

FOD Support

fatty oxidation disorder communication network



In this Issue

- From the Editor...1
- Family Stories...2
- Medical Update...10
- Nutrition Update...12
- Q & A...14
- NBS Update...15
- Love Messages...16
- Resources...18
- Kids Corner...19
- Donations...20

From the Editor



Dan, the boys (Kevin, 18, MCAD and Brian, 17) and I hope ALL of you had a HEALTHY and safe holiday season! Now that the winter holidays are over, we are in the process of planning for the coming year and beyond ~ I know you are expecting to hear about our **next National Metabolic Conference**, but because of time constraints (for the sponsoring Group) and other reasons, it will be postponed for this year. However, **the National Coalition for PKU & Allied Disorders is planning on sponsoring a Conference for the Summer of 2006**. Trish Mulkaley, their President, generously allows our Group, as well as 3-4 others, to participate. Past conferences have been held every 18 months, but because this is such a HUGE undertaking, it will require more time between conferences. We have not formally begun to plan, but I am collecting ideas, as far as LOCATION, SPEAKERS, TOPICS, and VOLUNTEERS. We usually ask 4-5 professionals to **share their expertise with our Families in regard to ANY issue related to children and adults with an FOD**. Our sessions will be open to young adults and adults ~ unfortunately we will NOT have daycare for children due to the high cost. So start brainstorming and email me (deb@fodsupport.org) with your ideas.

Please note that we have a **NEW EMAIL LIST** ~ if you were on the old topic list you'll have to sign up for our new List by going to our website's 'Join Our Email List' page. Once you are signed up, be sure to save the email that explains how to send messages, read the archives, how to change your password and set your options. It is important for you to check your spam filters because if they are set too high you will not receive list type messages or they will immediately go to your junk mail/spam folder and you may get "bounced" from the List. We also have access to all of our old topic list messages ~ so if new members would like to be able to read them you'll have to sign up on the website as well. I will approve your email address, but there is a delay from when you sign up and when you can get into the old archives. Although messages are no longer being sent over the topic list, there are hundreds of archived messages on various topics that may be very helpful for Families. While you're on the www.fodsupport.org site signing up for the List, be sure to check out all the other pages ~ we update with new information as often as we can (ie. Pharmaceutical update on prescription generic l-carnitine).

Thank you to our Families that shared their struggles and challenges with us in this issue by way of their stories. We welcome ALL of your stories and pictures and we will try to either print them in the newsletter or place them on the *Family Stories*, *Newborn Screening*, or *Love Messages* page on our site. We would especially like to encourage families dealing with some of the less common FODs (i.e. HMG, SCHAD, Carnitine Acylcarnitine Translocase, TFP, CPT 1&2 etc.) to share their experiences. We're also always looking for more low fat recipes, poems, and pictures.

Whether you're a Family or a Professional, we are **all striving to create awareness, education, screening and diagnosis, clinical treatment and research** ~ by sharing your story or your expertise...

'We Are All in This Together!'

Take care... 

Letters to the Editor

Dear Deb: I came across your website when searching for some information and wondered if you could help. As shown in the attached story (reprinted in our Family Story section), I have been diagnosed with CPT 2 and have learned to live with it for 5 years now. It has had a major impact on my life changing it in so many ways and up until recently I have coped, but now I am having great difficulty with my working life. As a result of losing many jobs through a lot of time off and people not understanding my problems because they haven't heard of it before, I am now trying to raise awareness of this disorder in England. However, I am not sure where to start and how to go about it, but I am willing to take any suggestions and would be more than willing to give myself for any tests. I'm very grateful for you taking the time to read this and I really hope you can help. Please be in touch. Many thanks ~ Brendan Embra,

Brendan_embra@hotmail.com

Dear Brendan: Welcome to our Group! You have found the right place for not only information but for great support. As I mentioned in my email to you, we have several adult onset CPT 2ers in our Group and the best way to network with them is through our Email List, which you can sign up for on our website. Additionally, there is another site (www.spiralnotebook.org) that has great info on CPT 2. Some of those that helped put that site together are also members of our Group. As for research opportunities, you may want to contact Dr Roe at Baylor (refer to http://www.fodsupport.org/fod_study.htm for more info). Some of our members are participating in the C7 oil research in Dallas, Texas. Dr Georgirene Vladutiu in Buffalo, NY also does research on CPT 2 but you would have to contact her at mitomaven@aol.com to see what she is pursuing. With your article below, you are definitely creating awareness of these disorders so you're off to a great start! I hope some of this info is helpful. Be sure to introduce yourself on our Email List and you will hear from others experiencing similar issues in their personal and professional lives. DLG



Family Stories -

Brendan's Story, adult onset CPT2 England

Printed in the Banbury Guardian, Oct 14, 2004 by Matthew Smith. Brendan comes back fighting: Brave young man's struggle to come to terms with rare genetic disorder

A young man suddenly struck down by a rare illness is fighting to raise awareness of the condition. Eighteen-year-old Brendan Embra, of Badger Way, Banbury, has even offered to be a guinea pig for medical research into the genetic disorder.

The complicated condition ended Brendan's promising football career with Oxford United and he had to miss a year of schooling. He was diagnosed 4 years ago with carnitine palmitoyl transferase 2 deficiency, a genetic disorder which makes his muscle cells disintegrate by a defect in the way his body metabolizes fatty acids. An attack can be triggered by illness, physical exertion, common painkillers such as paracetamol, alcohol, stress, trauma or a high fat diet. It could result in kidney failure and breathing problems.

Brendan has found no easy answers to his condition and hopes if more cases are discovered it will spark greater interest in research. The disorder has been reported in fewer than 200 people throughout the world. Determined to battle on, he started a part-time job last week as a learning support assistant at Blessed George Napier School where he is helping to teach children with learning difficulties.

Brendan said, "I'm really enjoying it and feel like I'm doing something worthwhile again."

Although Brendan had tried a series of part-time jobs, including telesales, barman, and football coach, he had not been able to cope. He was upset when an application for benefits was turned down by the Department for Work and Pensions because it did not recognize his rare disorder.

Brendans's Story... cont'd

He first fell ill in June 2000 during a football match. "I was getting aches and pains like cramps," he said. "I sat down and tried to rehydrate myself. But when I tried to get up again, I couldn't move."

