had apnea. At this time, however, they never checked her blood sugar. She was there for 3 months and in that time we thought that every day was going to be the last!

She didn’t smile for the longest time ~ she started smiling when she was 8 months. She had hypotonia and retained carbon dioxide.

The doctors suspected that Bruna was deaf and blind. Now I’m sure that she is not blind but we are not sure if she can hear. They wanted to give her a tracheotomy and a g-tube, but she started to eat and her carbon dioxide retention was lower.

To resume, Bruna has been in the hospital most time of her life (7 internments or hospitalizations). She started having hypoglycemia 3 months ago, although I think she was having that since birth but they only found out 3 months ago. The last one she had a 2!! Everyone in the hospital said it was a miracle that she didn’t die.

**SHE IS A SURVIVOR!!**

The doctor told us 2 weeks ago that she has a fatty acid oxidation defect, but it is unclassified at this time. Her study is being made in France.

I hope that you understand everything I wrote. There is so much more to say but I have a little difficulty in writing English. If you like you can call me, I think it’s easier.

I’m sending you a picture of Bruna and me. I have others I can share if you’d like to email me ~ her 1st day of living, her baptism, Bruna and her father, Bruna and our dear Dr Paula Azeredo Who is our Angel.

I hope you find her so beautiful and special as we do.

Thank you so much

Claudia Carmo
Portugal
Claudia.carmo@netcabo.pt

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**Family Stories - Michael’s Story, MCAD**

One Tuesday morning, when Michael was 13-mos-old, we got up and got dressed as usual. Michael went to a Mother’s Day Out program at our church. I dropped him off at 9 am. Around 11am or so I received a phone call that he had a fever and I needed to come and pick him up. I went to get him and brought him home. I gave him some motrin and put him in his crib. I took the video monitor in the room with me to keep an eye on him while he slept. A little while later I heard a strange noise coming from his room and looked down at the monitor. **He was having a seizure.** I rushed in to get him and called 911. The ambulance came and checked him out. They said he needed to go to the ER and either we could take him or they could. We opted to take him ourselves.

After many hours they said he had a virus and had a febrile seizure. This was the beginning down the road of a wrong diagnosis. About a month later, Michael was taking a nap and I noticed he had been asleep for an awful long time. We went in to wake him and he would not wake up, his breath was real shallow and he wouldn’t open his eyes. We rushed to the ER. They ran test after test, and no answers. For 4 hours he was unresponsive to pain or anything else. Finally he seemed to snap out of it. **The only conclusion they could come up with was that he had another seizure.** They kept us over night worrying that he was an epileptic.
Michael’s Story… cont’d

The next morning we had an EEG. All came back normal. With no real idea what had happened, they said it must have been a febrile seizure. But I knew he had no fever before or after his nap. In fact he had not been sick at all. So they sent us home. **For the next 2 years not much happened.** We had what we called small episodes where Michael would cry out in his sleep and seem to be very thirsty, but wouldn’t fully wake up. After some juice he would seem to be ok. This happened maybe a dozen times over the 2 years. Diabetes runs in my family, so I kept asking the Dr’s about that. They would check it out at his check up’s and nothing seemed to be out of the ordinary. These episodes wouldn’t even last long enough to get him to the Dr’s office. And I think sometimes they thought I was crazy.

**Then the episodes seemed to have stopped, until early one Sunday morning in February of 2003.** Michael came into my bedroom complaining of a stomachache. I told him to get into bed with me. He did and went back to sleep, every once in a while crying out. I got up and got him a drink. He sat up and drank a little and would try to go back to sleep. This went on for maybe an hour. Then it was time to get up for church, so I tried to wake him. **He would not wake up and I noticed he was cold and clammy.** I called our doctor (their office is open on Sunday mornings) and spoke with the nurse. She told us to take him to the ER. When we got there and went back the nurse took Michael’s temperature in his mouth. She said she would have to try another thermometer because that one seemed to be broken. She tried again. And again she said it was broken. She said sorry but I will have to do it rectally. When that reading came out the same as the other two she rushed us into another room. **We found out later his temperature was only 88 degrees. They said his body was shutting down.**

They ran tests, but nothing came back out of the ordinary. They drilled us, could he have gotten into anything. No, we said, again and again. They called in all sorts of Drs. Nobody had a clue. Finally after about 6 hours of tests (all coming back normal, and no more to run) they sent us up to PICU. Michael was more alert but his temp was still too low. **They said maybe he was Septic and continued to run more blood tests.** For 24 hours they had him on a warming device and had all his fluids running through a warmer. Finally his temp came back to normal.

The next evening they switched us to a regular room and we waited. Michael was seeing an Infectious Disease Dr and a Neurologist. On day 3 at the hospital our Neurologist came in and said he had a **preliminary result back on a condition called MCAD, but he really didn’t think that’s what Michael had because he was too old to be just now being diagnosed (comment from Deb – THIS is why we need to continue to EDUCATE those that don’t have correct info!).** But he would continue running tests.

