Blessed New Year FOD Families and Professionals ~ we all hope that 2011 will be a better year for everyone no matter what challenges you face! As a Group, we are diving right in with various endeavors to expand awareness of FODs across the country and world [displaying our brochures etc at professional conferences], as well as expand our financial stability to take us into the future. I will discuss some of that in the Editorial on page 2.

Valerie Fulton (LCHAD) will be representing the FOD Group at the Feb 27 – Mar 2, 2011 Society for Inherited Metabolic Disorders (SIMD) Conference at the Asilomar Conference Center in Pacific Grove, CA. We are once again sharing a booth with OAA’s Director Carol Barton. I’m sure we will display at 1 or 2 other Conferences this year but nothing is planned so far.

As difficult as it’s been economically the last several years, we were blessed several times in 2010 with ‘Angels on Earth’\textsuperscript{2} who gave extremely generous donations that allowed us to offer scholarships to Families wanting to attend our Atlanta Conference. And because of continued Family and Professional support we will be able to do that once again for the 2012 Conference! We hope to be able to have Families from all over the world as we did in 2010 from Iceland and Australia, in addition to the States and Canada.

If anyone has suggestions for where our 2012 Conference should be please share with me ~ since we have been mainly in the Midwest and south for almost all of our Conferences, we are considering heading out west ~ IF we can find a committed Host and Premi...
Because of a generous anonymous professional donation, we have been given the opportunity to expand our financial stability. Additionally, an FOD Family member donated some stock to the Group in Dec 2010, which is something we have never accepted before. Now that we have a brokerage account, we will be able to establish further conservative investments so as to insure that the FOD Group will be able to continue once Dan, Mary (our webmaster and Board member), and I decide that it’s time to pass the torch on to new Families.

Because our Bylaws state that our 3 Board members are FOD Families, there will come a time that we will explore voting in a new Director, Treasurer, and Secretary. Each position will be responsible for various tasks and roles, which no doubt will expand and change over time. Not that I’m going anywhere soon, but we do need to think ahead – and that’s why we decided to invest in the future as far as the long-term accounts are considered. That way we will have funds available for our insurances, accounting fees, website and phone costs, Conference funding, and other various expenses that keep our Group afloat every year.

Also, because of these generous donations, I was able to open a Grief Consultation office 3 blocks from our home in order to offer pro bono grief support to local bereaved parents, and other family members. Donations are accepted but they are not required in order to receive support early on in their grief or years down the road. I have started to get referrals from ‘advertising’ in the local papers and from writing some articles for the paper and a regional magazine, as well as mailing my brochure/card to surrounding county facilities, churches, and other professionals. It not only makes them aware of my grief support practice, but it’s another way to create awareness of FODs. I will be speaking with the staff at a local perinatal office in February which will also be an avenue to share how carrying an FOD baby can impact the mother’s health etc. Sharing my personal and professional FOD and grief experiences has been something I’ve wanted to do face-to-face with parents for a very long time, but we just didn’t have the money to do that ~ NOW we do and I am so grateful! I enjoy talking with parents over the phone and through emails, but there’s a different and special kind of connection when it’s one-on-one in person. So THANK YOU to all that have donated to our Group over the years so we ALL can continue to provide NEEDED emotional and practical support to our new, as well as our ‘old’ FOD Families and Professionals! There will ALWAYS be a need ~ do NOT underestimate the POWER of SUPPORT!

Deb Lee Gould, MEd, Director
www.bereavedparent.com
Hello, we are the Huber family and this is our journey with raising three wonderful children with MCAD. We will forever be grateful for the education, ongoing research and newborn screening test. Our hearts go out to any family that has lost someone to MCAD. What we have learned is that MCAD is a livable disorder and our three children: Colton 10, Haley 6, and Carter 4, are proving it everyday. We would like to share the 10 lessons in life that MCAD has taught us. I am sure there will be many more as we watch these children grow.

10 Lessons in Life - MCAD

Lesson #1 We try to consistently stick to the routine (meals, snacks, and bed time schedules). Easier said then done sometimes since our children love sports and outdoor activities.

Lesson #2 We never leave the home without nutritional drinks and snacks.

Lesson #3 We have a protocol letter in all vehicles so we can travel and will be ready in case of emergencies.

Lesson #4 If our children are sick with even a cold it will take more nursing to get well.

Lesson #5 We hand packets of educational material about MCAD out to any caregiver and teacher along with an explanation from mom and dad.

Lesson #6 Our children take extra snacks and drinks to school. This helps to keep their energy up and stay healthy.

Lesson #7 If our children are sick with the flu or fever we take them to the Children’s Hospital for the best care.

Lesson #8 We live in a very warm climate, so plenty of liquids and air conditioning are a necessity.

Lesson #9 Newborn Screening and expanded Newborn Screening should be done at birth.

Lesson #10 Our children are very brave during metabolic crisis.

We did not always know that MCAD, “the silent disorder” was living among us. Of course, you never forget when you receive the phone call from the doctors to say that the screening came through indicating that MCAD is present. When our daughter Haley was born in 2004, we got that inevitable phone call from our family doctor. I was completely devastated seeing this normal child and wondering what the future meant for her. We soon were in a metabolic clinic and had a wonderful doctor that helped us through all of our questions. Through the Metabolic Specialist I realized that the MCAD prognosis for Haley was found in the expanded newborn screening test. After Haley’s diagnosis, we went on to test our oldest son, Colton, who was born in 2000 and the results came back with positive MCAD. He had missed the screening since the MCAD was not added to the expanded newborn screening test until early 2001. Our baby, Carter, was born in 2006, after testing in the hospital and follow up with the Metabolic Specialist we soon realized his results would come back positive MCAD. He had missed the screening since the MCAD was not added to the expanded newborn screening test until early 2001. Our baby, Carter, was born in 2006, after testing in the hospital and follow up with the Metabolic Specialist we soon realized his results would come back positive MCAD. Looking back through the years we had many close encounters and are very blessed for our children. Every time we have an episode it is scary and exhausting, but at the same time we are relieved for the knowledge to get them better. The Children’s Emergency Room is now very familiar with us and the staff is wonderful. Three MCAD children and only five visits to the emergency room, I consider our family very fortunate. We are so blessed that we now have some education and know what to do in a metabolic crisis. We frequently visit FODSupport.org to find any information that could help our family and we appreciate all that you do for so many children. Thank you FOD Support.org ~ we look forward to growing, sharing and helping other MCAD families.

Sincerely,
The Huber Family
Chad, Kelly, Colton, Haley and Carter
South Carolina
ckhuber@comcast.net
In 2006 I was blessed with a beautiful baby boy, Chance Wayne Morton. Chance was 6 pounds 5 ounces and 19 inches long. We came home a day after his birth with no problems, but very excited to be in our own bed. He was latching well and looked as healthy as can be.

At four-days-old he was hospitalized for projectile vomiting. Everything he ate started coming back up. It seemed like more came up than what went down. The Drs admitted him into Children’s and from there all the tests started. They took blood and hooked him up to IVs. They had him on a bunch of machines that I didn’t even know what they were for. The second day in the hospital after all blood work came back normal, the nurse made me leave his room, while they did a spinal tap on him. I remember just non-stop crying and I was so scared. I heard him screaming in there. I felt so lost for the both of us.

We had about 5 different Drs - pulmonology and cardiology were first to come in. They found out he had bradycardia and one of his lungs was a bit larger than the other, but they said none of that was the answer to what was going on with him. We had some answers, but not the final one. We met with 2 metabolic Drs who were trying to explain what FODs were. They thought this was the problem. After putting him on medicine through IVs and some formula instead breast milk, he started tolerating foods okay. There still was no clear answer. They took more blood and said it would be awhile for answers.

A few more days went by and we got to go home. Chance got to be at my graduation. That was the best present ever! We went home on medicines - Carnitor® and MCT oil, as well as reflux medicine. That didn’t help and it made things worse. We went through about 3 different formulas and one that he was on for a week, later made him start throwing up again. He was on a heart monitor at home and we had to follow-up with the 3 Drs. The heart Dr said Chance would be on the monitor for awhile and after that he would be put on the Holter monitor. The Pulmonologist wasn’t concerned with him after another x-ray and said he was just fine and his lungs looked healthy. The Metabolic Dr was the hardest - we still didn’t have an answer. They did a skin test which took few months to get back.

In the meantime while waiting for results, Chance started gaining weight, doing really well, and only had a few visits to the hospital for IVs and anti-vomiting medicines. One of the hardest things was finding pediatrician willing learn about this with us. None of them seemed willing to help.

After a few months we got the right pediatrician and finally an answer to everything. It was VLCADD. His Dr didn’t add any medicines, but we do follow-ups every 3 months. I learned how to monitor his blood sugars at home, what to look for when he has a crash and what illnesses I can treat at home and which ones he needs help with overcoming. I was sooo shocked and felt so alone.

Over the years, Chance has grown pretty well. He has issues gaining weight but is doing remarkable. He is now 4-yrs-old and in preschool. He loves to learn. Loves all his friends and teachers. He still has his bad days and is in the hospital more than other kids, but he knows he has to take his medicines to help him stay healthy. We still see a Dr every 3 months for check-ups. He definitely is my Angel. I have learned so much about life from him. He is energetic and loves singing, feeding ducks and being outside. He gets his muscle pains alot more, but nothing holds him back from being himself. He is a bright boy and I love him so much.

Jayme Morton
spritebabe72@yahoo.com
feeling when your child is in crisis or about to go in crisis and you just feel helpless because there isn’t anything you can do to help them. Especially when you were told it was “just a bug” and days/week later he is still vomiting and won’t eat.

Finally in August of 2009 I took him to the ER and they started the same ol’ spiel. I talked to the Dr and told him that this had been going on for a long time and if it was a bug how come no one else in the house is getting sick. Especially his younger brother because they share everything. So he ordered a urine screen and some blood work. Cameron’s amino acid levels and ketones were 400 times the normal range, and by this time his kidneys finally decided to quit. They started an IV, and told me that they needed to give him a special IV and that it had to come from another part of the hospital and would take some time for it to be “walked over.”

Once we were upstairs in a room, more blood was drawn. He did not urinate for three days after that first time in the ER. So I guess we were lucky or blessed depending on how you look at it that he was able to go at all and provide the sample. They could not figure out why—apparently however they kept saying he was so dehydrated that he just didn’t have enough fluid to be able to go. I seriously think that it was at that point if we hadn’t pressured them to do more tests and had taken him home again that night he would not be here today.