Unable to fathom out what was happening, Brendan was taken to Stoke Mandeville Hospital in Buckinghamshire for examination where he stayed for nine days on a drip. Five months later he developed similar symptoms and was admitted for 3 weeks. Ever since, there has rarely been a month where he has not been in hospital. Sometimes it is for a day, others for weeks on end.

"Before I used to get cramps from head to toe lasting days on end," Brendan said. "Now it has started to show more in the way of muscle spasms and fits."

Brendan previously kept his condition private from all but close friends and family. But now he wants to highlight the illness, as he explained: "I'm sure if more was known about it there would be far more cases reported and research done."

Frequent hospital visits and the prospect of another attack meant Brendan failed to complete a diploma in sports science at Oxford College. "I tried to leave home to see if I could cope. But because I wasn't working enough I got into debt and it cost me my relationship with my fiancée," he said. Doug Turnbull, professor of neurology at the University of Newcastle upon Tyne, is an expert on the condition. He explained CPT 2 was an enzyme involved in transporting long chain fatty acids (a very important source of energy) into the powerhouses of the cell.

"Long chain fatty acids are very important for the production of energy in muscle during prolonged exercise. In patients with CPT 2 deficiency they can often perform short bursts of exercise, but struggle with more prolonged exercise. Treatment is quite difficult but usually consists of altering the diet with increased carbohydrates," said Prof Turnbull.

[If you would like to network with Brendan please email him at Brendan_embra@hotmail.com]



Family Stories - *Morgan's Story, LCHAD*

[As written for the 'March of Dimes Ride']

Morgan suffers from LCHAD, a condition that prevents her from efficiently oxidizing long-chain fatty acids. Although there are numerous health risks caused by this condition, this disorder is particularly dangerous during infancy.

During their early years, all children have limited glucose in their systems. Due to the small size of their livers, they also have very small "storage tanks" for glucose reserves. Given this limited short-term glucose supply, infants may frequently consume their entire reserve. This consumption may occur for many reasons, including battling colds and viruses. Some viruses, such as rotavirus, can quickly reduce a child's glucose levels because the child not only expends considerable energy fighting the virus, but also has trouble replenishing his or her glucose reserves due to lack of appetite and dehydration.

For most people (including infants), a reduced short-term glucose level is not life threatening. When glucose is low, our bodies look to fatty acids for additional energy. Our "oxidation" of these fatty acids, however, does not occur in one step. Instead, we complete this process in stages. First, we break down "long-chain fat" into "medium-chain fat." Next, we break down "medium-chain fat" into "small-chain fat." We then break down the "small-chain fat." Ultimately, this oxidation process leads to the creation of energy that allows our bodies to function, notwithstanding the temporary loss of our short-term glucose reserves. In short, the fatty acids in our bodies serve as a safety net when we use up our glucose storage tanks.

For most individuals, this process is taken for granted. Nearly everyone, including carriers of fatty-acid oxidation disorders ("FOD"), possess sufficient metabolic resources to break down fats during each critical sequence. Children who suffer some form of FOD, however, do not. Children with LCHAD cannot efficiently break down long-chain fat. Children with MCAD cannot efficiently break down medium-chain fat. Children with SCHAD cannot efficiently break down small-chain fat. Simply stated, children with fatty-acid oxidation disorders have considerable "holes" in their metabolic safety nets.

These holes raise numerous short-term and long-term problems. In the short term, children with these disorders are at risk any time they fast for a long period of time. Unfortunately, the length of a normal sleep cycle, under certain conditions, may be too long for a child suffering from a FOD. For example, if the child's body is fighting a virus, he or she may quickly consume their entire short-term glucose stockpile. If this stockpile disappears during the sleep cycle, a child with a FOD may never awake. Their glucose levels may simply spiral downward which, in turn, may cause hypoketotic hypoglycemia. That condition may lead to coma or death. For LCHAD children, such as Morgan, one of three die during an initial hypoglycemic episode. Regrettably, those deaths could easily be avoided if the child simply received another feeding during the night. A few ounces of a soft drink or juice could be the difference between life and death.

Morgan's Story... cont'd

Even where a child with LCHAD survives this initial episode, he or she still faces many long-term problems. The longer the child goes undiagnosed, the more severe these problems may become. Left undiagnosed, LCHAD children usually continue to eat as other children eat, which means a fair percentage of their food intake is fat. That fat consumption by an LCHAD child is very dangerous. If these children eat the same percentage of fat as most children do, they may quickly develop liver problems, heart problems (cardiomyopathy), muscle weakness and damage (hypotonia and rhabdomyolysis), kidney failure (as a result of rhabdomyolysis), and blindness (retinitis pigmentosa). With proper diet adjustments and supplements, these long-term risk factors can be appreciably reduced and, in some instances, eliminated.

Parents who know their child has LCHAD can take steps to avoid these short-term and long-term problems. Parents who do not know, on the other hand, are unwittingly walking through a minefield. Stepping on any one of those mines (e.g., hypoglycemia, cardiomyopathy, etc.) may result in death. Even after death, many parents do not know their child died from a FOD. It is now believed that a fair percentage of deaths attributed to SIDs should actually have been attributed to LCHAD or other fatty-acid oxidation disorders.

Even though an LCHAD child's life hinges on early detection, parents of these children have little, if any, reason to suspect they are navigating through troubled waters. The parents usually have no reason to know they are LCHAD carriers since they do not have any problem processing long-chain fats. LCHAD children also appear physically normal. Although they may some times have a doughy appearance and act in a lethargic manner, parents typically credit these characteristics to baby fat and a calm demeanor. It is only with the benefit of a positive diagnosis and hindsight that parents realize that these seemingly benign features are consistent with LCHAD.

But how can parents obtain an early diagnosis? Fortunately, with recent technological advances, it is now possible for children to be screened for LCHAD immediately after birth. Some states are already screening for LCHAD. Others have it under consideration. Also, the private sector offers parents the ability to screen for many disorders that may not be covered under state law, including LCHAD. Many times, however, parents are unaware of the need to screen for additional disorders. They may also not be in a position to afford the cost of private screening.

In Morgan's case, we were lucky. We almost lost her at nine months of age. When Nancy tried to wake Morgan one morning, she would not respond. She did not appear to be breathing. Her eyes appeared to roll back in her head. Nancy did not know if Morgan was dead or alive. We were blindsided. In an instant we went from believing we had a completely normal nine-month old girl to realizing she may be gone forever. We will never forget the anxiety of not knowing whether she could be revived. Even after her revival, we quickly realized we were far from being out of the woods. We will never forget our anxiousness as we waited for the results of her CT scan to determine if she had suffered any permanent brain damage. We will never forget our feelings of hopelessness as intensive care doctors were at a loss to tell us why our daughter came so close to death.