They collected Michael’s urine for 24 hours and sent it off. **The Dr came back after the tests were run and said they came back positive, BUT he still didn’t believe it was what Michael had. He kept saying he was too old for this diagnosis.** They took more blood and said there was one more test to confirm or deny the MCAD. On Friday morning, **after 6 days in the hospital, it was confirmed. Michael had MCAD.** They had started him on Carnitor® on Tuesday, so his levels were up before we went home. We now know what Michael’s episodes were and why we went to the hospital the second time. **It has been over a year now since he was diagnosed and he is doing great.** So that’s Michael’s story with a very happy ending.

April Wiesner, Mom to Michael, 7yrs, MCAD
Ashley, 11yrs, Carrier
LawiesJr@aol.com

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**Deb’s Updated Mail and Email Address**

Please update your address book –
Mailing Address ~ 1559 New Garden Rd, 2E, Greensboro, NC 27410
Email address is deb@fodsupport.org
Backup address at fodgroup@triad.rr.com
Family Stories - Shawna’s Story, MCAD

My name is Melissa Cummings and my husband’s name is Douglas. Our little angelic girl, Shawna, came into our life on April 24, 2003. She was perfect in every way. The next morning the nurse brought Shawna in for a feeding and we were told that she had started to turn blue when they fed her and that we should just keep an eye on her. The nurse said Shawna was just trying to eat too quickly and that everything was fine. We went home the next day with a perfect bill of health for her and I. We had finally completed our family and were ecstatic.

One week later (I will remember that moment for the rest of my life) we received a phone call from her pediatrician. All I could think was that something has to be wrong. Pediatricians don’t just call to check up on newborns. He informed us that one of Shawna’s Newborn Screening blood tests had come back abnormal. The doctor didn’t have much information to offer at the time and said that someone from the Albany Medical Hospital was going to be calling to set up an appointment for further tests. I sat in shock and just thought what does this all mean. Cheryl Clow, a nurse who I now know was sent from heaven to care for my daughter and others like her, called one hour later and explained that there was a possibility that Shawna had something called MCAD, Medium Chain Acyl CoA Dehydrogenase Deficiency. Cheryl explained that MCAD is a genetic Metabolic Disorder. Cheryl needed to see her that Monday to run some tests and that Shawna needed to be fed every three hours.

We drove the four hours to Albany. Cheryl explained in detail that MCAD is a disorder that affects the production of a specific enzyme. The enzyme that breaks down stored fats in the body was not produced, so if she fasted (due to lack of eating, vomiting, diarrhea, etc) her body would not be able to break down stored fats. If that occurred she could have seizures if not treated right away. Cheryl explained to us that 80-90% of people affected with MCAD have it on a specific gene (the common mutation). When they had looked at Shawna’s DNA they did not find the gene there and that the new mandatory testing was giving some false positives. We were so sure that our little girl could not possibly have this but she needed to be retested ~ better to be safe then sorry. Cheryl was very clear that it was also a possibility that she did have it and that we should continue to feed her every three hours. Cheryl was an angel about answering all of my questions and calming a lot of my fears. She explained that the test would be sent to Baylor Institute in Texas and that it would take up to 2 weeks to get the results (sooner if it’s a positive test). If the tests came back positive we were going to need to have the other two kids tested. With all that said, the tests done, we traveled back home hoping that our little angel was not affected.

The whole time I wouldn’t let myself research it on the Internet because I kept thinking there is no way she had this rare disorder and that the test would come back negative. I also knew that if I started looking at things on the internet I was going to get the worst case scenarios and that it would drive me insane. Two weeks later I got the answer no parent wants to hear. I couldn’t take it all in and Cheryl knew this without me saying anything. She only gave me basics and asked that I call her the next day. After calming down I called my husband and could only cry. I now had to tell my son, he looked at me and said, “It’s okay mom we can take care of her because we love her.” I felt inept to deal with it all because I hadn’t let myself do the research. I didn’t know how or where to start. The next day I spoke with Cheryl and set up another appointment for even further testing. Shawna was tested to see if she had a Carnitine deficiency. The other two kids were tested to ensure that they were not affected by MCAD. With luck and God on our side, the carnitine test came back normal. It felt like a victory was won for our side. Then a couple weeks later, both of our other kids tests came back normal, that was a LARGE victory for us again.

I found the FOD (Fatty Oxidation Disorder) Family Support Group. I emailed them my name, address, phone number and email address. Thinking that I would get some small pamphlet in the mail I continued my research, sobbing at many of the stories. The very next day the phone rang and it was Deb Gould. I still have never met Deb Gould face to face but I know she is a dedicated person. She sent me all of the back newsletters and helped me to understand MCAD in a way that allowed me to be able to explain it to others. There are times when I get disgusted with people. They say things like “She looks normal you wouldn’t even know there was something wrong with her.” It is those times when I calmly state, “She is normal, she is special, and she eats special things.” I like my son’s response much better, “She is not any different from a vegetarian. They eat a special diet and so does she, that doesn’t make anything WRONG with her.” He is very protective of his little sisters.