He was stabilized in the hospital and finally started eating, drinking and going potty and after our 7-day stay we were released and told that more blood work and tests were still pending and to follow-up with our regular Dr. Well, we went to the regular Dr and they referred him to genetics. But it was going to be 2—3 months before we could be seen.

One week to the day that he was released from the hospital, we were back in the ER at Southwest. He was having difficulty breathing, his stomach was sucking in so far you could see his ribs—he was really struggling. I called his Dr and she said he is probably having an asthma attack and to take him to the closest ER. They gave him breathing treatments and his oxygen level actually started falling instead of getting better. They put him on oxygen and had him transferred to Kosairs again.

Now they said he had asthma and gave us Qvar abuterol and a nebulizer. Dr Asamoah from genetics was able to come see him as an inpatient and do a skin biopsy. It didn’t grow for whatever reason so we had to make another appointment and have it repeated. By this time it’s already the end of September before we were able to see him again. Cameron was on levocarnitine 10ml 2x day and a low protein diet. Dr Asamoah said they suspected a metabolic disorder but had to do another skin biopsy. They sent it off to Philadelphia for testing. Everything came back normal, so they sent it to another lab. That lab’s results indicated that it was an FOD and that it was possibly CPT 1 or 2. Well it’s now March and they tell us they still don’t know but they’re getting closer.

About 3 weeks ago he called and told me that it was CPT 2. It’s amazing how bad news can be such a relief (LOL!), because we finally knew what he had and what we were dealing with. I had mentioned to family and friends once that I really didn’t care if they came and told me he was terminal, I just wanted to know what he had so we could move forward and deal with the outcome no matter what it would be. Thanks to the Group I know the questions I need to ask and the things I need to ask for such as an emergency protocol letter. I am so glad I found this group *side note thx Deb!!

He has not gone into crisis again but has been in and out of the ER and his regular Dr several times for breathing and respiratory infections.

Cameron now takes levocarnitine 10ml 2x a day, cornstarch 1tbls mixed with 8oz of milk at bedtime (because of low blood sugars in the morning and to maintain him at night), Qvar which is a steroid inhaler 2 puffs 2x day for his asthma, and of course has an abuterol rescue inhaler. The dietician sent us samples of MCT Oil in a powder. She said there are two types and that we should try the powder first and to restrict his fat to 28g a day—which I don’t understand because they say too much fat is bad for these kids but that they have to have some fat for development and growth—sounds like a no win situation to me (LOL).

His symptoms are vomiting, diarrhea, not urinating, low blood sugar (however this is not always the case because the abuterol will actually make his sugar high), breathing difficulty (not 100 percent convinced that this is asthma and not a result of CPT 2 because most times 80% the rescue inhaler and nebulizer do not help and we end up in the ER), lethargy, and just a normal day to day symptom is that he will say his legs/arms etc are hurting, and that is a sign that he needs to eat right away because he is probably out of energy and breaking down muscle protein for energy. He also has heat sensitivity. He can be outside for 3 mins and his face will be beet red like it’s sun-burned. He also goes to speech therapy.

If you ever have questions or just need someone to listen, do not hesitate to contact me. It’s always good to have a support system, especially someone that has “been there.”

Thanks for the snack suggestions over the Email List—they were all great and most things that Cameron so far is enjoying. The Gatorade was also a good tip. My wife sent some to school with Cameron and a note stating that if he goes outside or any physical activity that he is to drink it. I hate sending him to school because I know they are not doing what they are supposed to, but it’s send them to school or go to jail here in KY!

James, Dad to
Cameron, 6 years old, CPT 2
Jacob, 3 years old (going to ask to have him too)
jenkins1974@insightbb.com
My daughter Amberly has LCHAD. LCHAD is Long Chain 3-Hydroxy-Acyl-CoA Dehydrogenase (LCHAD) deficiency. I’m Amberly’s mother, Michal and my husband’s name is Abe. We live in British Columbia, Canada.

I realized I was pregnant with our fourth child in October 2007. Our oldest child, a girl, was five. We also had a four year old boy and a thirteen month old boy. This child would complete our family.

The beginning of my pregnancy was typical of my others. I was a little uncomfortable, but I never had to throw up and I was more tired than usual. I started gaining weight almost right away and it was soon obvious that I was pregnant. In the beginning of March I started feeling thirstier than usual and by the end of April, I was drinking about a gallon of water a day and had cravings for anything juicy. This made me start suspecting that I had gestational diabetes.

On Monday, May 5, I started having flu like symptoms, but no fever. My appetite was poor, I was very tired and my whole body was just uncomfortable. Sleep was almost nonexistent and my mouth was very dry, so I needed to drink water often which resulted in lots of bathroom breaks. By Friday, I was getting the shakes. I was cold from inside, but not on the outside. It would start in my back and I would just shiver. Warming up in the bathtub helped and it would go away for a while, but kept coming back. I craved water, chewed ice cubes and ate a lot of freezies. I started losing weight and that finally clued me in that I was not doing too well. I had never lost weight during pregnancy before and I had started to notice that my stomach wasn’t really growing anymore. None of these symptoms was that extreme, except my craving for water or anything cold and juicy.

On May 14, I had another appointment and I told my doctor of my thirstiness and that I thought I might have gestational diabetes. She told me we would do another diabetes test (I had already had one at six months) - I was now almost eight months pregnant. She also wanted me to have an ultrasound and blood work. The blood work was standard though, with all of my pregnancies at that stage, because of my negative blood. I went out of the Dr’s office hoping that we would find a reason for how I was feeling and feeling as if maybe the Dr was more concerned than she had let on. I was also a bit worried that maybe my baby wasn’t growing like she should. I tried to reassure myself that maybe this was all just due to it being so near the end of my pregnancy and maybe it was just a bit more stressful to my body because this was already my fourth one. Lots of people couldn’t sleep at the end of their pregnancy and were exhausted.

The next day, the nurse from the clinic called and told me that my blood work wasn’t normal and that I should come in for more blood work the next day and to come in right away if I started bleeding because blood work results showed that my platelets were low, but they needed to confirm if the numbers were correct. I started spotting that day and decided to take it easy to see if it would stop. I wasn’t worried about low platelets because I thought that I would be bruised up if my platelets were low and I wasn’t. (I thought maybe there had been a mistake with the blood results).

The next morning, I was still, spotting, so we decided to go to the hospital to have things checked out. We dropped the kids off at my parents and went to town, which is about an hour and 15 minutes from our place. We left home at 8 am and I started getting contractions at 8:30. They were between 6 and 8 minutes apart and not too hard, but enough that I knew, we would be having our baby that day. When we got to the hospital, they put the monitor on me to check my contractions and listened to the baby’s heartbeat. My contractions were about 5 minutes apart by then. They also did blood work right away because of my abnormal tests on Wednesday. They were a bit concerned with the baby’s heartbeat and that worried me, but then the Dr checked to see how far I was dilated and laughed. She was surprised, but she said I was fully dilated and that’s why the baby’s heartbeat was sounding funny - she was about to be born. The doctor said that she would quickly change so she could deliver my baby. I was surprised too. The contractions were mild and still only about five minutes apart. Then my contractions stopped, but I started having mild pushing pains. Amberly Faith Peters was born at 10:30 am. She was a small baby at 4.5 lbs and 16 inches long.

I had delivered a 9 pounder just 21 months ago, so the delivery was pretty easy and she seemed very, very tiny. She didn’t cry when she was born and I just got a quick look at her before a different Dr and nurses took her away to work on her. Meanwhile things weren’t going so well for me either. I was hemorrhaging and with my platelets so low that was a big concern. They also gave me a shot to try and stop the bleeding. And the bleeding just didn’t want to stop. My platelets dropped to 39. Normal platelet levels are between 160 or 260 and 360. Yikes! Amberly had breathing issues, acidosis and a low apgar score (I think her apgar score was 3 out of 10). She also had low blood sugar levels. The Drs had no clue as to why any of this was happening. I had had three previous healthy pregnancies and deliveries and three healthy children. What was going on?

They decided to send us to BC Women’s and Children’s Hospital in Vancouver. Amberly was stable at around 4pm and got sent out, but I wasn’t stable yet so she went alone, by air ambulance. They brought her to me to say goodbye, but I wasn’t feeling very good and hardly had the energy to look at her. That’s when I realized that perhaps I was dying. The Drs were doing all they could to help me, but it wasn’t getting better yet. I was receiving blood, lots of platelets and I don’t know what else. My liver wasn’t functioning properly and my kidneys were having a hard time as well. My Dr later told me that I went into DIC.

Family came, and I said what I thought might be my final words to my husband and parents. I had talked to my oldest child soon after Amberly was born, to tell them that they had a sister and that she was sick and that I was sick as well. They didn’t realize how bad it was because I had already been telling them that I was a bit sick at home because I just couldn’t do much at home already and I had talked about babies sometimes being sick too. We didn’t tell them how bad it actually was. Lots of people were praying and God, in his mercy, let us live. I started getting better late in the evening and at 2am they sent me and my husband to Vancouver to the same hospital where Amberly was. That was my first plane ride ever! My husband’s too. Who would have thought? After we got there, both Amberly and I started to get better. The Drs didn’t know what had happened, but were happy to see us on the mend. Amberly was on IV for the first two days and on antibiotics for 5 days because they thought maybe she had an infection, but tests came back negative.

The hospital we were at was about 15 hours away from our home if you were driving. It takes about 3.5 hours to get there if you take a plane, because we have to drive about an hour and a half to get to the nearest airport. There was talk about getting transferred to a hospital that was nearer to home, and we were getting excited to be closer to home so maybe someone would be able to bring our other children there to meet
their baby sister. It seemed as if Amberly was getting stronger every day, but on Thursday she started doing worse with feeding. We had been struggling with getting her to nurse and she had been doing pretty well on Wednesday, but after that she would latch on, but get tired after a few minutes. This really bothered me because all of my other children were breast fed and I really wanted that to go well for us again. It just seemed as if Amberly was not hungry enough yet. In hindsight, it makes sense because she was not getting the proper nutrition to build up energy. They were still talking about transferring us, but I think the Drs were a bit concerned that Amberly was not really keeping her body temperature warm enough on her own yet.

It seemed like they didn’t want to tell us much about what was going on with her because it was still a mystery as to why she had been sick at birth, but it seemed like it was probably connected to whatever I had had but the Drs couldn’t figure out why I had gotten sick either. All they knew was that I didn’t really fit into any category for a diagnosis after many, many tests, but my symptoms were very similar to the HELPP syndrome. I think maybe the only thing that didn’t fit was that I didn’t have high blood pressure. Both of us were getting better, medically anyway, every day. I was getting stronger and gaining energy and Amberly was gaining weight even though she had little interest in eating because she was getting fed by NG tube.