Despite these feelings, we also realized how lucky we were. We were lucky to be living in a large metropolitan area and only a few miles from Scottish Rite, an outstanding children's hospital. We were lucky that the emergency room doctors immediately knew to check Morgan's glucose levels, which were exceedingly low. We were lucky that the doctors almost instantly started an IV. We were also lucky to live near Emory University, whose pediatric genetics department was able to ultimately diagnose Morgan's condition. Yes, we were lucky in many respects.

But many parents do not have this luck. They may not live in a major metropolitan area. They may not have quick access to a children's hospital. They may not live near specialists who are aware of LCHAD or how to diagnose it.

With expanded newborn screening, we can eliminate any reliance upon luck. Instead, we can substitute existing, available technology for chance. We can give parents the comfort of knowing whether their child has any of thirty inherited disorders, including LCHAD. With early detection and treatment, many of these children can enjoy fruitful, productive and fulfilling lives. Without early detection, these lives may be destroyed or significantly impaired. **These human costs are unnecessary and can be avoided through expanded screening.**

Mike and Nancy McConnell
Dunwoody, Georgia

FOD Family Questionnaire

If you do NOT see your name on the **Family List** or on **this issue's Update**, it is because I (Deb) never received the FOD Family Questionnaire that I sent you in the Family Packet when you first registered with us. **If you would like to be listed** for networking purposes, please go to '**Online Forms**' on our website (www.fodsupport.org) and **print out the Questionnaire**. Then **SIGN it and DATE it** so I have your permission to list you. Please **mail it to me via the regular mail**

(see page 1 of this issue for address) so we can list you in the next List Update.



Family Stories - *David's Story - MCAD*

My second child, David, was born on June 14, 2004. During my delivery, I had a temperature that worried the doctors. After breaking my water and seeing it was "mucky" they were even more concerned. I on the other hand was fine. After all, I had a very healthy little girl at home waiting for her new baby brother. What could go wrong? **David didn't cry when he was born. That worried me but they assured me he was fine** and that they just wanted to watch him because his temp was a little low and I had a high temp during delivery. He was taken from me after only holding him for a few minutes. Still, they said he was fine and I knew he would be back soon to start feeding.

Over the next 2 days my son was with me a total of 2 hours and only ate for a total of 30 minutes. **He was lethargic, ice cold, had low sugar levels and breathing problems.** He was placed under a warmer, hooked up to a heart monitor and a breathing monitor to keep track of his oxygen level. He was also put on an IV and had 2 spinal taps. During the first 2 days he was given 2 bottles of formula to get his sugar level up. I was not happy. I wanted to breast feed him but the doctor said it was more beneficial to have the bottles of formula.



The evening of the second day he was given to me and stayed with me for a good while and ate some. **The next morning he again ate some and was tested for sugars and was fine. He was to be released and could go home.** But I couldn't leave. They said I needed to be watched because of the small temp I had during delivery. This made my husband and I very mad. The past 2 1/2 days were all about my son and how he was not doing well now all of a sudden he's fine but I'm not. We asked to speak to the doctor because we were going home! **While waiting for the doctor, they took my son for the last routine tests and returned to tell me that he was lethargic, ice cold, had low sugar, and breathing problems. He was put back under the warmer, hooked back up to the heart and breathing monitors and put back on the IV.** They said a doctor would be in shortly to talk with us. When the doctor came in, we were briefly told that while waiting to get the routine test done David's sugar level dropped, his breathing became irregular and again he got cold. He had been away from me for 4 hours with no food and had only eaten a very small amount that morning. The doctor said **David's newborn screening test came back showing he had MCADD** and that they would like to retest him. Of course we said yes. We had no clue what MCADD was and neither did the doctor.

That night I was allowed to sit with my son again and hold his hand but I could not feed him or pick him up. In the morning a specialist came in to briefly explain to us about MCADD. She assured us she would come back the next day to tell us more. One of the student nurses over heard this and, on her own, printed out what she could find on the Internet about MCADD. The next day we got the same information from the specialist and not much else but by having the information from the Internet, we were able to ask questions and understand more of what was happening to our son. David was put on Camitorâ. When the doctor asked if I would allow another bottle, I said NO. If my son has this thing called MCADD and part of the "treatment" is frequent feeding, then stop taking him from me and let me feed him. I asked him, "Doesn't it make sense that if he needs to eat, I be allowed to feed him every 2 hours rather than them keeping him in the nursery away from me with no food?" That's when things began to improve. I started feeding him every 2 hours and slowly he was taken off the IV.

One week after my son was born he was well enough to go home. After being home only a couple of hours with our new baby, our little girl Kloey, whom we had not seen much in the past week, was taken to the emergency room. We were told she had pneumonia. Fearing that the baby might catch it, Kloey went back to her nanny's for the next 4 days while we went to all of the doctor appointments that were set up for the baby. Our son is now 6 months old. He has his sugar level tested 2 times daily. He eats whenever he wants but can't go longer than 4 hours without eating during the day and he sleeps 7 hours at night (with comstarch in his last bottle). His sugar levels are doing wonderfully and he is getting so big. Currently he is waiting to be evaluated by Newborn Intervention. He is unable to put any weight on his legs, but over all he is fine. We have been lucky and had no hospital stays and no ER visit (although I have been a few times out of my own fear). Our little girl tested negative for MCAD but carrier status is still unknown. Scary as it has been, **I'm so thankful our son was diagnosed at birth.** Although, I feel the problems that he had at birth were triggered by them not allowing me to feed him. Things have worked out for the best, however. My husband's grandmother lost 2 sons to what the doctors thought was crib death (SIDS). I'm thinking there may be a connection. There are still days when I fear the worst. I'm not sure that I will ever stop fearing. I do know now that **I need to use my fear to push myself to learning more about MCADD, teaching others, and enjoying every moment with my kids.**

Brandie Cole
Bree3382@aol.com



Family Stories - *Update on Jake Bray - LCHAD*

[To read about Jake's birth/newborn screening diagnosis story visit <http://www.fodsupportors/jake.htm>]

We finally got Jake home from the NICU and had our diagnosis, but we were very overwhelmed. The doctors and nurses at the hospital (Winchester Hospital in MA) had NEVER had a baby diagnosed with LCHAD via NBS and none of them even knew what it was! We had started doing some research online and at the time there wasn't much out there on LCHAD so we had primarily been reading all kinds of out of date studies which totally freaked us out mostly stating that the mortality rate was extremely high and all kinds of other alarming information. By the way Jake's mutation number is G1528C (both copies).