There was a moment that changed my feelings from guilt and anger to hope and determination. I was on the phone with my brother-in-law when he said there are so many people out there that abuse, neglect, and don’t want their children and their children are healthy. Yet we love our children and God gave us a child with a defective gene. It was then that I remembered something my best friend had once said to me, “God wouldn’t give us more than we could handle.” He gave us Shawna because He knows we would do anything to protect her and keep her happy and healthy. My husband is a pillar of strength and encouragement, when I feel my lowest or start to feel the guilt he makes it all okay. I have never in my life known anyone like him. While we know that there will be ups and downs, we know we can make it through anything. We know this because we have an amazing support system. Our families are always there to help where they can no matter what the request. We always have an enormously supportive and caring group of friends who are so happy to help us with anything. I work as a teacher and chose to take some extra time from work so that Shawna would not be as exposed to illnesses. I was going to have to go without pay and possibly lose my benefits. I could call them coworkers but I call them special friends ~ they got together and raised money to help us to care for Shawna and the rest of the family. Our family and friends hold our hands and lend their shoulders and ears. My retired aunt even watches the two youngest so that I don’t have to worry about Shawna as much. My aunt has even dealt with one of Shawna’s episodes. Cheryl Clow is an astounding woman who is more dedicated to her job then any person I have ever met. Our friends and families are walking this road with us.

Melissa Cummings
mndcummings@yahoo.com
Our precious son Noah passed away after a long fight for life on March 23, 2004 at 10:21 pm. Noah was admitted to Wolfson Children's Hospital of Jacksonville Florida on February 13th, 2004. That Friday started out as a normal day. My husband William and I got the boys ready to go to the local store. Before we left I fed Noah, it was around 6:30 am. **Noah had a Metabolic Disorder called Tri-Functional Protein Deficiency.** It was very important for us to feed him every three hours no matter what. During that feeding Noah was his normal self. He took most of his feeding and he was very alert. He seemed fine during our trip to the store. As I was placing Noah and his car seat back into the car that is when I noticed his skin coloring. He was very pale and his skin was blotchy and very cold to the touch. And he started moaning while his eyes fluttered. I knew something was wrong.

The past two previous weeks we had two false alarms with Noah. **My first son Caleb died from TFP at the age of fourteen days. He went undiagnosed so I have never raised a TFP child before. Our geneticist told me to bring Noah in if I felt that he was acting differently or if I felt he was becoming ill. So we did have two previous trips to the doctors but everything turned out to be okay. Well that day there was something wrong. We rushed Noah to the hospital. On our way there I did call our doctor to inform him that we were taking Noah to the hospital. He then in turn notified the hospital of Noah's case and what the protocol was for his disorder.**

William dropped Noah and I off at the ER entrance. William had to take our other son, Caden, to the babysitter. We knew we would be at the hospital for a long time. **As soon as I walked in I told the staff who I was and what was going on. I also informed them that our doctor called them to tell them that we were on our way. I also handed them a copy of the protocol letter. I was ignored. I was told to sign in and please take a seat. I didn't take a seat. I stood in front of the desk stomping my foot. Finally after a few minutes the lady behind the desk took a look at Noah. She rushed us in back.** Once we were in the actual ER Noah's breathing and heart stopped. I watched them work on Noah for almost an hour before he was stable enough to be moved to the PICU floor. My son went through hell that day. **He survived! It was not his time to go.**

**Noah hung on for five and a half weeks. I was so blessed to have witnessed his first real smile a few days before he passed away.** During the finally weeks of Noah's life William and I were on an emotional roller coaster ride. We classified our days as a good or bad day for Noah. **We truly felt that Noah was going to pull through.** When I would visit Noah I would often read healing scriptures to him and play CD's for him. Noah loved listening to music. During this time I read the book Charlotte's Web to him. I had an eerie feeling that once I finished the book Noah would pass away. So for a few days I didn't read it to him. Instead I read from the Bible. But three days before he passed away I started reading it to him again. I finished it the day before he passed away. I will cherish that book forever.

On his last day of life he was started on a new study/protocol. **We will never know if it would have worked. Noah's heart was too weak from a second crisis that he had during his second week in the hospital.** He ended up with cardiomyopathy. Noah's little body couldn't take anymore. During his final week of life he ended up with two chest tubes, one upper lobe lung collapse and a bacterial infection.

When I arrived that night, I greeted Noah with a kiss on his forehead. That is when I noticed how pale and cold he was. He seemed fine during our trip to the store. He reminded me of Caleb the day he passed away. For the first hour of my visit he didn't open his eyes like he usually would once he heard my voice. I sat down in the chair next to his crib and started praying and reading healing scriptures while I played a CD with healing scriptures. Around 9:30 pm my friend Angel and her husband Eddie arrived. I just met Angel the week before and I met Eddie that night. **All three of us went to Noah's side. Once there, Noah opened his eyes and squeezed mine and Angel's fingers for about ten minutes. Then Noah closed his eyes and all the alarms went off. The staff worked on Noah for twenty minutes. They were about to take extreme measures. That is when I found my voice and told them to let Noah be with God and his big brother Caleb. My son was gone the moment he closed his eyes.**

William, Caden, and our close friends soon arrived. **We all spent the next four hours holding and loving Noah. We have all learned so much from Noah. Noah has personally given me so much. I now have closure with Caleb's passing...I was haunted by his passing for two and a half years. I was always thinking what if he would have been tested at birth...what if we would have placed him on a low fat diet. I now know that nothing could have been done for him. We did everything medically for Noah. The standard protocol does not work for the type of TFP my sons had. However it did prolong Noah's life for four months. For that I am grateful. We were able to have Noah at home with us for two months. I will cherish those four months forever. I love and miss you BIGGIE much Noah.**

Love, Mommy

Noah's Memorial Website: [http://www.geocities.com/n0ah_riley/NoahGrabow.html](http://www.geocities.com/n0ah_riley/NoahGrabow.html)

Firstly, let me say what an honor it is to work with families with Fatty Acid Oxidation defects. I really appreciate Deb Lee Gould giving me the opportunity to tell you about some of the research we presented this year at the American College of Medical Genetics and the Society for Inherited Metabolic Disorders Meetings in Orlando, Florida.