Friday afternoon while my sister-in-law, who had come to visit with her husband, and I were with Amberly, we received a disturbing visit. A team of metabolic specialists came to tell us that my daughter’s newborn screening test had come back positive for a metabolic disease called LCHAD. I had never even heard of LCHAD and had always thought that testing positive for any of the diseases they checked for with newborn screening was so rare, that I didn’t even consider that it would ever affect us. The Drs told us that they had to do another test to confirm the diagnosis, but in the meantime Amberly would be put on a special formula consisting of monagen, polycose, flax oil and breast milk, for LCHADers.

I wanted to believe it, but in the back of my mind I had a feeling that something was different with my daughter otherwise why should she have been sick? She was only a month early and my first daughter had been born 3.5 weeks early and had been 7 lbs and 2 ozs and not had any complications. My husband and I hung very tightly onto the words “have to confirm” yet, because that meant there was a small chance that she didn’t have it.

Meanwhile we had gotten papers on LCHAD to look over from the Drs and after getting a chance to read them, it was harder to cling to that hope that this was all a mistake. The stories sounded familiar. It made sense that I had been sick and we realized that it was likely that this was why we were still in the hospital.

Amberly did not do well on the new formula, she kept throwing up. We tried to get her to take it by bottle, but by noon on Saturday we decided to just feed her through NG tube. She kept throwing up though and on Monday, with the final confirmation of LCHAD, they changed her to another formula with portagen, polycose, breast milk and flax oil. The new formula worked a lot better and we were finally able to bottle feed her again.

Breast feeding was not an option anymore. After that came the teaching of LCHAD. What she could eat, mixing formula, sick day management and the dangers of fasting.

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It was pretty overwhelming at first, but gradually we began to understand what it would be like to have a child with an FOD. They taught us how to insert an NG tube and gave us the supplies we would need if she would need to be tube fed at home and gave us an emergency protocol letter to use for hospital emergency visits. Out children finally came to visit us with my parents when Amberly was two weeks old. She was gaining weight and starting to take the bottle quite well, but would still throw up occasionally. It felt good to have our family together again. We thought we might be able to go home with my parents and our children, but the Drs wanted Amberly to be on full bottle feeds with no NG for at least a couple of days before we went home. We were discharged a week later, when Amberly was three weeks old. She was 5 lbs and 13 ozs when we came home.

My husband and I were still a bit nervous about managing everything at home, but also very anxious to have our family together again.

I think the plane ride and hour and a half ride home by car was a bit much for Amberly though, because the next day we struggled with getting her to feed again and ended up having to put in the NG tube. This gave her a bit of a break and after a couple of days she was fully on bottle feeds again and started acting like a totally different baby. She was awake a lot more and started crying to be fed more. This was pretty exciting because for a while just after her diagnosis we wondered if she would ever want to eat!

Since then she has done very well, but we have had to put in the NG tube numerous times during an illness and have struggled with getting her to eat. She is a very picky eater and it is often a challenge to get her to eat enough and as often as she should. She is healthy and active so we try not to let the feeding issues bother us too much as we manage to get in enough to keep her healthy most times. She has had the stomach flu three times and has ended up in the hospital every time, but has suffered no lasting effects. Other than that we have been able to manage her illnesses at home.

She has been a blessing to us in many ways and I wouldn’t trade her for anything. It seems to me that each child you have will teach you something, but some more than others. I look forward to knowing her better every day and as we learn more about this disorder called LCHAD I believe it will seem less and less a disorder and more just a way of life.

Michal Peters
Canada
m2010peters@gmail.com

FOD Awareness ~ Booth Displays

Valerie Fulton (LCHAD mom to Adam) will be representing the FOD Group at the Society for Inherited Metabolic Disorders (SIMD) Conference in Pacific Grove, CA on Feb 27-Mar 2, 2011. Carol Barton of the OAA will share the booth with us.
Legal Corner:
EXPANDED NEWBORN SCREENING 2005 to 2009
MISSED OPPORTUNITIES -- POTENTIAL LEGAL CLAIMS

Over the past five years, it’s become clear that many children who should have had the benefit of expanded screening did not. By early 2005, the newborn screening expert group convened by the American College of Medical Genetics and HRSA’s Maternal and Child Health Bureau had issued a consensus report that all children should be screened for 29 “core disorders.”

On May 12, 2005, the American Academy of Pediatrics officially endorsed that report. Thus, by at least mid-2005, the standard of care was clear: If not already performed by state mandate, hospitals and pediatricians at a minimum must give parents information about expanded newborn screening for the 29 core disorders.

Tragically, there were many babies born after June 1, 2005, whose parents were not given this information. Many suffered severe injury due to late diagnosis and treatment -- injury that would have been prevented if the child had been properly screened.

Parents should know that these children potentially have very valuable legal claims that, among other things, could pay for all future medical and life care expenses.

Most states have long statutes of limitations for child claims, so it is likely that there is still time to bring suit on behalf of most children who were injured because they were not offered screening -- even babies born as far back as 2005.

Among the 29 core disorders are these 22 metabolic disorders:

- 3-MCC 3-Methylcrotonyl-CoA Carboxylase Deficiency
- ASA Argininosuccinate Aciduria
- BKT Beta-Ketothiolase Deficiency
- CBL A, B Methylmalonic Acidemia (Vitamin B12 Disorders)
- CIT I Citrullinemia Type I
- CUD Carnitine Uptake Defect/Carnitine Transporter Defect
- GA-1 Glutaric Acidemia Type 1
- HCY Homocystinuria
- HMG 3-Hydroxy 3 - Methylglutaric Aciduria
- IVA Isovaleric Acidemia
- LCHAD Long-chain L-3- Hydroxycyl-CoA Dehydrogenase Deficiency
- MCAD Medium-chain Acyl-CoA Dehydrogenase Deficiency
- MCD Multiple Carboxylase Deficiency
- MSUD Maple Syrup Urine Disease
- MUT Methylmalonic Acidemia
- PKU Phenylketonuria
- PROP Propionic Acidemia
- TFP Trifunctional Protein Deficiency
- TYR 1 Tyrosinemia Type 1
- VLCAD Very long-chain Acyl CoA Dehydrogenase Deficiency
- BIO Biotinidase Deficiency
- GALT Galactosemia

Two endocrine disorders also are covered:

- CH Congenital Hypothyroidism
- CAH Congenital Adrenal Hyperplasia
- Sickle cell anemia is covered

If your child suffered injury because of delay in diagnosis/treatment of one of these disorders and you were not offered expanded/supplemental newborn screening, you can contact attorney Chuck Hehmeyer (215-568-6190 or cphehmeyer@raynesmccarty.com). For many years, Chuck has specialized in handling these types of claims across the country. [Note from Deb: Chuck is a great friend to our Group—he has helped many of our Families and from other groups too!]
I just wanted to say to the Group that I feel that although we all go through a lot, there are several goods things that can come out of having to deal with disorders. My six-year-old daughter has become so understanding when it comes to people with disabilities because she can relate. She had a lemonade/bake sale (low-fat baked goods!) and she originally said that she wanted to raise money to buy an American Girl doll. After talking to her about charities, she decided that she wanted to donate all of the money to the FOD family support group. Latte Land donated coffee and she sat out in the cold weather two days in a row and ended up raising $85.00 for the support group. I could not be more proud of her! To make things even better, my best friend matched her first days donations of $50.00 so that she can go and buy her American Girl doll!!

Desereae Minor  kdminor@gmail.com
Overland Park, Ks  Brynn-unknown FOD possible VLCAD  Dane-4 same

My name is Melissa Cummings and I have four children. Their ages range from 18 years old to 5 years old. On April 24th 2003 my daughter Shawna was born. Through the Newborn Screening she was diagnosed with MCADD. She is now 7-years-old and is a beautiful, energetic and loving young lady. She is very active, she attends literacy class, K-Kids (they do charity work for the school and community), Swimming Lessons, Gymnastics and Tae Kwon Do all outside of the regular school day. Our youngest child, Zachary Richard, was diagnosed 2 years ago with a Mitochondrial Disorder called Pyruvate Dehydrogenase Deficiency.

In October of 2008 Shawna was granted a wish from the Make-A-Wish Foundation of Central New York. Shawna’s wish was to go to Disney and meet all of the Princesses and Tinkerbell. Our whole family went with her to Florida. We stayed at Give Kids the World Village, which is a wonderful place that made the whole family feel special. Make-A-Wish sent us to Sea World, Universal and Disney (both the Magic Castle and Animal Kingdom). She got to be a Princess for a week along with her sister. All of us got to feed dolphins, pet stingrays, ride some rides and meet all of the Disney characters. It made her feel so special and it was the first time in a very long time when my husband and I did not have to worry about every little detail, Make A Wish took care of everything.

This year for her birthday she asked if she could have a Walk-A-Thon to raise money for the Make-A-Wish Foundation so that another child could feel special and happy too. She wanted to be able to give that feeling of hope, strength and joy to another child. At first I was meeting road blocks so to get some help I called my best friend, Julie Johnson, who works at St. Lawrence University in the Center for Civic Engagement. Through lots of planning and the help of St. Lawrence University I was able to make Shawna’s dream come true. On October 9th we hosted the first annual Shawna’s Walk For Wishes.

Our goal was to raise $6000 which is approximately how much it costs to sponsor one wish. The local newspapers picked up on the stories and did numerous articles about MCADD and Shawna, it was nice to know that we were able to spread the word and educate others about FOD’s.

We are so fortunate because the day of the walk it was a beautiful October day with not a cloud in the sky. We had 50 volunteers ranging from family and friends to some of the Sororities at St. Lawrence University. We had approximately 200 walkers register to walk some were elementary kids who knew Shawna and some were local sports teams. We were ecstatic when we counted the money and it was just over $12,000 and we knew we would be able to sponsor 2 wishes!! We had far exceeded our goal.

When walkers registered we handed out food, t-shirts and most important an information packet about FOD Support. Thank you to Deb for all of the FOD brochures. I had a lot of people reading the pamphlet and asking me questions about MCADD and the FOD Support Group. We even met a woman whose child had just received a positive Newborn Screening for a FOD - it was an amazing feeling to meet another family and realizing just how far we have come since April of 2003. We are already planning a Softball Tournament, Golf Tournament and the 2nd Annual Shawna’s Walk For Wishes. We would love so much to have more FOD families there to join us. Please feel free to contact me at mndcumings@yahoo.com.