A few days after we brought Jake home from the NICU we saw a Metabolic doctor at Children's Hospital in Boston for the first time, Dr. Deborah Marsden. Finally, someone who knew about LCHAD! She gave us a great explanation of the disease and its many complications to the metabolism and biochemistry of the body during acute illness. It was definitely information overload but at least it was factual current information and there was a treatment - basically a low-fat diet, frequent feeds and medicine... and there was hope for a happy, healthy life!

Jake's story may be a bit more complicated than most LCHAD cases. He started off being healthy and not catching any viruses for almost a year, although he was a very fussy, whiney and difficult baby. He screamed so hard at times that we thought he was going to go into cardiac arrest or something (if that's even possible). He would arch his back, become rigid and would shake and scream as if he were being tortured and wanted to jump out of his skin! We later figured out that he had pretty severe acid reflux and constipation (which many Chiders have due to low muscle tone) and it took a few months and an upper GI test to figure out those problems and get him on the right medicines. Those were a few LONG months of constant crying, vomiting up every bottle, pooping through every diaper because of the laxatives he needed in order to move his bowels and sleepless nights to realize that our bundle of joy was going to be a handful. Our patience was tested to the limit having such an unhappy baby! For a couple who waited 5 years after marriage to get pregnant because they weren't even sure if they wanted to have a child dealing with all of this put an incredible strain on our marriage. Not to mention the stress of dealing with several different doctors every time we didn't know what was wrong (Metabolic doc, Pediatrician, GI Doc), not being able to leave him with a babysitter and eventually not being able to work. It was also very frustrating that the pediatrician didn't know about LCHAD nor did most other doctors because it is so rare, so the pediatrician and other specialists would often point us back to the metabolic doctor and vice-a-versa! Once we got Jake on some meds for the reflux and constipation he was a bit less fussy but he was still very difficult and much more whiney than most babies and still had spells of extreme screaming if he went a day or so without a bowel movement. At one point we even had a massage therapist come to the house which really seemed to work well and I learned the technique which I still sometimes use on him today if necessary. Of course we were always worried if something was going wrong metabolically that we couldn't see because he was just so unbelievably fussy, or if there was anything else wrong with him that we hadn't looked into or if he was just colicky.

Fortunately we would see the Metabolic doctor every month at first and eventually every 3 months to make sure everything was okay metabolically. We would discuss his nutrition (he was on a special low-fat formula called Portagen since he couldn't metabolize the fat in my breast milk) with the nutritionist and we would get some blood labs drawn (liver function tests, CK, carnitine levels, and essential fatty acid levels). His labs always looked good so at least we knew as far as the LCHAD was concerned he was doing well and we just kept telling ourselves that some day he wouldn't be an unhappy baby anymore!

We were always very careful not to visit anyone who was sick with anything worse than a clear runny nose so we kept him healthy for a while. Then he had his first of many acute illnesses that caused him some problems. We found out he was allergic to our cat and he got a few really bad sinus infections (which are very hard to diagnose in a young baby). Those put him in the hospital a few times before we figured out what was causing the coughing and vomiting. During these acute illnesses his liver function and CK levels would be pretty elevated (usually between 1000-3000). Once we got past that we had a few months hospital free and then... **he started vomiting severely every month for seven months in a row** (yes Jake vomited more than any baby I've ever known)! In comparing notes with other FOD parents vomiting is common but not quite like this. Jake was apparently unlucky enough to also have something called **Cyclical Vomiting Syndrome**... what are the chances! I have actually met one or two other parents of FOD kids who also have this so perhaps there's a link but it has not been proven. This may sound crude but I actually figured out it was on a cycle because it seemed we were in the hospital every time my monthly cycle came around! The doctors kept writing it off as viral and I wasn't buying it after several months and actually had to page Jake's GI doctor while we were admitted at Children's in Boston and asked them about CVS which I had heard of in some of my research on vomiting and another FOD parent had told me about it at that time as well. I had demanded someone come see Jake immediately and sure enough he was diagnosed with CVS... a very bad combination with LCHAD!

Jake's last vomiting episode from CVS was in May 2004. Unfortunately **it led to his worst metabolic crisis since birth!** He had been vomiting for about 24 hours, at first not too badly and we were getting some fluids into him which seemed to be enough to maintain his blood sugar. It was over night when he took a turn for the worse and it was hard to tell if he was sleepy or lethargic.

Update on Jake Bray... cont'd

When we checked his blood sugar again it was 11! He looked bad but not horrible so we thought the glucometer was wrong and made a HUGE mistake of driving him to the ER (at Children's Hosp in Boston) instead of calling 911! It ended up being rush hour morning traffic as well as a BIG MISTAKE! I gave him sips of milk with cornstarch in it the whole ride (of course the one time I didn't have the glucose paste in his diaper bag this happens) that was probably the only thing that kept him out of a coma!

When we got to the ER he was in and out of sleep and became unresponsive. They had trouble getting an IV into him (as they always do, he has really tough veins and had had so many IVs that his veins have a lot of scar tissue in them and don't work very well for IVs anymore. They had to give him a shot of Glucagon to keep his sugars up and it took about an hour of 4 people (doctors, a nurse and an IV nurse) to get an IV into him anywhere they could stick it. Meantime his blood sugar was 11 per their glucometer! It was so horrifying to watch, that image will be in my head as long as I live along with the worry of whether he was going to make it through this and if he did, was he going to have brain damage from having blood sugar that low! They finally got an IV into his lower leg by squeezing and pumping his foot and leg repeatedly to get blood flowing to the vein enough to get the IV in! It was a metabolic Fellow who got it in ~ she will forever be my hero! Jake remained pretty unresponsive and lethargic for probably about 15 hours or so and was put into the ICU. It was so scary to see him lying there barely conscious. I was afraid to leave his side. His IVs wouldn't stay in. They kept infiltrating after about 5 hours and we had to go through the same torture to get another one in. One of the doctors suggested that we put Jake in for surgery and have a port-a-cath implanted into his chest with a line going into his heart so that he would no longer need IVs to be put into his veins, instead they would be easily inserted into this device by poking through his chest. Of course after just going through such trauma and still not being out of the woods the last thing we wanted to hear was the word "surgery" for anything! But we knew it was this or several times a day of pinning Jake down while he's screaming and getting poked over and over again with failed IV attempts. So although we were very nervous we went through with it and we are so glad that we did. It has saved Jake so much trauma with IVs and it's great for blood draws too. The only down side is it needs to be flushed by a visiting nurse once a month with saline and heparin but it's very quick and easy.