We recently looked, with great detail, at pregnancies that gave rise to a child with a fatty acid oxidation defect. We followed these pregnancies to see if they evolved into maternal liver disease (MLD). Generally speaking, MLD’s are a category of liver problems that occur during pregnancy such as Acute Fatty Liver of Pregnancy or HELLP syndrome (hemolysis, elevated liver enzymes, and low platelets). These liver conditions are very rare, and can cause the maternal liver to stop working properly, and if not recognized, can put both mom and baby at risk.

What we did was to look at the pregnancies of ALL FAODs, not just the ones with long chain defects. What we found was all categories of FAOD are at some risk of developing these problems; roughly 16% of all infants with FAOD had a pregnancy complicated by one of these maternal liver conditions. Although this may not seem like a large percentage, it is in comparison to the general population, where it occurs less than 1% of the time.

Although our research is far from done, we hope to look in more detail at the biology of why this happens. Perhaps the most important concern is for patient advocacy. When the rare occurrence of HELLP Syndrome and/or Acute Fatty Liver of Pregnancy occurs, the suspicion for a potential fatty acid oxidation defect should be immediately raised. This is especially critical in states that are not currently performing expanded newborn screening for FAODs. Good communication with the pediatric care provider should occur from the obstetric team to ensure that this follow up assessment of the newborn infant occurs.

The clinical collaborators at the different locations during the initial part of this project have been: Dr Vivian E. Shih, Massachusetts General Hospital Neurology Service, Chief of Metabolism Dr Harvey L. Levy, Children’s Hospital Boston, PKU Program Dr Louise E. Wilkins-Haug, Brigham and Women’s Hospital Maternal/Fetal Medicine Clinic, Dr Cecilia Larson, New England Newborn Screening Program.

We are very interested in checking the fatty acid intermediates in pregnant women who have had a child with a FAOD in the past. If you are interested in participating, or would like to know more, please contact me at:

**Laboratory/Research Office:** (617) 726-3884 (Massachusetts General Hospital Amino Acid Lab) Clinical Appointments: (617) 355-4695 (Children’s Hospital Boston)
I can also be reached via email at mfearing@partners.org

**Abstract:** Maternal Liver Diseases in the Pregnancies of Infants with the Spectrum of Fatty Acid Oxidation Defects Compared to Matched Population Controls

*Marsha K. Fearing, Harvey L. Levy, Louise E. Wilkins-Haug, Cecilia Larson, Vivian E. Shih

1-Massachusetts General Hospital Neurology Service, 2-Harvard Medical School Scholars in Clinical Science Program, 3-Children’s Hospital Boston 4-Brigham and Women’s Hospital, 5-New England Newborn Screening Program.

**Background:** Infant fatty acid oxidation defects (FAOD) are rare inborn errors of metabolism, occurring in 1:12,000 births. Common clinical features of long-chain FAOS include hypoglycemic and hypoketotic encephalopathy, hypotonia, cardiomyopathy, and sudden death. Increasingly, fetal long chain FAODs are associated with rare maternal pregnancy complications affecting the liver. These include acute fatty liver of pregnancy (AFLP); hemolysis, elevated liver enzymes, and low platelets (HELLP) syndrome; and pre-eclampsia evolving into HELLP syndrome. This relationship was initially described in the long chain FAODs, specifically Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHADD). A few isolated case reports emerged implicating the shorter chain defects, but the true prevalence among fetuses affected with the entire spectrum of FAOD is unknown. Maternal liver disease (MLD) in the general population has a low prevalence rate estimated at 0.1-1.5% for AFLP and 0.6-1.0% for HELLP syndrome. Given the paucity of these conditions, elucidating the true epidemiological relationship is difficult. The lack of literature comparing the entire spectrum of FAOD and pregnancy outcomes compared to the population led us to perform the following study.