Melissa (NY)- Mito/MADD and MOM to
Zachary- 4 yr- Mito (PDHD)
Shawna- 7 yr- MCADD
Seira - 8 yr- unaffected
Vincent- 18 yr- unaffected
ATTENTION FOD FAMILIES ~ FUNDRAISING EFFORT AT ITS BEST!

I will donate 95% of my proceeds to FODSUPPORT toward our RESEARCH FUND!

Ever hear of Silpada Jewelry? No, well let me introduce you to the beautiful fine sterling silver jewelry we have to offer!

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Please go to my website and check out over 500 pieces in the new catalog! Then send me an email with your order and your phone number and I will contact you personally for payment information.

Every order is shipped directly to you for just $4.00!

LET’S REALLY MAKE THIS HAPPEN! THE OPPORTUNITIES ARE ENDLESS WITH SILPADA!

LAST YEAR I WAS ABLE TO DONATE OVER $2500!!!

Brenda Goodman (FOD mom)
Independent Representative for Silpada Jewelry
Fine Sterling Silver Jewelry
email: doublebny@aol.com
home: 216.292.5938
website: mysilpada.com/brenda.goodman

♥♥♥ Thank you, Jena, for sending out your annual Holiday cards to MANY of our FOD children and adults! ♥♥♥

Jena sent out lots of holiday cards as part of her own FOD project! She personally printed the child/adult’s name on the card and wrote a note and mom, Janet, did the graphics design and printing for her.

The cards were for Christmas, Hanukkah, Kwanza, Happy New Year, or a non-Santa version of a Christmas card. Each was personalized.

For those who have more than one child - each one received their own card! They sent cards to quite a few siblings last year and did so again. For those of you with tiny babies, they sent them cards too. The more, the merrier. This is Jena’s project, and she wants to share.

Last year cards were mailed to several countries, as well as many of the US states and Military duty station addresses, too. There is no such thing as too much mail when one is on active duty! Let’s look forward to Dec 2011!

Janet Longmore wordminder@yahoo.com
mother of Jena, almost 18, LCHAD/Mito and John, almost 19, ADD/Aspergers

Please remember our families in your thoughts and prayers throughout the year

[For entire list please refer to our Jan 2010 issue]

Mom, Jaimie, and siblings Mason and JaeLynn will love Parker forever...

‘Being deeply loved by someone gives you strength ~ loving someone deeply gives you courage’

~ Lao-Tzu
I cannot find a more fitting quote to describe our journey over the last two months. While I struggled to find enough time in my schedule for the holidays, it was Gareth who taught me the true meaning of holiday spirit.

As many of you know, Parker Hammon recently lost his heroic battle at the age of six. Parker fought strongly those six years, overcoming many odds and teaching many people about the seriousness of SCAD and other metabolic disorders. Parker also had a way of touching many of our lives, including Gareth’s. Gareth always referred to Parker as his “first FOD friend” and while the two never met, Gareth loved his friend Parker unconditionally.

Shortly after Parker’s passing, Jaimie, Parker’s Mother (and one of the strongest women I have ever met), had let it slip that she was going to sell some personal belongings to cover Parker’s funeral expenses. As I told my husband the story, I never realized that Gareth was listening from the other room. Overhearing this adult conversation, Gareth, unbeknownst to us, began formulating a plan.

At the bus stop the next morning, Gareth shared his plan with me. He wanted to sell Hot Chocolate to raise money for Parker’s funeral. As I choked back tears of pride, I also realized that what he wanted to do was next to impossible. We had busy schedules, schoolwork, and holiday preparations to fill our days. Thanking him for such a kind idea, I quickly brushed it from my mind. How would we sell hot chocolate anyway? “Nice idea, but too much work right now” I thought to myself.

But when Gareth came to me with his birthday money later that day and told me to send it to Jaimie for Parker’s funeral, I was overwhelmed. That night, having a “proud Mommy moment,” I posted Gareth’s comments and actions to my Facebook page. Almost immediately, friends and family were asking how they could help. Friends wanted to send money to buy Hot Chocolate, others wanted to send Gareth money to send to Jaimie. FOD families asked if they could send donations to the funeral home to help. Before we knew it, Gareth’s plan was taking shape before our eyes.

Within a week, friends and family had sent over $600 to the funeral home. Gareth raised another $50 on his own to send in. As I posted our thanks to my Facebook page, I had no idea that Gareth’s simple desire to help his friend would be like a pebble tossed into the water, causing a ripple effect that no one could ever imagine. Within hours of posting our thanks, a Facebook friend, who had not seen the original post, responded. Upon hearing the story, she began asking her friends to help. Before we knew it, an organization with ties to Newborn Screening advocacy got behind us and there were donations pouring in from around the world, as well as online auctions to raise money for funeral expenses.

We were witnessing a Christmas Miracle as the compassion of others streamed through the computer. Any faith in mankind that I had lost over the years was restored tenfold. And through it all, Gareth just kept asking “Is Jaimie going to be okay?”.

A week before Christmas, Gareth received a call from Jaimie. As she spoke to him, Gareth’s face lit up like nothing I’ve ever seen. Jaimie had called to tell Gareth that enough money had been raised to pay for the funeral in full. Gareth’s wish, the one I had tried to discourage in the beginning because “I” was too busy,” had come true. He had helped Parker’s Mommy.

If the story ended there, it would be the perfect ending. But there was more to come. Not only did Gareth help to share love and compassion during the holiday season, he also shared a story – the story of Parker. That, in itself, was worth so much more than the donations received. By sharing Parker’s story, it has raised awareness of these types of disorders. It has allowed other families in similar situations to not feel alone. It has provided a connection for many that goes far beyond a simple wish to sell Hot Chocolate.

And Gareth doesn’t want to stop there. He wants to continue to raise money for families like Parker’s. While we are still in the planning stages of “Butterfly Angels,” we are hoping to have a fund set up with a local bank or organization shortly so that Gareth can continue to raise money for those in need, all while raising awareness of FOD disorders.

I’ve heard it said that Christmas should be seen through the eyes of a child. This year, I was blessed to see Christmas through the eyes of an innocent 8-year-old, who showed me through his innocence, persistence, and compassion, the true meaning of friendship and the spirit of giving this holiday season. Thank you, Gareth, for a lesson well taught.

Kim DiPaolo, proud mom to Gareth (MCAD)
garethsmommy@yahoo.com
What causes Retinopathy in Long-chain 3-hydroxyacyl-CoA Dehydrogenase and Mitochondrial Trifunctional Protein Deficiencies?

Autumn Fletcher, Melanie B. Gillingham, Cary O. Harding
Oregon Health & Science University
Portland, Oregon

A. Introduction

Mitochondrial Trifunctional Protein (TFP) and Long-chain 3-hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency are disorders of fatty acid oxidation (FAO) caused by different mutations in the same protein (TFP) [1]. FAO is important at all times for energy production in the heart and muscle, and is critical for all tissues in the body during times of fasting, illness and prolonged exercise [1]. Although deficiencies in many enzymes in the FAO pathway are described, only LCHAD or TFP deficiency can lead to chorioretinopathy and vision loss.

Chorioretinopathy is defined as any disease involving the choroid, the area of the eye containing the blood vessels, and the retina, the light-sensing portion of the eye, responsible for vision. There is no known reason that FAO disorders should cause retinopathy because the retina is believed to use glucose, not fat, for energy [2]. However FAO proteins, including TFP, are found in retinal cells [3]. The fact that the proteins for FAO are present in these cells leads us to question if there is a need for them in the eye. However, we cannot assume that FAO is necessary in the retina because only patients with LCHAD or TFP deficiency and not patients with other FAO disorders suffer from this retinopathy.

TFP is an enzyme with three activities that are essential for the generation of energy from long-chain fats. Trifunctional Protein is made up two different peptides called subunits. These different subunits are designated alpha and beta subunits [1]. A cartoon of TFP with subunits and their activities labeled is shown in Figure 1.

The alpha (a) subunit contains two of the three enzymatic activities, hydratase and dehydrogenase activities. The beta (b) subunit contains the thiolase activity. Figure 2 outlines the pathway that uses these activities for energy production, and illustrates the build up of byproducts in isolated loss of the dehydrogenase activity, LCHAD deficiency. Two separate genes provide instruction for the cell to create these protein subunits.

For the purposes of this review, a gene is a piece of DNA that codes for one protein or part of a protein. The building blocks of DNA are called nucleotides and sequences of genes are called alleles. Everyone carries two alleles of each gene in the genome; one comes from your mom and one from your dad. Typically, a change in the sequence of a gene, called a mutation, can result in a non-functional protein. The building blocks of proteins are amino acids. There are different types of mutations that make changes in the protein created, but many mutations result in a change in the amino acids. Some mutations lead to a stop instruction that creates a short protein that often gets degraded in the cell.

Figure 1. A cartoon illustration of TFP with subunits and activities labeled.

Figure 2. Pathway of long-chain fatty acid oxidation, with blocked dehydrogenase step and metabolite build up illustrated.
Deficiencies in FAO are inherited in a recessive manner. Recessive inheritance means that to have clinical symptoms of LCHADD or TFPD, the patient inherited two mutations; one from mom and one from dad. To have a child with an FAO deficiency the parents must both carry one mutation that codes for a non-functional subunit; in genetic terms these parents are carriers. It also means that both parents must pass that mutation to the child. Statistically this will happen 1 in 4 times that carrier parents have a child.

The gene for the α-subunit of TFP is abbreviated HADHA ([http://www.ncbi.nlm.nih.gov/omim](http://www.ncbi.nlm.nih.gov/omim) OMIM # 600890), and the β-subunit gene is HADHB (OMIM # 143450). Many patients with LCHAD deficiency have two copies of a common mutation in HADHA, designated c.1528G>C, that leads to selective loss of the LCHAD activity of the protein. In other cases the change in DNA results in a truncated, or shortened protein because of a stop instruction being substituted for the next amino acid. Patients with defects in the β-subunit of TFP, due to two alleles of HADHB leading to a non-functional or unstable protein, tend to have a slightly different set of complications than patients with defects in HADHA. The progression of retinopathy tends to be much slower with little vision loss in patients with deficiencies in all three activities of TFP, as compared to patients with isolated loss of the LCHAD activity [4].