Anyway, getting back to that hospital stay he ended up recovering pretty well. It took him several days to get his strength back and the doctors warned that this may be a big set back for Jake developmentally if he had sustained any brain damage from the low blood sugar. The only odd thing that we noticed right away was he was eating really funny. He was kind of swishing food around in his mouth and not biting down fully as if it were the first time he'd ever eaten food before. But he did get over that within a week or so. It was hard to tell otherwise because he wasn't really talking at that point and was already behind by about a year with his receptive and verbal language. Actually, surprisingly, shortly after that hospitalization he started talking and making tons of progress. It was almost as if he had gotten a kick-start or something. We still haven't done an MRI to see if he sustained any brain damage from this episode (or at birth for that matter) simply because we don't want to put him under general anesthesia just for a test right now. He's been through way too much for being 2 1/2 years old and the risk of general anesthesia for an LCHAD kid even with a glucose IV in is too risky for us right now. I do want to do this at some point but we may wait until he's old enough to just cooperate for the test won't need to be put under general anesthesia.

Since this last crisis he's been doing very good except now that he has been healthy for several months we are focusing heavily on his speech and occupational therapy. He is FINALLY starting to talk and is putting a few words together so we are very excited about that. However he has been doing some odd behavioral things for a while now. **We are trying to figure out if he was doing all of these things before his bad crisis in May 2004 or after it wondering if some of this may be due to potential slight brain damage that may have occurred during his major crisis at birth or in May '04 or who knows could his developmental issues be because of LCHAD or some other reason?**

He was always a very fussy and very difficult baby and just never seemed right. I can't even explain it but he just didn't do much smiling and cooing and typical things that babies do. Now as a toddler through early intervention therapy they have helped me figure out what it is that seems so different about Jake to me. **He is very rigid, has trouble transitioning from place to place / activity to activity, even changing his clothes. He won't let me cut his hair, nails (I have to do it in his sleep), changing his diaper or shirts is a nightmare. He also shakes his head from side to side really fast a few times a day, spins in circles and sometimes stares at objects that he is holding a little too close to his face and stares at them oddly. So recently figured out that he has some tactile and sensory issues.** Given this combination his pediatrician has diagnosed him with PDD (Pervasive Developmental Disorder) which falls under the autism spectrum. So yet another very emotional and stressful time for us in the roller coaster ride of life with Jake. We are waiting to see a developmental pediatrician to confirm this but his speech and occupational therapists agree that this diagnosis makes sense. We really haven't had much of a break; it's just been one thing after another with him, from LCHAD to CVS, to developmental issues. Not to mention the strain on our marriage, it's definitely tough making time for each other and letting all of this stress go. Prior to Jake we were boating and offroading every weekend, now we hardly ever get out for dinner!

So there you have it! Jake is definitely one of the more complicated LCHADers having the cyclical vomiting syndrome and PDD. We are trying to keep positive, it surely is a daily struggle. But he is the most lovable and sweetest little boy I have ever seen. I think somehow he knows that he is special and really appreciates the little things we do for him. **He is making progress every day and knock on wood this is the longest stretch in a while that we've kept him out of the hospital for.** I've probably made him sound like a complete invalid, you would actually never know by looking at him that he has any medical or developmental issues, you'd have to spend time with him to notice that he's "different."

My two cents on FOD kids is

These FOD kids are all very alike but also very different for various reasons. Some were diagnosed earlier and had a better chance for a pretty normal and healthy life, some were diagnosed later after several complications, some have other medical or developmental issues (which haven't been proven to be linked to FODs but lots of parents seem to have similar stories, especially with developmental delays). If you are reading this and just received a diagnosis my best advice is to ask lots of questions and talk to as many FODers or FOD parents that you can. You will find that unfortunately most Pediatricians and other doctors don't know anything about these disorders and the Metabolic doctors are great but since Metabolism apparently isn't a very appealing medical specialty these days these doctors are stretched pretty thin (apparently they need further schooling than most specialists and don't get paid well at all!). **I would highly advise joining the www.fodsupport.org support group** and share your story, make some contacts and read the email posts since many contain excellent information, especially the ones that come from some medical professionals on the List.

Michelle Bray
mnbray@comcast.net



Family Stories - Zack's Story - LCHAD

Zack was born early at 36 weeks because my wife Amber suffered from severe preeclampsia. Zack weighed a normal (for his gestational age) 5lbs 5oz at the time of his delivery. There were no complications with the delivery. However, they kept him in the NICU for a week because he was not able to maintain his body temperature. Zack also had pneumothorax that went away after a few hours on 100% oxygen.

Zack was released from the hospital a week after he was born and came home with us. The next 3 weeks were full of happiness as I was lucky enough to have a month paternity leave. **When Zack was 3 weeks old he woke up around 5am and had horrible projectile vomiting.** This continued a few more times over the next hour while I was trying to get in touch with his pediatrician who told us to **go to the emergency room.** Several tests and 24 hours later we were discharged from the hospital after the doctors told us he was just dehydrated.

Over the next month Zack's health slowly degraded as he started to throw up with almost every feeding. Because of the persistent vomiting Zack had only gained 1 pound since birth at 2 months of age. Also, around this time Zack became completely lethargic and didn't hold anything down for almost a day. **We rushed him to the emergency room again.**

Doctors weren't exactly quick with answers during this hospitalization that lasted 5 days. They ran countless tests ranging from a CT Scan to a Renal Ultrasound. We did find out that **his ammonia and liver enzymes were elevated.** Because of this they **started running tests to determine if he had a metabolic disorder but stopped after the first one,** an organic acid profile which came back normal, when a urine culture came back positive for a urinary tract infection. **Although our gut instinct told us there was something else going on** I was relieved with this diagnosis since it wasn't serious. At this point I was naïve and didn't question the doctors because I thought they knew everything. However, Amber wasn't relieved by this diagnosis but we all brushed her concerns aside. I wish I would have listened to her concerns and demanded then that the doctors continue the tests to determine if he had a metabolic disorder. Looking back at pictures of Zack around this time it is easy to see how sick he looked, **definitely not just a urinary tract infection.**

Zack was released from the hospital and was prescribed Zantac for reflux and an antibiotic to treat his urinary tract infection. Over the next 1 1/2 months Zack actually showed dramatic improvement and gained a lot of weight. However, we were still concerned that there might be something else going on. I don't know why we thought this because he was acting fine but something just didn't seem right.