**Method:** 50 case infants with fatty acid oxidation defects (FAOD) were identified in the New England region, either clinically or by expanded panel tandem mass spectrometry (MS/MS) newborn screening. A conditional logistic regression model was established, paring each infant affected with a FAOD to 25 unaffected controls for each case. Infants were matched by date of birth and hospital setting, generating a total of 1300 infant-mother pairs. Primary outcome analysis compared pregnancies affected by a fetal FAOD to controls for outcomes of MLD (AFLP, HELLP syndrome, and pre-eclampsia that evolved into HELLP syndrome). Isolated pre-eclampsia was not included in MLD. The pairs were phenotyped for secondary outcomes in antenatal, intrapartum and neonatal characteristics. Subgroup analysis was performed comparing the fetuses with long chain FAO defects to fetuses with medium/short chain FAOD defects. A Bonferroni correction was applied where appropriate to establish cutoffs for significance for the primary outcome.
Results: Case and control infants analyzed were similar with respect to mean gestational age (case = 38.2 ± 2.1 weeks; controls = 37.8 ± 3.6 weeks), mean birth weight (3264 ± 577 grams; 3308 ± 446 grams), and maternal age (30.2 ± 5 years; 28.4 ± 6 years) for the FAOD and control infants respectively. Primary outcome analysis revealed MLD occurred in 16% of all FAOD pregnancies (equally represented in long versus short-medium chain defects) compared to 0.88% in the general population (OR = 20.4; 95% CI = 7.8-53.2). Secondary analysis of isolated pre-eclampsia without hepatic involvement was not significantly different between the case (6%) and control pregnancies (6.1%); gestational diabetes mellitus was not significantly different in cases (10.0%) and controls (6.8%). However, post-natal results included elevated rates of clinical neonatal jaundice that was significantly higher in case versus control [FAOD 36%, control 8% (OR 6.25; CI = 3.42-11.4)] infants.

Of the fetuses affected with FAOD, 32% (n = 16) had a defined long chain defect, and 68% (n = 32) had a medium or short chain defect. There was no demographic difference among maternal age, but groups differed slightly on birthweight (long chain = 3.410 ± 0.52 Kg; short/medium chain = 2.940 ± 0.57 Kg). Subgroup analysis comparing fetal long chain FAOD to controls and fetal medium-long chain FAOD and maternal liver disease demonstrated significance in both groups (long chain FAOD OR = 50.0 p<0.001; short/medium chain FAOD OR = 12.3 p<0.001; Bonferroni correction p < 0.025).

Conclusions: MLD is significantly higher across the entire spectrum of FAOD demonstrating an 18.1 fold increase in the pregnancies of FAOD neonates compared to our control population. Notably, the prevalence is equally high in the pregnancies of infants with short and medium chain defects and not isolated to those infants with long chain FAOD. This implicates the entire spectrum of the acylcarnitine intermediates. Future studies, in considering pregnancies affected with fetal fatty acid oxidation defects, should examine the relationship of all FAOD with respect to the pathophysiology of the maternal liver disease for improved future health outcomes.

New FOD related articles – the articles listed in our last newsletter are now on our website in pdf form on the Medical Information page/Medical Articles

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Nutritional/Recipe Update

MCT Oil was purchased by Novartis Medical Health, Inc from Mead Johnson & Company effective February 13, 2004. MCT Oil is a modular source of MCT (Medium Chain Triglycerides) for patients unable to digest or absorb conventional fats. Consumers should consult with their physician on the use of this product. Refer to http://www.novartisnutrition.com/us/productDetail?id=593&source=summary for more information. NOT to be used by MCADers.

Some Families have found an alternate MCT Oil through Sound Nutrition. It is called original Thin Oil and is basically the same quality as the other Oil. It is less expensive by the case (12 16.7, 500ml bottles) - it costs about $7 a bottle. To order just call Sound Nutrition at 1-800-437-6863.

Banana Pudding Recipe

MCT Oil: 20 grams
Sugar: 125 grams
Bananas: 3 or 4 bananas, the browner they are, the sweeter the pudding tastes
Egg: 1 or Whites: 2
Flour: 250 grams
Baking powder: 1 teaspoon
Salt: 1/2 teaspoon
Nutmeg: 1/2 teaspoon

Mix Sugar and MCT oil until they look like a cream
Mashed bananas and add to the Sugar/MCT oil
Then add the egg or white eggs
Mix all with energy; In another bowl mix the flour with the Baking Powder, Salt, and Nutmeg
Then add to the banana mixture and blend well
Put the mixture into prepared (non-fat shortening & floured) baking pan, and bake in a preheated oven, low-medium temperature (165º) for about 45 minutes.

NOTE: An egg has 6 grams fat, so it is very little fat for a pudding
Q: Is there any Standardized Treatment for FODs?

A: These are young diseases, and until expanded Newborn Screening started picking up more and more we felt they were extremely rare, except MCAD. As Expanded Newborn Screening continues to grow we will have to revamp their incidence rates. We also have to adjust what populations we think are affected by FODs since we now have found them in Newborn Native Americans, Hispanics, and African Americans. In the meantime, because they are so rare, most Doctors have seen very few and there really is not consistent, agreement about how to care for them. The only published Protocol is from Ross Metabolics – The December 2002 Issue of the American Dietetic Association has an article by Rani Singh, RD, PhD at Emory giving an overview of her Survey of 123 Metabolic Centers across the country. Essentially the only things that were consistent were fasting avoidance and increased frequency of meals. I have hope - the NIH Consensus Conference for treatment of PKU occurred in 2001, 50 years after it was discovered. So, as some of you have identified we cannot say any Doctor is absolutely correct or incorrect in how they manage any given child with an FOD, so long as the child remains healthy and growing. Deb Gould works incredibly hard to update the list of the Doctors and Metabolic Centers who are staying up to date with all the new things we are learning almost daily, about how to treat these disorders.