B. What is LCHADD/TFPD retinopathy?

The disorder now called LCHAD deficiency was initially described in 1989, and by 1992 at least 10 patients were identified [5, 6]. Trifunctional protein was first discovered in 1992, coincidentally the same year two patients were described that had compound deficiency of all three enzymatic activities called TFP deficiency today [7-11]. In 1996 it was first suggested that pigmentary retinopathy is a diagnostic sign of LCHADD in the presence of an unknown FAO defect [12].

In a 1997 a retrospective study of 19 patients that carry two copies of the common mutation was performed. It was observed that as early as four months of age children with LCHADD begin to develop dark spots in the retina or pigment clumping [13]. In the early stages of retinopathy, retinal function appears to be normal although the center of the eye may appear to have less pigment. As the retinopathy progresses there is more clumping of pigment in the retina but age-appropriate performance and visual acuity. Later, the pigment clumping begins to disappear in the center and move outward. The eye blood vessels deteriorate. Patients may have loss of night vision, followed by loss of color vision. In late stages all central vision is lost [13].

In 2002, Tyni et al suggested that TFP is expressed in the retina, despite the commonly accepted fact that the retina uses glucose for energy [3]. Then, in 2004, a pathology report showed that retinal pigment epithelial cells (RPE cells) die before the cells that absorb and process light, known as photoreceptors [14]. RPE cells are a supporting cell type that provide nutrients to the photoreceptor (PR) cells, also known as rods and cones [2]. No experiments have shown why RPE cells die in retinopathy of LCHAD and TFP deficiency.

C. Do all patients with mutations in HADHA or HADHB get choriorioretinopathy?

Mutations in HADHA and HADHB have varying effects on folding of the protein subunits into their correct formation [15]. The common mutation in TFP, c.1528G>C, causes isolated LCHAD deficiency [16, 17]. As of November 2010, there are 32 published mutations in HADHA, and 29 published mutations in HADHB. Function of the enzyme complex requires folding and assembly of subunits to occur correctly [18]. Various mutations in both the α- and β-subunits have been reported to destabilize the protein complex, leading to a decrease in all three enzyme activities and lower total protein levels in patient cells carrying these mutations. The common mutation does not have this effect; levels of protein in cells with two copies of the common mutation are comparable to levels in control cells [15].

Interestingly, the common mutation in TFP, c.1528G>C in the HADHA gene, is very prevalent in patients of European descent but is relatively absent in Asian populations [13, 19-21]. In a recent Chinese study using 1200 blood samples from individuals of Han descent, not one carrier of the c.1528G>C allele was found [22]. Genotyping of patients in Japan and Korea reveal mainly HADHB mutations [19, 23]. In contrast, a study in Finland estimates the carrier rate of the common mutation in the Finnish population to be 1 person in 240 (1:240) [28]. In a similar study in Poland the carrier rate was recently estimated at 1:189 overall; one particular region had a carrier frequency estimate of 1:73 [29]. Other countries have relatively high carrier frequencies of the common mutation; estimates put the carrier rate of this allele in the Netherlands at 1:680 [30]. The carrier rate in the United States has never been investigated to our knowledge. Only 5 cases of LCHADD or TFPD have been found through newborn screening in the last 5 years in the newborn screening program that screens all children born in the states of Oregon, Idaho, Alaska, Nevada, and Hawaii (personal communication, C. Harding).

D. Are OHACs and OHFAs involved in vision loss?

Published reports of retinopathy in LCHADD and TFPD were not found in any study containing only cases from Asia. In contrast, the prevalence of retinopathy varies from 20% to >50% of patients in published LCHADD and TFPD studies from Europe and the U.S. [26, 31]. This correlation suggests that the common mutation is associated with retinopathy in LCHADD/TFPD, although it falls short of being convincing. More research is needed to confirm a connection between the common mutation and retinopathy in these deficiencies.

The β-oxidation pathway is shown in Figure 1, with the blocked step in isolated LCHAD deficiency illustrated. When long-chain fatty acids are oxidized for fuel in the cells of patients with LCHADD, the hydratase creates a 3-hydroxy fatty acid, but further oxidation by the dehydrogenase is blocked, resulting in the build up of 3-hydroxyacylcarnitines (3-OHACs) and 3-hydroxy fatty acids (3-OHFs) in patient blood (Figure 1) [32]. Generally, increased 3-OHFs and 3-OHACs are seen in blood of patients with LCHAD deficiency and to a lesser extent in patients diagnosed with TFP deficiency [33, 34]. These 3-OHFs and 3-OHACs increase with metabolic crisis, prolonged fasting or prolonged exercise when the body attempts to use long-chain fatty acids for energy [35]. When patients with LCHAD or TFP deficiency are healthy, eat regular meals, and consume a low-fat diet these levels fall and the progression of the retinopathy is slowed [4]. In addition, early diagnosis, treatment and decreasing the number of metabolic crises are associated with slower progression of retinopathy [36]. A direct mechanism connecting metabolites with retinopathy is not yet clear, but there is a strong connection between high 3-OHAC concentration in blood and decreased retinal function in a prospective study of patients with LCHAD and TFP deficiency followed over 5 years. This study concluded that lowering 3-OHAC byproducts with diet treatment slows the progression of retinopathy [4].
E. Conclusions and future directions

The current evidence supports the theory that progression of retinopathy correlates with the presence of the common mutation. Patients with at least one c.1528G>C allele can have high 3-OHACs when they are in metabolic crisis or when they eat a diet with excess long-chain fat. These patients also commonly develop chorioretinopathy. In contrast, TFP-deficient individuals who lack the c.1528G>C mutation have lower byproduct levels and do not as often progress to vision loss, although some retinal changes are still evident.

Multiple theories have been put forward in the past twenty years regarding the cause of LCHAD and TFP deficiency retinopathy. One theory suggests that the number of metabolic decompensations and/or hospitalizations for hypoketotic hypoglycemia the patient experienced is positively correlated with vision loss [32]. We would argue that during metabolic crises, levels of byproducts are likely high. Therefore the retinopathy seen in patients that have undergone multiple hospitalizations could also be due to episodes of severe byproduct build up. Patients that carry at least one c.1528G>C allele that follow the recommended diet and have fewer metabolic decompensations also have lower 3-OHACs and slower progression of their retinopathy. Studies are in progress to tease out whether low blood glucose episodes or higher levels of byproducts can lead to retinal cell death.

We are currently creating retinal cell models of TFP and LCHAD deficiency; molecular studies to determine the mechanism of cell death in the retina are ongoing. RPE cells engineered with specific genotypes will create supporting or refuting evidence that genotype and or byproducts play a role in the progression of retinopathy in children with TFP and LCHAD deficiency. Understanding what causes retinal cell death is the first step towards developing an effective treatment to prevent vision loss.

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References


**Medical Update...cont’d**

We oppose the IEP provision. We also oppose the passage of any bill containing the IEP provision. We oppose the provision for the following reasons:

- **Seclusion and restraint are not educational practices, strategies or methodologies.** At best, they are emergency interventions.
- **Seclusion and restraint plans are not behavior plans.**
- **Seclusion and restraint plans are not discipline plans; they are punishment plans.**
- **Placing seclusion and restraint plans into IEPs is tantamount to declaring them “programs” within the meaning of special education law.**
- **As S.3895, Finding 4 states, “seclusion and physical restraint are not therapeutic.”**
- **As S.3895, Finding 4 also states, “[seclusion and physical restraint] are not effective means to calm or teach children and may have an opposite effect while simultaneously decreasing a child’s ability to learn.”**
- **The use of seclusion and restraint as educational practices has been repudiated in therapeutic institutions including hospitals, psychiatric facilities and other residential settings for people who have challenging behaviors, even though these therapeutic institutions are staffed with medical and other highly trained professionals, and even though the physical environment itself is better suited to applying seclusion and restraint than are our nation’s schools.**
- **A student’s IEP or educational plan is not a place to insert a seclusion/restraint plan that may result in serious injury or death to the student or to the school personnel who are tasked with implementing such a plan.**
- **The current IEP process includes provisions designed to address student behavior challenges, including the use of functional behavior assessments [FBAs] and behavior intervention plans [BIPs]. These provisions have been in place since 1997 and were strengthened by Congress in 2004. Including seclusion/restraint plans in IEPs directly imperils all of the work that**

**Educational Advocacy Corner:**

**IS A BIRD IN THE HAND REALLY WORTH TWO IN A BUSH?**

http://www.ourchildrenleftbehind.com/
Advocacy Corner...cont’d

Congress, schools and parents have done to encourage the use of behavior plans.

- IEPs are the “contract” between school districts and parents that define their child’s educational expectations. Seclusion/restraint plans in IEPs, like speech therapy, physical therapy, testing accommodations, assistive technology, classroom placement, extended school year, etc., create an expectation of services to be provided. With this model, districts will naturally favor the use of seclusion/restraint plan over the development of a positive behavior support plan as the preferred method for reacting to challenging behavior.
- No effective mechanism exists for parents to challenge the inclusion of seclusion/restraint plans in a student’s individual safety plan, educational plan or behavior plan. Due process mechanisms existing for IEPs are costly, cumbersome and time-consuming and produce additional stress for already stressed out students, parents and families.
- Including seclusion/restraint plans in IEPs will increase, rather than reduce, the use of seclusion and restraint.
- Although parents are members of the IEP team and therefore would be able to participate in the decision whether or not to insert a seclusion/restraint program into the IEP, there is no provision explicitly requiring that parents participate as FULL MEMBERS in any group or activity creating the seclusion/restraint plan itself. This also currently is true with respect to the conducting of FBAs as well as the development of a BIP. Parents will be asked to agree to seclusion/restraint plans that have been developed without them.

The points we list above are only a partial list of reasons why Our Children Left Behind [OCLB], self-advocates and parents oppose the inclusion of IEP seclusion/restraint plans in IEPs.

Parents also are concerned about the IEP proposal because of its evident reliance upon IDEA’s due process provisions that regulate how parents can challenge elements of an IEP. IDEA’s due process mechanism is neither fair nor effective for parents. While parents are forced to pay their own legal costs for due process hearings, districts have access to insurance pools that may pay $100,000 or more per case for a districts legal fees and costs associated with the hearing. That insurance is provided to the districts for free by their insurance carriers as a value added benefit. District due process costs not covered by insurance are paid for by tax payers. Under these circumstances most parents have no reasonable possibility of challenging a district’s decision, over the parent’s objection, to insert seclusion/restraint plan into their child’s IEP.