Amber was constantly taking him to the pediatrician because she thought something was going on. **Even though he looked fine he was acting different than his normal. This was very hard to explain to doctors because he didn't appear to be sick.** Zack's pediatrician ran a blood test and it showed that he was severely dehydrated which prompted him to refer us yet again to the local children's hospital.

By now I'd had enough with the first question that came out of a doctor's mouth at the local hospital being "Are you first time parent?" I realized that they were asking this to determine how credible our statements should be taken. It was during this trip to the ER that **I demanded a complete metabolic workup** be ordered because 3 trips to a hospital in 4 months wasn't right. It also wasn't right that Zack was almost 5 months and couldn't lift his head or roll over and had head lag when you picked him up. Of course these concerns were blown off after I answered yes to the first time parent question.

Luckily, with some persuading from Zack's WONDERFUL pediatrician, Dr. Kamber, the local hospital agreed and the tests were ordered. A couple days later a **presumptive positive came back for LCHAD.** As you can tell I'm not that impressed with our local children's hospital but I will give them credit for diagnosing him quickly even though it took quite a bit of persistence to get the labs ordered.

No one at the hospital had ever heard of any FOD especially LCHAD which appears at a significantly lower occurrence than MCAD. In fact, when they came in to tell us the lab results the only information they had on it was what came from an outdated textbook from around the time LCHAD was discovered. This was very scary because at the time this was published it was thought that **greater than 90% of children affected by LCHAD would die by age two.**

As you can imagine Amber and I were heartbroken. However, after several days surfing the web, sending multiple e-mails to Valerie Fulton, and frantically making calls to Deb, I learned that this is only true if they are not diagnosed until severe liver or heart damage has occurred. Luckily, with Zack, this was not the case. I also learned that even with heart and liver damage LCHAD can be managed most of the time through proper diet and constant supervision by a professional who specializes in FODs.



Zack's Story... cont'd

Nothing else spectacular happened in this LONG hospitalization (3 weeks) except Zack did have a g tube placed and a nissen performed. The g tube has been a huge success however the surgeon failed to inform us that the nissen can cause something called dumping syndrome in which the stomach empties much faster than it should. This can cause a sudden spike in glucose levels which causes the body to produce too much insulin which drives the glucose down very low. Zack's levels were all over the place from less than 20 to over 300! Unfortunately, the normal treatment for dumping syndrome is eating high fat foods, which Zack obviously cannot have.

The local geneticist made a hard effort to try to research LCHAD but it was evident we needed to find a new doctor after all the trouble we had with the hospital and the fact that the local geneticist had never treated LCHAD. **After a very long search we found Dr. Hainline & Dr. Wappner who practice at Riley Children's Hospital in Indianapolis, IN.** The difference between the care he was receiving locally and the care at Riley was night and day. It is amazing there! I highly recommend this facility. They were even able to accommodate me with a broadband connection so I was able to work while Zack was in the hospital at Riley (Ok I am jumping ahead sorry!)

When we first saw Dr. Hainline in September Zack's LCHAD was still not under control. His liver functions and CK levels were still high. Also, Zack was going through the dumping syndrome cycle on a daily basis. His glucose levels during all this went as low as 20 or 30. Again, this was happening daily. **I know you're probably wondering why we didn't take him to the ER, etc. I want you all to know that we did. We took Zack to the local hospital 4 or 5 times in the month after he was diagnosed and called the local geneticist almost every day. They STILL blew us off even after knowing his diagnosis.** This was a horrible time in our life, we felt completely helpless and alone. There was no one we could go to for help and all we wanted to do was help Zack. It brings tears just thinking about this time. I don't ever want to go through something like that again. It was hell.

Needless to say when we told Dr. Hainline about all of this during our September visit all he could do was shake his head in disbelief. I felt so much better after this visit and was comforted in the fact that we now had a place that would help Zack. Because of less than impressive lab results Dr. Hainline thought it was best to admit Zack to get everything under control. What was only supposed to be a 3 or 4 day admission turned into another 3 week admission but it was well worth it. **During this hospitalization at Riley we discovered damage to his heart caused not by LCHAD but from untreated high blood pressure that the local hospital did nothing about!** Also, the mystery of his roller coaster like glucose levels were discovered when he was diagnosed with dumping syndrome.

Are you seeing the theme here...? **I can honestly say the things that cause us the most problems on a day-to-day basis aren't even caused by his LCHAD.** They are the things that the local hospital missed and didn't treat which became worse (ie. dumping syndrome, heart damage from untreated hypertension etc.). **One thing I learned from all of this is to always question what you are told.** Even the best doctor doesn't know all of the answers and you should always get a second opinion if your gut instinct tells you to.

However, **Dr. Hainline, Dr. Wappner and the wonderful staff of Riley Children's Hospital have done an awesome job.** For the most part Zack's labs are normal, the damage to his heart has stabilized and he has MUCH more energy. With the help of Amber and physical therapists, Zack is able to physically do most of the things that he should and he is very advanced mentally.

I now look forward to Zack being with us for many many years and for his health to continue to improve under the supervision of Dr. Hainline. We will get through this and I deeply appreciate the support we have received from the group. Thanks for all that you do and Happy Holidays and **remember, if you know something is going on and your doctor won't listen be persistent!**

Andy and Amber Weedman

Zack, LCHAD, 9 months

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More info and stories on LCHAD refer to www.lchad.com

LCHAD Email Network and New LCHAD website

Valerie Fulton (Adam, LCHAD, <http://adamslchad.com>) is email networking many of our LCHAD Families, just as Gina is doing with VLCAD. If you'd like to become a part of her email network contact Valerie at valchadmom@yahoo.com

Please be sure to visit www.lchad.com to learn more about LCHAD ~ **this site is in honor of Zack Weedman and was created by his dad, Andy.** As with the Email network, it's a great way to learn about LCHAD and to find support from other LCHAD Families going through similar experiences.