Carnitine is a normal chemical in our bodies that serves to assist long chain fats entering the mitochondria and waste products leaving the mitochondria. Anytime there is a problem that affects mitochondrial function, more carnitine is used up removing waste products. In FODs, Carnitine deficiency is considered secondary to the Fat enzyme problem and theoretically once a diet is instituted, the Carnitine deficiency should correct. However, as many of you know, many children with FODs have problems with muscle pain and weakness, and sometimes changes in appetite or even sleep when carnitine is removed. It is a very individual response and good discussion to have with your Doctors. We do know that during fevers and severe illnesses that stop the child from eating, there is increased muscle breakdown that releases both proteins and fatty acids. For some children, the release of those fatty acids can make the child very sick. That is why we had such a lengthy conversation in Orlando (Oct 2002 conference) regarding doubling oral dosages or using IV Carnitor® during acute illnesses. You can also reference the various Emergency protocols on the FODSupport.org website.

Cornstarch: The only studies on the use of Cornstarch occurred in the early 1980s specific to Glycogen Storage diseases. Cornstarch, and many other complex carbohydrates, as well as brands of Cornstarch were tested for maintaining normal blood sugars in these children without triggering an Insulin response. Insulin causes increased Glycogen, not a desired thing in children with Glycogen Storage diseases. Theoretically, some Doctors feel Cornstarch may help maintain blood sugar levels in children with FODs who have PROVEN hypoglycemia (low blood sugar). As was presented by multiple Medical Experts in FOD management in Orlando (Drs Korson, Roe, Rinaldo, Winters), not all children with FODs have a blood sugar problem, and focusing heavily on blood sugars has the potential to prevent early treatment when muscle breakdown releases toxic fatty acids. In order for Cornstarch to hang around and protect children with FODs from low blood sugars over 6-8 hours, it must be given in cool, sugar-free liquids. Otherwise, Insulin is triggered and the Cornstarch is metabolized much faster than desired.

Diet ~ We now know that giving children too many simple carbohydrates contributes to obesity and Insulin resistance without improving metabolic stability long-term. Current diets for children, adolescents and adults focus on eating more complex carbohydrates during periods of health and using simple carbohydrates only during periods of illness and stress. These were all outlined as my article printed in the Jan 2003 newsletter. We also now know that the AMOUNT of fat children eat is not as important as the TYPE of fat and maintaining normal essential fatty acids. And most importantly, stopping lipolysis (fat breakdown from muscle) during acute illnesses is critically important, especially in long chain fatty acid defects, and may require IV D10 and IV Insulin. New things are being learned almost daily...keep sharing!

Lynne Metabolic NP LAWPNP@aol.com

Q: Our 19-yr-old has finally been approved for SSI and they want to see all our household income plus all the household bills for several months. Is my income going to negate any SSI she qualifies for as an adult? Will I be able to charge her rent? What chance do I have of being repaid for her extensive phone bill? We are supposed to bring bank info for her (but she doesn’t have one) and representative payee info to the next meeting. Isn’t getting it for kids different than getting it for an adult?
Question & Answer … cont’d

A: This is a tricky question and a tricky situation. Your question is why you have to show the Social Security office your financial information when it is your adult daughter who is receiving the money. When they ask to see your expenses they are looking at monthly house payment or rent plus utilities [electric, natural gas and water, but not telephone]. Social Security Income [SSI] payments can be used by a recipient in three ways: Sharing expenses, Paying rent, or Family assisting with expenses.

"Sharing expenses" means the recipient is paying an equal share of the rent and utilities as the other people living in the house. If the rent and utility payments for the household come to $2,000 per month, and 4 people live in the house, the recipient's "shared expense" is $500 per month. If only 2 people live in the home, then the recipient's "shared expense" is $1,000. This does not mean that the other residents must actually pay for their share. The recipient's 9-yr-old sister is not going to have to prove that she paid her share. It is just the formula Social Security uses to allocate the recipient's money for those expenses. The rub for shared expenses is that if the recipient's share of the shared expenses is greater than the actual amount the recipient receives in monthly support then the Social Security will not permit the recipient to justify payments under the "shared expenses" method. So if the recipient's "share" of the total expenses is $1,200 per month and the recipient's check is only for $600 per month, Social Security will say that the recipient is not truly sharing expenses and therefore cannot use the "shared expenses" approach to account for where the monthly income goes. One of the two methods below will have to be used instead. The "shared expenses" approach is the best method for the SSI recipient and the family. The recipient's monthly income via SSI will stay the same and the family will not have to report any of the recipient's money used to meet family rent and utilities expenses as income.

"Paying rent" means the recipient of SSI pays X dollars per month for rent and utilities. It does not matter how much the person from whom the recipient is renting actually pays for house payments and utilities. The Social Security will expect that the rent recipient will show the rent as income on their tax returns. Very simple and very basic. This method of accounting for the use of the SSI money does not harm or affect the amount of the recipient's monthly SSI check. It does however, the homeowner to report the "rent" amount as taxable income. The recipient pays the rent, whatever the amount, and the landlord reports the rent as income. The rent obviously cannot be more than the amount of the recipient's monthly income. Social Security does expect that the recipient will be using a portion of the monthly income for costs other than rent and utilities.