The IEP seclusion and restraint provision has generated controversy. Some organizations have taken the position that they can support the bill even with the IEP provision intact, because the other provisions in the bill strengthen protections for students throughout the United States, including in states that currently have no laws or policies regulating the use of seclusion/restraint.

Other organizations are taking the position that the IEP provision is fatal to the bill. They cannot accept legislation that permits the planned use of seclusion/restraint by including it in IEPs and other similar plans. OCLB supports this position.

There is no doubt that seclusion/restraint are non-therapeutic interventions that create a significant likelihood that those who are being secluded or restrained – our nation’s children, often our children with significant disabilities – and those who seclude or restrain them will be hurt or killed. Seclusion/restraint are dangerous and should not be used on our nation’s children, period.

We are sensitive to the fact that S.3895 provides protection throughout the United States. If not for the IEP provision, OCLB would strongly support this bill. But for the reasons stated above, we believe the IEP provision, creates a greater likelihood that students will be injured or killed as a direct result of the inclusion of seclusion/restraint plans in IEPs.

More importantly, this provision will provide a strong legal basis to condone and in fact promote the use of dangerous practices that – according to Congress itself – have no educational or therapeutic value. We cannot agree that the benefit of federal regulation of seclusion/restraint in our schools outweighs the potentially fatal cost of legitimizing the use of seclusion/restraint use in our children’s IEPs.

Compromising in order to produce a bad bill that emboldens the use of seclusion and restraint – the Bird in the Hand – is not acceptable to us. There will be no time to go back and “fix things.” The damage will be done and our children will be the worse for it.

The graphic prepared by OCLB self-advocate, Michael Igafo-Te’o, summarizes our feelings in one word. “Ouch!” Seclusion and restraint never should be sanctioned as part of educational programming. As Michael, who has been secluded and restrained, clearly understands, seclusion and restraint hurts and kills children.

Tricia and Calvin Luker
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Tricia Luker tluker@ralawcenter.com

The OCLB Team
Sandy Strassman-Alperstein, Deidre Hammon, Jackie Igafo-Te’o Shari Krishnan, and Calvin and Tricia Luker, along with self advocates Benji Alperstein, Daniel Alperstein, Brianna Hammon, Michael Igafo-Te’o, and Nicholas Krishnan
Join us on the evening of April 12, 2011, and all day on April 13, 2011, for a discussion by top researchers, medical professionals and families living with FAODs. Topics will include current research, potential future treatments, as well as day to day practical choices and living with metabolic disorders.

Our mission for this meeting is to provide a forum for sharing questions, concerns, ideas and solutions in managing FAODs, as well as educating ourselves on the latest research in screening, diagnosis and treatment.

Although this meeting is designed primarily for those with FAODs and their families and friends, interested healthcare professionals seeking more information about these disorders are always welcome.

Speakers:

Cary O. Harding, MD, OTSU Doernbecher Children’s Hospital, Associate Professor, Oregon Health and Science University School of Medicine,

Margaretta Seasmore, MD, Director, Genetic Consultation Services, and Director, Biochemical Disease Detection Laboratory, Yale-New Haven Hospital, Professor, Yale University School of Medicine.

Lynn A. Wolfe, MS, FNP, ACNP, BC, Undiagnosed Disease Program, National Genome Research Institute, NIH, National Institutes of Health, Office of the Clinical Director.

SCHEDULE

April 12, 2011
Reception
Meet the Speakers
6-8 pm

April 13, 2011
8:30-9:00 AM Registration & Coffee
9:00-10:00 AM Keynote Address: Dr. Cary O. Harding, “What’s New in Research and Treatment of FAODs”
10:00-10:15 AM Break
10:15-11:15 AM Roundtable Discussion: Dr. Harding, Dr. Margretta Seasmore, “Developments in Practical Care for FAODs”
11:15-12:15 PM Lunch
12:15-1:30 PM Heart Healthy Lunch Provided
1:30-2:30 PM Roundtable Discussion: Dr. Harding, Dr. Seasmore, Ms. Wolfe, “Practical Issues”
2:30-2:45 PM Break
2:45-4:00 PM Planning next meeting; evaluation & Close

Program Cost: $40.00 per person (includes lunch)

A limited number of rooms have been reserved at the Holiday Inn at a reduced rate. Please call (203) 336-7700 before March 1, 2011 and mention “Team Ella” to reserve a room at the discounted rate.

Roundtable discussions will include pediatric and genetic healthcare professionals experienced in caring for patients with metabolic disorders such as FAODs.

A supervised children’s room is available during the day on April 13, for those traveling with children. A Heart-Healthy lunch is provided with the cost of the program.

TEAM ELLA includes family and friends of Ella, 6 years old with VLCADD, diagnosed through newborn screening.

Registration

Name: ________________________________
Address: ______________________________
Phone/Email: __________________________

No. Attending Meeting, April 13: _______ No. Attending Reception April 12: _______

☐ Traveling with child needing

No. of Children: _______ Age(s): _______

Space in Children’s Room

Gender(s): ________________________

Program Fee: $40.00 per person (includes lunch) Total Enclosed: ____________

Mail Form and Payment to Team Ella, 22 Woodland Drive, Clinton, CT 06413

Directions to Holiday Inn: north of Old Mill Road (exit 12 off I95) to Route 35 North, to the Black Rock Turnpike North. Continue on the Bridgeport Avenue Frontage Road to the Holiday Inn (on the left).
The primary goal of this project is to improve access to psychological and developmental evaluations for patients with fatty acid oxidation disorders (FAODs). The second major goal is to improve education about the developmental and psychological issues in fatty acid oxidation disorders. This project will demonstrate a method for developmental and neuropsychological screening for children with FAODs detected by newborn screening, describe the developmental course of children with these disorders, and disseminate information learned about the screening method and likely outcomes. This is important because the fatty acid oxidation disorders are the most frequently identified conditions detected by expanded newborn screening and, at the same time, some of the least understood with respect to long-term outcomes. This project will be a collaborative effort between Children’s Hospital Boston, the New England Consortium of Metabolic Programs, the Fatty Oxidation Disorder (FOD) Support Group, and the Genetics and Metabolism Psychology Network (a national network of psychologists interested in developing standard evaluations for patients with genetic and metabolic disorders). In addition, this project will support the goal of the Metabolic Clinic Quality Improvement Learning Collaborative (being sponsored by the NEGC and directed by John Moeschler, MD) to obtain developmental outcome data on newborn screened children. Specific activities for this project include the following:

**Primary Goal 1: Improve Access to Psychological and Developmental Evaluations for Patients with Fatty Acid Oxidation Disorders (FAODs)**
1. Identify all children with fatty acid oxidation disorders detected by Newborn Screening in New England
2. Administer developmental and neuropsychological screening questionnaires to parents or guardians of children with fatty acid oxidation disorders in New England
3. Provide feedback to families and their healthcare providers regarding the results from the questionnaires and, when needed, recommendations for further evaluation via early intervention programs, psychologists or other specialists

**Primary Goal 2: Improve Education about Fatty Acid Oxidation Disorders and Disseminate Information to Families and Healthcare Providers**
1. Summarize the developmental course and neuropsychological functioning in children with specific fatty acid oxidation disorders
2. Design and disseminate educational materials for families about developmental issues in specific fatty acid oxidation disorders
3. Publish in a peer-reviewed professional journal manuscripts on the screening method and developmental issues in fatty acid oxidation disorders

**Dear Parents:**

You are invited to participate in a research study on children under 15 years of age with fatty acid oxidation disorders (FAODs) identified by newborn screening. The purpose of this research study is to gain a better understanding of how children with FAODs are developing and specifically, we would like to know more about the medical and developmental/educational outcomes of children with these disorders. You are being sent this letter by your metabolic clinic or the Fatty Oxidation Disorders Support Group, which are helping me by distributing this letter to families who might be interested.

This study is being conducted at Children’s Hospital Boston, with a small grant from the New England Genetics Collaborative. Susan Waisbren, PhD (Psychologist for the Metabolism Program) is the Principal Investigator for this study. Our goal is to enroll 80 children with FAODs.

If you decide you would like to participate, a member of the research team will call you to administer questionnaires over the phone. The questionnaires are: 1) Adaptive Behavior Assessment System, Second Edition (ABAS-II), which assess your child’s everyday skills and developmental progress, 2) Behavioral Assessment System for Children, Second Edition (BASC-2), which provides information about your child’s emotional well-being and identifies attention deficit disorder, anxiety, depression and atypical behaviors and 3) Behavioral Rating Inventory of Executive Function (BRIEF), which measures skills related to information processing, such as memory, attention, planning, organization and ability to finish tasks. We will also ask a few background questions about your child’s school experiences. These questionnaires take about 45 minutes to complete. We will also ask your permission to obtain medical records on your child, including the newborn screening finding and confirmatory and other follow-up laboratory results.

There are very minimal risks to participating in this study. You will not need to visit Children’s Hospital Boston and all of the information needed will be collected over the phone or through a medical record review. As part of the study, there is the risk of loss of privacy. To protect you from this risk, your child will be assigned a unique study number. Information linking your child to this number will be stored in a separate file in the research office. All information collected for the study will remain in a locked file and the computer database will be accessible only through a special password.

There are no direct benefits to you for participating in this study; however, your participation will help us have a better understanding of how children with FAODs are doing. We will also send you the results from these questionnaires, which may be helpful to you in planning your child’s school program. Finally, results from this study will be used to develop materials for families and teachers about MCADD. We will send you these materials after the study is completed.

Your participation in the study is completely voluntary. There are no costs to you for participating in this study. You will not receive any payments for your participation. You may withdraw from the study at any time. We will inform you by letter of any research findings that may be relevant to your child.

If you would like to participate in this study please return the enclosed form with the best time and phone number for us to reach you. If you do not want to participate in this study please return the enclosed form and mark off that you do not want us to contact you. If we do not hear from you within two weeks of sending out this letter a member of the study team will call you to see if you are interested in participating.

Thanks so much for considering this request.

Sincerely, Susan E. Waisbren, PhD

---

**REPLY FORM**

**Increasing Access to Care for Newborn Screened Children with Fatty Acid Oxidation Disorders**

| NAME OF PARENT/GUARDIAN: | | |
| NAME OF CHILD: | | |
| DISORDER: | | |
| PHONE NUMBER: | | |
| BEST TIMES TO CALL: | | |
| EMAIL ADDRESS: | | |

I am interested in talking to you about participating:
I am not interested. Please do not call me.