Medical Update

Procedural Changes for Mayo Supplemental NBS Kits

Mayo's Biochemical Genetics Lab has been experiencing many **problems (see bullet points below) with providing supplemental newborn screening (SNS) kits directly to parents**, and some of these are outlined below. **As a result of these concerns, a great deal of downstream work is generated for the laboratory and supporting departments. While we will continue to work directly with physicians, hospitals or other health care providers to offer SNS for their patients, the confines of our current system unfortunately do not allow us to continue to deal directly with individual families.**

We have explored the **options available to parents through Baylor Medical Center and Pediatrix Screening. Both laboratories have very efficient and effective systems in place that allow them to work directly with families** meanwhile minimizing the roadblocks created within our system. **As such, Mayo will no longer be offering parent kits and will provide callers with the toll-free numbers for both Baylor and Pediatrix.**

- As you are likely aware, patients have reported that when they ask their physicians about SNS the physicians respond in a variety of manners including "You need to call and get the information yourself" or "SNS isn't really necessary." Even when the parents obtain the information and kit, many physicians and hospitals refuse to facilitate the testing for their patients. In the majority of instances where this is occurring, they will assist the parents by collecting the specimen. The parents are expected to figure out how to send the specimen to us and deal with related logistics. Sadly, and perhaps even more frustrating for the parents, are instances in which the hospital and/or physician refuses to collect the specimen.

- For medical and legal reasons, we are unable to accept specimens directly from patients or family members. Results must be sent to a health care provider, not directly to patients. The Biochemical Genetics Laboratory at Mayo has tried to make accommodations for SNS by requesting that when parents are responsible for shipping the blood spot card, they include specific information regarding the referring physician/hospital so that there is a health care provider to receive results. Unfortunately, this information is not always accompanying the specimens. Due to this, SNS results are not getting to the practitioners or the families in a timely manner, which is upsetting to the families. In fact, in some cases, the physicians/hospitals are sending the results back to Mayo indicating that this is not their patient or that they didn't order the test. Luckily, we have not yet had an abnormal test result for one of these babies. If we had an abnormal test result it is possible that the result would have been delayed and in turn follow up testing and treatment could have been delayed as well. Regardless of whether the screen is normal or abnormal, families should receive the results in a timely manner.

- Our infrastructure is not set up to bill patients directly. When the physician and/or the hospital is unwilling to fully facilitate the testing process or when they do facilitate fully but are unwilling to receive a bill for services, we have requested that the family include payment in the form of a check when the specimen is shipped. As with the results, physicians/hospitals are sending the bills back to us indicating that they will not pay because this is not their patient or they did not order the testing. Payment is not occurring for a significant percent of SNS specimens we receive when the testing is ordered from parent kits.

- We have spoken with physicians/hospitals who have sent SNS samples to us in response to parent requests. In addition, we have had a number of instances in which a pediatrician requests testing for his/her own newborn. Upon further discussion with these providers, a) they are not willing to make this service available to all of their patients, b) they indicate that they will tell their patients about SNS and give parents the numbers for the various labs offering SNS, but they do not want to maintain a supply of screening cards for their practice, and/or c) they do not want to "choose a provider." This creates additional work and roadblocks for both the parents and the screening laboratories.

- Many times a parent will call just a few days before their delivery date and request that a parent SNS kit to be sent to them as quickly as possible. Unfortunately, we do not have a system in place to send the parent kits via overnight delivery. These parents are often frustrated for many reasons: 1) they found out about SNS from their provider at the last minute, 2) they have to do all the leg work to obtain information, compare labs, and get a kit, and 3) they are unable to get a kit overnight from Mayo. In addition, many parents find out about SNS after their baby is born and the optimal time for screening is missed (within the first 7 days of life) because they do not receive a screening kit quickly enough.

Please contact me if you have any questions or concerns.

April L. Studinski, MS, CGC
Genetic Counselor

Mayo Clinic College of Medicine, Biochemical Genetics Laboratory
voice: 507-255-3681 fax: 507-266-2888

<http://www.mayoclinic.org/laboratorygenetics-rst/biochemical.html>

Please ship clinical specimens to:

Mayo Medical Laboratories
200 First Street SW
Rochester, MN 55905
Telephone: 800-533-1710

Going to the Dentist

Heather Miller-Kuhncneck, MS OTR/L BCP

[Please note: This article was written for dentists; parents might find it helpful to print out a copy for their dentist, as well as read it themselves.]

Introduction

Sensory defensiveness, one type of Sensory Processing Disorder, occurs in children with a variety of diagnoses and disabilities. Therefore, in a dental practice that serves children with special needs, this disorder may likely affect some portion of your clientele. A thorough understanding of sensory defensiveness and strategies to alleviate difficulties caused by this disorder would be beneficial for dental professionals, in order to make the dental visit more pleasant for all involved. Therefore, this paper will present information allowing dental professionals to recognize the signs and symptoms of over sensitivity. In addition, helpful techniques to reduce the discomfort and anxiety of their patients with sensory defensiveness and increase compliance with dental procedures will be provided.

What is Sensory Defensiveness?

Sensory defensiveness has been defined as the behavioral indications of over-reactivity to common sensory experiences (Lane, Miller, & Hanft, 2000; Wilbarger & Wilbarger, 1991). Sensory defensiveness can occur in any of the sensory systems, of which there are really eight, rather than five. They are as follows:

- Tactile system (touch)
- Vestibular system (sense of movement in relation to gravity)
- Auditory system (sound)
- Visual system (sight)
- Proprioceptive system (position of our body parts, joints, and muscles, as well as the amount of force being used with movement)
- Gustatory system (taste)
- Olfactory system (smell)

Inner senses (hunger, elimination, etc.)

Typical over-reactions to sensations that others might not find noxious range from mild to severe, depending on the stimuli received and the overall amount of stimuli the child is being exposed to. The range of behaviors includes gaze aversion, physical withdrawal, blocking of the stimuli, vocal outbursts, aggressive behaviors, and tantrums.

A child with sensory defensiveness may exhibit the following during a dental visit:

- Tendency to pull away from or over-react to unanticipated touch, particularly touch to the face
- Over sensitivity to teeth cleaning by the hygienist
- Fear responses to moving backwards in the dental chair
- Difficulty tolerating the bright light above their head
- Fear responses to the noises of the dental equipment, including the polishing brush
- Fear responses to unexpected office noises, such as intercoms, door alarms, or beeps
- Extreme dislike of the polishing paste due, to the texture
- Over-reactive gag responses to dental tools or x-ray materials

Responses to the smell or feel of the glove materials

Fear responses may escalate to physical responses if the fear is not respected. Typically a child will demonstrate "flight or fight" behaviors. First, they will try to escape from the stimuli that are distressing, but if that cannot occur, they will become more and more physically reactive in any attempt to remove themselves from the situation. A child may be able to tolerate one type of stimuli but become more and more agitated if multiple stimuli are added.