"Family assisting" is where the recipient lives with the family and the "shared expense" formula described above does not apply because the recipient's portion of the rent and utilities exceeds the recipient's monthly SSI check. The thought used to explain "family assisting" is that the recipient isn't paying their fair share of the expenses and the family is assisting the recipient by paying the part of the recipient's expenses that exceeds the monthly check amount. This way is the least favorable of the three accounting methods because the Social Security will consider the "family assisting" amount as income to the SSI recipient, and therefore will reduce or lower the amount of the recipient's monthly SSI check to account for the money received via "family assisting." This method will reduce the recipient's SSI check by as much as 50%.

So the recipient's entitled amount is determined by using one of the three categories above. The amount of a recipient's check is standard for each formula. Some states, like Michigan, supplement a recipient's SSI with a separate check. Those supplemental amounts vary from state to state and are keyed in amount to which of the three categories is used above. You may want to check with your local Arc or local consumer representative group to gain more precise information about supplemental amounts and Social Security practices in your state.

This answers the larger question you raise. Parental or other family income is not going to impact the amount of income the adult SSI recipient receives. Also, I do not believe you will be able to get retroactive payments for past telephone expenses. I suggest you contact your state Mental Health provider or family services agency to see if additional assistance is available to help pay for other expenses. For example, in Michigan we have an "Adult Home Help" program that pays home helpers [usually family members] a minimum of $333 per month or more for helping the SSI recipient with activities of daily living. Michigan SSI recipients automatically qualify for adult home help services although the amount paid is determined on an individual basis.

I hope these things help. Do consult with your local consumer representative group for further specific information for your state and situation.

Tricia
TriciaLuker@comcast.net

[Tricia obtained her expertise in special education, community supports, and parent advocacy through her direct efforts for her daughter, Jessica, who died a few years ago from an Organic Acidemia. She is a Program Director for various MI organizations. She and her husband, Calvin (an attorney), develop and provide legal, advocacy, and training services to those with disabilities and their families.]
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/ every now and then to update yourselves on what your state is adding to their NBS panel of tests. There was a series of interviews on the Today Show the last week in June ~ although it created awareness of expanded NBS the March of Dimes did NOT represent all of our Support Groups’ efforts to expand to the full panel of disorders ~ they only pushed their agenda of recommending 9 disorders. The Families that were interviewed (the Burkes, Kretzmanns, and Robin Haygood) however did a GREAT job of promoting ALL the disorders! I believe after the onslaught of angry and frustrated emails and calls to the MOD and the Today Show that they got the message that we were NOT going to sit by and only recommend a few disorders! So keep up the great work!

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®.

FOD Family Questionnaire

If you do NOT see your name on the Family List or on this issue’s Update, 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.

LCHAD Email Network

Valerie Fulton (Adam, LCHAD) is email networking many of our LCHAD Families, just as Gina is doing with VLCAD. If you’d like to become a part of her email network contact Valerie at vallchadmom@yahoo.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.
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

- Carolyn and Terence Finn
  - Emily - Birth Feb 13, 2002 Death April 3, 2004

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

- Lance and Dawn Goldsmith

- Deb and Don Gould

- Shelly and William Grabow
  - Noah - Birth Nov 18, 2003 Death March 23, 2004

- 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

- 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
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Welcome to new babies!

• Kevin Lee Nawn (MCAD), brother to Alex (MCAD), was born on December 14, 2003. Proud parents, Wendy and Chris, said he weighed in at 8 lbs 1oz and 20¾” long. Alex is a great big brother to Kevin!

• Alaina Grace Eick entered the world on July 21, 2004 at 2:16 pm. Her parents, Ronda and Bret, love their newest 7lbs14oz.baby! Sister, Alexis is 6 and brother, Myles (partial complex 1), is 3 — they’re enjoying Alaina as well.
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Condolences

We were very saddened to hear of the death of Noah Grubow (TFP) on March 23, 2004. He has joined his brother, Caleb (TFP) in heaven. Noah’s parents, Shelly and William, would like to thank everyone for all your prayers during Noah’s courageous fight to live. Our deepest thoughts and prayers go out to Shelly, William, Caden, and the entire Grubow Family at this time of saddened hearts.

We also would like to express our condolences to Cynthia Brown, whose 9-yr-old daughter, Miranda (Unclassified FOD) died March 21, 2004 after getting a virus.

Carolyn and Terence Finn are also mourning the death of their young daughter, Emily (CACT) on April 3, 2004. Emily experienced major complications due to cardiomyopathy.

All of our FOD children will ALWAYS be with us in our hearts!
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Love is always bestowed as a gift — freely, willingly, and without expectation…
We don’t love to be loved — we love to love

~ Leo Buscaglia
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Finn Thanksgiving Foundation, headed by the Finns in cooperation with an Episcopal church in Scarborough and the Fatty Oxidation Disorders (FOD) Family Support Group, will promote medical research and offer assistance to families facing these types of diseases.

Fundraising Information

If you’d like to participate in a fundraising project that would benefit the FOD Group, please read our last newsletter ~ Jan 2004 ~ we have several projects going on with Pampered Chef, Tupperware, and PartyLite Gifts.