---

**Please return to:**

Susan Waisbren, PhD
Susan.waisbren@childrens.harvard.edu or Lydia.Carr@childrens.harvard.edu

Or call: 617-355-7346

Or mail to:

Susan Waisbren, PhD
Children’s Hospital Boston
1 Autumn Street #525
Boston, MA 02115
Last July I had the pleasure of attending the FOD/OAA Conference in Atlanta to represent the National Institute of General Medical Sciences Human Genetic Cell Repository at the Coriell Institute. I met many wonderful families who kindly shared their stories. I listened with great empathy to the parents who bravely shared their personal struggles with guilt over their child’s diagnosis and I listened with anger and frustration to the young parents who have had their emergency protocols blatantly ignored when their critically ill child presented to the local hospital ER. I want to thank all of the parents and families who stopped by for candidly sharing your experiences and for helping me to gain a deeper understanding of the complexities individuals with inherited metabolic diseases are faced with daily.

For those who I did not get a chance to talk with, I am the genetic counselor for the NIGMS repository at Coriell. The NIGMS Human Genetic Cell Repository is a biobank that collects blood or tissue samples and clinical information from individuals with inherited genetic diseases and makes cell lines and DNA for scientists to use in their research. Samples and corresponding clinical information that are donated to the repository are anonymized and made available to qualified researchers all around the world through an online catalog. Having a centralized source of well-characterized cell lines and DNA allows scientists to spend more of their time and funding on studying how cells function, identifying new mutations, and developing ways to diagnose, treat, and possibly prevent metabolic diseases.

Our goal is to continue to build our collection of fatty acid oxidation disorders and organic acidemias to create a larger, more diverse and more valuable resource for scientists studying the causes of and potential treatments for inherited metabolic diseases. More details about the diseases currently represented in the repository are in the table below. There are many diagnoses for which we do not have any samples. If you are interested in donating a sample to help us build this valuable research resource for fatty acid oxidation disorders and organic acidemias, please contact me either via email tschmidl@coriell.org or by phone at 856-757-4822 for more information.

Thank you again for sharing your stories and thank you to those who have already donated samples!

Sincerely,
Tara Schmidlen, MS CGC
Certified Genetic Counselor
NIGMS Human Genetic Cell Repository
Coriell Institute for Medical Research
403 Haddon Avenue
Camden, NJ 08103

FAQs about the NIGMS HGCR

* Deb also has the stats for the OAAs for the table below — please email her if you’d like them as well

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Number of Samples</th>
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<tbody>
<tr>
<td><strong>FATTY ACID OXIDATION DISORDERS</strong></td>
<td></td>
</tr>
<tr>
<td>2,4-Dienoyl-CoA reductase deficiency</td>
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</tr>
<tr>
<td>Carnitine palmityltransferase I deficiency (CPT I)</td>
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</tr>
<tr>
<td>Carnitine Palmitoyltransferase II deficiency (CPT II)</td>
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<td>Carnitine/acylcarnitine translocase deficiency</td>
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<td>Carnitine uptake defect</td>
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<tr>
<td>Glutaric acidemia type II (GA II)</td>
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</tr>
<tr>
<td>Long chain 3-Hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)</td>
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</tr>
<tr>
<td>Long chain acyl-CoA dehydrogenase deficiency (LCAD)</td>
<td>4</td>
</tr>
<tr>
<td>Medium/Short chain 3-Hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)</td>
<td>0</td>
</tr>
<tr>
<td>Medium chain ketoacyl-CoA thiolase deficiency (MCKAT )</td>
<td>0</td>
</tr>
<tr>
<td>Medium chain acyl-CoA dehydrogenase deficiency (MCAD)</td>
<td>14</td>
</tr>
<tr>
<td>Short chain acyl-CoA dehydrogenase deficiency (SCAD)</td>
<td>4</td>
</tr>
<tr>
<td>Trifunctional protein deficiency (TFP)</td>
<td>2</td>
</tr>
<tr>
<td>Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)</td>
<td>1</td>
</tr>
</tbody>
</table>
I found this on the University of Louisville web site taken from https://louisville.edu/medschool/pediatrics/wcec/preparation-and-storage.html. Also a good general research starting point is https://louisville.edu/medschool/pediatrics/wcec

Storage Concerns

* MCT oil should be stored in tinted glass containers. Refrigerate after opening.
* If possible, foods prepared with MCT should be stored in glass or metal, instead of plastic. Especially true of foods that are expected to separate, such as some salad dressings. Oils such as MCT will, over time, soften plastic surfaces. Glass or metal are preferred for storage.

Storage Times for Prepared MCT Foods

<table>
<thead>
<tr>
<th>Store in the refrigerator for 2 weeks:</th>
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<tbody>
<tr>
<td>MCT Margarine</td>
</tr>
<tr>
<td>MCT Mayonnaise</td>
</tr>
<tr>
<td>MCT Gravy</td>
</tr>
<tr>
<td>MCT salad dressings</td>
</tr>
<tr>
<td>MCT garlic bread</td>
</tr>
<tr>
<td>MCT croutons</td>
</tr>
<tr>
<td>MCT snack mix</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Keep in the refrigerator for 3 days</th>
</tr>
</thead>
<tbody>
<tr>
<td>MCT chicken salad</td>
</tr>
<tr>
<td>MCT egg or tuna salad</td>
</tr>
<tr>
<td>MCT beans and franks</td>
</tr>
<tr>
<td>MCT potato or macaroni salad</td>
</tr>
</tbody>
</table>

James Dad to:
- Cameron 6/CPT2-Asthma
- Jacob 4/Unknown (waiting for Cameron's DNA testing results to test Jacob)

Louisville KY Jenkins1974@insightbb.com

My daughter, Lauren is 8 years old and she also has CPT2. She was diagnosed a year ago, just after she turned 7.

Some of the things I pack for her at school are:
- Applesauce cups
- Box of raisins
- Dried cranberries
- Pear cups in lite syrup
- Fat free chocolate pudding cups
- Low fat mozzarella sticks from Trader Joes
- Apple slices
- Strawberries
- Grapes
- Baby carrots-not high carb, but healthy!
- Low fat or fat free dried cereal
- Pretzels
- Low fat crackers- there are quite a few if you read all the labels
- Cereal bars- many varieties are low in fat
- Homemade muffins- zucchini, carrot, apple
- Mini bagels
- Low fat popcorn
- Fat free yogurt tubes- freeze first and they stay cold in a lunch box

She has PE 2x a week, and I send her out with a bottle of Gatorade to drink before and during any activity so she is getting carbs and fluids so she won’t get dehydrated, especially in the warm weather.

For lunch she usually has a turkey sandwich, fat free mayo, on low fat bread or roll, fruit and sometimes a fat free brownie or cookie. I usually pack a couple of choices off the above list, so sometimes she will also have one of the above with her lunch. We have lunch program at her school that provides nutritional info, so there are a few things she can eat- mainly pasta or chicken and rice that are very low in fat. I try to let her do that every once and a while so she can eat what some of the other kids are. Her special treat if there is a class party, is a rice crispy treat- the kind that are pre-package from the store. They have 2.5 grams of fat, so they aren’t too bad, but still fun when the others are eating something that they shouldn’t be.

Hope some of these food ideas are helpful!

Jennifer Orange County, CA jenlsmith@earthlink.net

Lauren, 8 yrs. CPT2

Websites I get supplements. I thought everyone could use the info:

DHA plant based chewable (for Kids) supplements BRAND: Spectrum
WEBSITE: HealthSuperstore.com $9.63 for 90 capsules

MCT oil BRAND: NOW Foods WEBSITE: House of Nutrition
$13.00 for 32 oz

Walnut oil BRAND: Hain WEBSITE: Unknown but I bought 12 at one time for a good price

Children's Multivitamen BRAND: Flinstone’s Complete SAM’s (I can’t find a website that can beat their price)

I have also used Vitacost and Amazon, but I always compare prices

Dawn Hudnall (FLW, MO)
Rodney 5 LCHAD
Reagan 3 LCHAD
hotrodswife alexanderdawn@hotmail.com

BLOGS:
- My friend, Alta, blogs ‘Tasty Eats at Home’ (listed on this site near the bottom).

Beth Meyer Richardson, TX bemeyery@sbclocal.net
Ella Grace (5 1/2 - MCAD) Evelyn Arnais (3 1/2 - unaffected)

I have just started B’ serendipitious adventures where I share my fat free/low fat recipies. I hope some of you will find them helpful and inspiring. This is a bilingual blog, all of the recipies will be in Norwegian and English. If you get curious on my other posting please let me know and I will translate them for you.

Please give me feed back both positive and negative so I can make the blog better.

Beatrice 30, TFP
Norway
beatbostad@yahoo.no
Q: We actually were in the hospital before Christmas with the stomach bug but got to come home before Christmas Eve. We are home now and he still really doesn't have an appetite. We can get him to eat enough to keep his sugars up, but at night for the last 3 nights he has thrown up. He has kept everything down during the day— it's just at night. The Dr switched us to soy formula, which he still hasn't had at full strength, it's been half soy, half pedialite with added sugar. So after he throws up we take a bath, and have some jello to get us through the night. He has also been very constipated. I am just wondering if any other MCAD babies who have had the stomach bug, have done this, is his tummy just really weak? The Drs also have him on an acid reflex medicine. Just so worried about him—not only keeping him hydrated but keeping his sugars up! He also will not take straight up pedialite, juice, nothing— I mean nothing in his bottle but milk! Any other suggestions to get him through the night?

A: The issue for MCAD deficiency (or anything else) is the amount of glucose needed/given, not its concentration. D5 is 5%, D10 is 10%, so 100 ml of D5 is the equivalent of 50 ml of D10, as far as glucose goes. The significance is in the water along with the glucose—you will need to make twice as much urine to get rid of the extra water, if you use D5. The opposite problem is the concentration of the glucose if the vein blows and you get an infiltrate--D5 is easily tolerated, D10 usually, and anything higher is likely to cause a burn, sometimes with loss of skin. When we use intravenous nutrition for everything (total parenteral nutrition--TPN), we will use a long catheter so there's no chance of leakage around the place where it enters the vein, and the tip is in a larger vein (with greater flow), closer to the heart. We can then use 20 or 25% glucose, and give a lot of calories without risking a burn.

So what concentration of glucose we use is balanced between need and risk. An infant might require 8 mg glucose/kg body weight/min, which comes out roughly 500 mg/kg/hr, or 12 gm glucose/kg24hr. If you used D10, this would be 120 ml/kg/24hr. This is roughly 2/3 as much fluid as a baby would be taking as milk. You don't absorb all the liquid given by mouth, though. If there is any question of heart problems (not in MCAD, but sometimes in the long-chain disorders) we don't like to overload the heart. For a baby in true heart failure we are often forced to restrict fluids to well under 100 ml/kg/d. So we have to use very concentrated calories, which means a central line. Another favorite source of calories for IV use, lipids, are of course off-limits to the FOD babies.

So back to an 8-month-old with MCAD def and upset tummy: IV glucose is the way to go, as concentrated as the vein will allow, so as to get in enough calories to suppress catabolism (fat breakdown)–not as many as a newborn would need, but still quite a lot compared to an older child. This is about the simplest management issue for a metabolic disorder, so I would hope a good pediatrician, pediatric endocrinologist (or gastro specialist), with telephone guidance as needed from a metabolic specialist, would feel comfortable taking care of the baby.

The need for carnitine in these children is a discussion for another time. I prefer my young patients to be on it all the time.

The significance is in the water along with the glucose

I can't recall using a g-tube of any sort in an MCAD child, either temporary (nasogastric) or permanent (G-button), since the episodes of illness are transient. For some of our other conditions, especially organic acidurias with major appetite problems, a g-button is a huge help. But when the problem is an upset tummy due to a virus, putting the food directly into the stomach isn't going to help. IV glucose for a few days should be sufficient to get through the period of vomiting and poor appetite. MCAD deficiency is quite exceptional for the metabolic disorders, as most days are completely free of any hint of metabolic decompensation or danger, and we mostly have to be sure the baby/child/older person doesn't go too long between meals.

Reach for the Stars!

I wanted to share what happened today. Carter, 6, MCAD, ran in a 1-mile Fun Run that our church sponsors. He ran, did a little walking, and sprinted to the finish! We were so proud of him and pleased for him :) I gave him a little gel pack, just in case he needed it (I knew he wouldn't), and his PE teacher stuck close on her bike, but he did great! He finished in about 11 1/2 minutes.

I hope that's inspiring to those of you who have little ones. I remember how terrified we were of Carter's future. He may not be "normal," but who cares? We like him this way! :)
The Phillips’ family would love to introduce Abigail May Phillips into the world. She was born August 11, 2010 at 4:03 am. Abby weighed 5 pounds 15 ounces and was a bundle of red headed joy.

I was excited when I found out I was pregnant, although a little nervous since my previous pregnancies were full of puking my guts out. My son’s Drs (TFP) wanted to do an early genetic test to determine if my third child was affected with TFP or not. I told them I didn’t want to until later… I didn’t want to lose my baby. I felt so good throughout my pregnancy that I was thinking that this baby couldn’t have been affected. At 33 weeks, the latest we could wait, I had an amnio done at UCSF to determine carrier status. Two weeks later, we found out the positive (TFP) results. We then started to make the plans to deliver at Stanford so the baby could be carefully watched after birth. The induction process started on Monday, August 9 at 39 weeks gestation. She finally decided to grace us with her presence Wednesday, August 11. The OB staff was wonderful at Stanford. Abby was taken to the NICU after her first breastfeeding session. There she and I remained for the next 6 days. Finally, the next Tuesday, we were allowed to return home. What a joyous day. Abby has been such a joy in our lives. We are looking to the road ahead and being as positive as we can.

Angie, mom to Nathan 2 1/2 TFP  Abby 1 week TFP  Ali 4 carrier CA angienphillips@gmail.com

I wanted to share the great news that we welcomed Caitlyn Janette into our family on 8/19. She weighed 7 lbs 7 ounces and is doing great! Our hospital did an amazing job of taking care of her before we received the test results, especially since I developed Eclampsia AFTER I had her (blood pressure was great during the pregnancy, it was very strange) and she had to be kept in the nursery while I was treated with a Magnesium IV to prevent a stroke or seizure. Her big sister, Maren, is wonderful with her and big brother, Jack, is having a little trouble with the adjustment but doing his best. Thanks to everyone for emailing me.

Heather DeBar  Maren 5, MCAD  Jack 3- not affected with MCAD, ketotic hypoglycemia, carnitine deficiency and delays heatherdebar@comcast.net

Hi my name is LeeAnn Gattone and I have a 3.5 yr old daughter, Kristin, and a 17 day old son, Dominic, who was diagnosed with LCHAD at 7 days old. On the second day after giving birth his blood sugar was 20. The hospital had no NICU so we were transferred to R W Johnson Children’s hospital in New Brunswick NJ. While the Drs were doing testing his NBS came back positive and that led to more genetic testing. When he was diagnosed they ordered special formula called enfaport. I also nursed him 2x a day. This was devastating to my husband and I - we had no clue that we were carriers for this disorder and our daughter is completely healthy. I am very thankful for this support group and look forward to talking to other parents and adults with LCHAD.

LeeAnn  Bridgewater NJ lchadmomy831@hotmail.com

Drake was born 15 July 2010, weighing 7 lbs 11 oz, and 19 in long. He's got an older brother, Duncan, and sister, Miranda, who were very excited to welcome him into our family. He's the first of our children affected with MCAD.

Rachel and Seth Johnson
Duncan, 5, carrier
Miranda, 4 unknown if she's a carrier  Drake, 4 mos, MCAD

Carter Aiden Foster (MCAD) was born February 3, 2010 and he weighed 7lbs 14ozand was 19 1/2 inches long!

Proud parents ~ Kristen and Chad Foster

Please note that we also have an FOD KidsKorner/Adults Gallery and other Pictures on our homepage. To submit a pic please email Deb.
Carson was born Feb 20, 2010 and he is doing wonderfully. He hasn’t shown any symptoms and we even made it through a bad cold at 5 months of age. We see Dr Wong at Children’s Hospital of Los Angeles every 3 months and he is fantastic. He’s very knowledgeable and provides us all the time we need during our appointments.

I do want to thank you for your website and support. When we first heard the news, we were devastated. My wife went into “autopilot” on taking care of Carson immediately with two hour feedings, etc. and I went into “how can he have as normal of a life as possible” and started researching as much as I could. I came across your website, signed up, and you called within 24 hours. It felt so good to hear from a parent of a child with MCAD. I then posted a note on your “Google Groups” and received numerous emails of encouragement. It was nice knowing that we were not alone.

After the shock wore off, we soon realized what everyone was telling us, “Carson will live a very normal life with controls and precautions.” He’s been a terrific baby and luckily has been healthy with a big appetite. He goes right back to sleep during the night feedings and always has a smile on his face. February seems like it was so long ago.

Thanks for everything you do. Keep up the good work. Matt Hammond mhammond@coreland.com

Kourtney Rae Luchau was born on November 14th, 2010. She was 8 lbs 5 oz and 20 ½ inches. Kourtney decided to make her entrance 6 days late on her big sister Brooklyn’s 3rd birthday! Her newborn screening results came back NORMAL! We are still deciding if we will find out Kourtney’s carrier status now or wait until she is older. Brooklyn is a great big sister and loves to help with Kourtney’s diaper changes and baths. Her dolls are dressed in newborn diapers and onesies.

Virginia
Mom to Brooklyn (3 – MCAD) and Kourtney (1 month – normal NBS)
Fargo, ND
Hope everyone is well this new year!
vdahlen@microsoft.com

~ NEEDED FOR THE JULY 2011 ISSUE ~

M edical Update ~ Please Submit

PROFESSIONAL ABSTRACTS/ARTICLES OF ALL KINDS (Drs, Nutritionists, Genetic Counselors, Social Workers, etc.)

FAMILY STORIES & Pictures

The ‘Silver Linings’ of FODs ~
All too often we are reminded of the difficulties associated with FODs. Hopefully our Email List support will help us remember the ‘Silver Linings’ to these disorders as well ~

What is your ‘Silver Lining?’
Complied by Kim — please send your ‘Silver Linings’ to Kim at gareths mommy@yahoo.com to be included in our July 2011 issue.
**DONATIONS**

*since our July 2010 Newsletter*


**Tshirts, Bracelets, Ribbons, CafePress, GoodSearch browsing, or iGive shopping:** Michelle Cotton. Deanna Swiech.

Thank you to all that have bought products from companies on the Internet that support the iGive and Cafepress.com program of donating a certain percentage to Groups like ours. All of those links are on our homepage, right sidebar boxes.


We greatly appreciate donations to help with daily costs, website fees, supplies, Conference costs, phone calls around the world, rent for the Grief Consult office, and raising funds for FOD Clinical Training and FOD Research and long-term investments.

**US Checks can be made payable to ‘FOD GROUP’ and mailed to:**

FOD Group PO Box 54 Okemos, MI 48805

We also have a Secure PayPal link on www.fodsupport.org

ALL US donations are tax-deductible.

Our Tax ID # is 83-0471342.

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**2010 Donations/Expenses for the FOD Group**

*Ending Balances thru Dec 31, 2010*

<table>
<thead>
<tr>
<th>Fund</th>
<th>Ending Balance 12.31.10</th>
<th>2010 Expenses 12.31.10</th>
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<tbody>
<tr>
<td>FOD General Fund</td>
<td>$6600</td>
<td>$120,000</td>
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<td>FOD Clinical Trust Fund</td>
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<td>FOD Research Trust Fund</td>
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<tr>
<td>FOD Petty Cash Fund (balance)</td>
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[The bulk of Expenses were for Conference costs, scholarships, and opening a Grief Consultation office (rent and furniture) to offer pro bono grief support to local Bereaved Parents & Families and also anonymously donating funds from an undesignated professional donation to 2 entities.]

**The 2010 return will be on our site after May 2011**

All Grief Consult donations are deposited into the General Fund, as are Bracelet and Ribbon Sales, Cafepress.com, iGive, Goodsearch, and any donation that isn’t designated for the other Funds ($100,000 given by an anonymous professional). The General Trust Fund is to save/earn interest for the 2012 Conference and other annual costs. Once the Research and Clinical Funds reach @$50,000 we will be able to offer grants to clinicians and researchers in the US. We also have a 1 yr certificate and long-term stocks/bonds earning interest and dividends.

---

**Communicate With Us**

Please **ADD** me to your mailing list [Conference years]

<table>
<thead>
<tr>
<th>Family</th>
<th>Professional (please circle one)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Name/Address or Address Correction (circle one)</td>
<td></td>
</tr>
</tbody>
</table>

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

Name/Address:

Please include ideas for future issues or your questions

---

**‘Do all things with love’**

~ Og Mandino