Below is an article that was in THE JOURNAL NEWS (Original publication: June 7, 2004), written by Robert Marchant about a new Foundation created in memory of the daughter of one of our Families ~ they are raising funds that will promote medical research and offer assistance to families facing these types of diseases.

Foundation for an Ossining youngster

Like proud parents everywhere, Terence and Carolyn Finn speak with pride about the little girl with sandy brown hair and a gap in her front teeth who took center stage in their lives, littering their neat living room of art books, fresh flowers and Japanese prints with a toddler's toys.

Their eyes brighten when they talk about Emily's first canoe ride at Lake Mohonk, the meticulous care she took when deciding which cartoon to watch, the little jokes she made on the family couch.

But unlike other parents, the Finns must speak in the past tense about Emily, their only child.

A rare and severe form of a genetic metabolic disease carried her away from her comfortable Ossining townhouse at the age of 2 years and two months. The genetic malady -- carnitine acylcarnitine translocase deficiency, or CACT deficiency, part of a larger group of metabolic diseases that affect around 1 out of 15,000 births — made it impossible for Emily to produce an amino acid that processes fatty acids.

As a result, toxins built up inside her body and destroyed her vital organs. The disease finally caused her heart to stop April 3, when she went to sleep in a hospital room at New York Presbyterian Hospital in upper Manhattan with her parents at her side and never woke up.

Emily is still center stage in the Finn household through the creation of a memorial medical fund established in her name. The Emily Finn Thanksgiving Foundation, headed by the Finns in cooperation with an Episcopal church in Scarborough and the Fatty Oxidation Disorders (FOD) Family Support Group, will promote medical research and offer assistance to families facing these types of diseases.

The Finns said they felt obligated to honor Emily's memory by helping others who may face a similar predicament. "It's what they call an 'orphan' disease. There's no initiative to study it because it affects so few people," said Carolyn Finn, 40, who works in human resources for a Manhattan media firm. "She was our beautiful daughter, and through her short life, she really supplied a lot of medical knowledge, and a part of me would like to continue that element of Emily, helping doctors to understand her condition. Part of me wants to continue what Emily started."

Terence Finn, 32, an information technology specialist, said support from their family, church and community was crucial, and it was time to repay the kindness they had received. "We've been so blessed, we want to help others," he said. The fund, which has already received several thousand dollars in donations, could provide a few valuable insights into a condition that is little understood.

Dr. Wendy Chung, an assistant medical professor and the director of clinical genetics at Columbia University, said, "We learned a lot about the condition from Emily. Because we only come across children like Emily one or two times a year, it's hard to make great strides forward because the condition is so rare." Small advances into the treatment of the malady "could be incredibly useful," she noted, and medical progress was often propelled by committed group of activists. "A lot can be said for people pushing. Often times it's the squeaky wheel that pushes things forward faster," she said.

Since Emily could not eat normal food or baby formula, she subsisted on a carefully prepared mix of nutritional supplements and oils. The Finns are hoping better treatment options might be developed in the future, and they are also promoting public awareness of metabolic disorders and the need to detect them at birth, since the earlier the diagnosis is made, the better the outcome.

Newborn screening methods are set by individual states and vary widely across the country. Advanced screening methods for babies that can test for more than 40 metabolic diseases, which cost around $40 to $80, are not required in all states. New York requires that newborns be tested for eight metabolic illnesses, as does Connecticut. Some states like Oregon, Massachusetts and North Carolina require testing for more than 30, while Kentucky requires four, Utah only three.

The foundation for Emily Finn has its address listed at St. Mary's Church in Scarborough, where the Rev. Hillary Bercovici described Emily as "this little pixie of a kid, with a real force of character."

He said the foundation would be a fitting tribute. "Some kind of funding might increase the life span of another child," said Bercovici, a former paramedic. "I can't think of a better way to commemorate her. This kind of transformation, losing a child, is something really awful and to make it healing for others, it's a good way to handle grief. To transform grief into hope, it's a wonderful thing."

Send e-mail to Robert Marchant at rmarchan@thejournalnews.com
Kids Korner

Henry
(Unclassified FOD)
and Sam F

Caden and Carson R
(both GA 2 and
MCAD)

Brett (VLCAD), Morgan, and Jake

Julio Rivera-Smith,
Carnitine (MCAD)
Conference Pics

GA2 Families

Krystena Richards
and Gwen Abele
(GA2 moms)

Speaker Panel: Dr. Barb Marriage, Lynne Wolf, Dr. David Whiteman, and Dr. Mark Korson
Family & Professional Donations

**Family Donations:** Lezlie and Ron Meyer in honor of Hayden (LCHAD/TFP). Kathy Cramblitt in honor of her grandson. Pampered Chef donations. Christine and Shayne Aldana in honor of Lukas (MCAD). Kathy and Sam Walh in honor of their nephew, Jack Geiser (MCAD). Grandma Gerry Lee in memory of Kristen Marie Gould (Undiagnosed MCAD) and in honor of Kevin (MCAD). Thank you to all that have bought products from companies on the internet that support the iGive program of donating a certain percentage to Groups like ours.

**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 at this time.

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.

To laugh often and love much... to appreciate beauty, to find the best in others, to give one's self... this is to have succeeded.

~ Ralph Waldo Emerson~