Intervention Strategies for Sensory Defensiveness

Sensory defensiveness is often treated with two types of sensory input: deep touch pressure and heavy work. Deep touch pressure is firm touch provided to the skin by way of massage, vibration, brushing, lycra clothing, ace wraps, sandwiching between pillows, heavy weighted clothing, or lying under something heavy. Heavy work includes any activity that provides resistance to the muscles and joints of the body. Activities such as pushing or pulling something heavy, hanging from a trapeze bar, jumping, lifting or carrying heavy items, or squeezing something against resistance can all be considered heavy work. Using deep touch pressure and/or heavy work before and during distressing events can help calm a child with sensory defensiveness. See the box below for specific ways to use these techniques before or during a dental visit. Lastly, a child with sensory defensiveness will best be able to handle discomforting inputs when they are not unexpected. Using verbal preparation can be very helpful. Before doing anything that involves distressing sensory input, warn the child that it is about to occur so they can be prepared and not startled. Also, giving a set time limit that the input will occur may also be helpful (i.e., "we are going to do this until the count of 20," or "we'll be done when the clock says X," etc.).

Suggestions for Reducing Sensitivity During Dental Visits

- Have the child wear the X-ray vest during the entire appointment to provide extra weight and deep pressure.
- Have the parent do oral deep pressure or vibration in the form of electric toothbrush, mini massager, or rubbing with toothette prior to appointment.
- Have the child eat something very chewy prior to the appointment.
- Allow the child to have a fidget toy that provides "heavy work" during the session.
- Have the child wear a heavy or tight hat before and during the appointment.
- Use firm touch whenever touching the child.
- Verbally warn the child before each thing you do.
- Allow child to wear something that blocks the bright lights if he/she is sensitive to this.
- Allow the child to listen to calming music over headphones if the office noises or equipment noises are distressing.

Conclusion

It is difficult for individuals with sensory defensiveness to cope with the fact others do not share their discomfort and others actually may enjoy situations that they find so upsetting. For a child with sensory defensiveness who may not be able to verbalize or even recognize the problem, the accompanying feelings of anxiety and frustration can be overwhelming. Therefore the impact on functional behavior can be significant. Having a dental professional who is understanding and attempts to make the experience as comfortable as possible by respecting their fears and reducing the level of stimuli that is distressing may make the difference between a visit that is successful and one that is not.

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About the author:

Heather Miller-Kihaneck currently teaches in the graduate occupational therapy program at Sacred Heart University in Fairfield, Connecticut. She has practiced in pediatrics for years, and has specialized in school-based practice and outpatient occupational therapy using Ayres' sensory integrative approach. She has edited a book on occupational therapy for children with autism, and has been the quarterly editor for AOTA's School System Special Interest Section. She is currently developing an assessment tool to examine behaviors indicative of sensory integration dysfunction in the school setting. She can be reached at Hmilleyt@yahoo.com or kihaneckh@sacredheart.edu.

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Nutritional Update and Recipes

For our July Newsletter ~ Dr Phyllis Acosta (recently retired from Ross Metabolics, Abbott Labs) will share an article on FODs and Nutrition.

L.C fat free Gingerbread Boys (MCT oil version)

Wisk to gother in med. bowl:

3 tablespoons MCT oil

3 tablespoons applesauce
1/2 cup sugar

2 tablespoons dark molasses

2 tablespoons Second Nature Eggs

Mix to gother in small bowl, then add to the above mixture:

1 5/8 cups flour

1 1/2 teaspoons baking soda

1/4 teaspoon cloves

1/4 teaspoon ginger

1/2 teaspoon cinnamon

1/4 teaspoon salt

Stir the mixture until well blended. Chill 4 hours. Roll out to scant 1/4" thickness on floured surface, cut cookies. Place on very lightly sprayed (oil baking spray) cookie sheet and decorate with mini chocolate chips *for eyes and buttons. Bake 8 minutes at 375 degrees. Remove immediately from cookie sheet to a wire rack to cool. Add other decorations as desired. The dough is somewhat sticky, so the big key to rolling them out is working quickly while the dough is COLD, flouring the surface and rolling pin as needed. Makes about 15 4-inch gingerbread boys. **Low Fat with MCT oil. Crispy outside, tender inside.**

Nutritional info per cookie: Calories: 79; Fat (not including MCT oil): 0; Cholesterol: 0; Sodium: 14 mg; Total Carbohydrates: 18g (Fiber: 0, Sugars: 7g); Protein: 0g. *Mini choc chips have 4 grams fat per tablespoon. Each tablespoon contains 45-50 chips, so the fat from the choc chips is extremely minimal.

Yummy Chocolate Banana Cake

2 cups unsifted flour
 1 1/4 cups sugar
 2/3 cup unsweetened cocoa
 1 1/2 tsp baking soda
 1/2 tsp salt
 1/2 tsp cinnamon (or more if you like)
 4 egg whites
 1/2 cup Safflower oil
 1/2 cup water
 1 tsp vanilla
 1 cup plain nonfat yogurt
 1 banana, mashed

Spray 13x9x2-inch baking pan with cooking spray. In medium bowl stir flour, sugar, cocoa, baking soda, salt and cinnamon. In large bowl with mixer at medium speed, beat egg whites, safflower oil, water and vanilla until blended. Add yogurt and flour mixture, beating until smooth. Beat in banana until smooth. Pour into prepared pan. Bake in a 350-degree oven 40 to 45 minutes or until cake springs back when lightly touched. Cool in pan on wire rack. *(For those who can use MCT oil, it can be substituted for the Safflower oil, making this a practically 0-long chain fat recipe). This recipe works very well as cupcakes...I usually have some gem-size ones frozen, ready to pull out when others in the family or in a class are having cupcakes. One gem-size cupcake has approx. 1.5 grams fat, as made in the original recipe with safflower oil.

Both Recipes by: Diane Nielsen (Stephen, age 3, TFP)
valanddiane@mstar2.net

MCT Oil was purchased by Novartis Medical Health, Inc from Mead Johnson & Company effective February 13, 2004. MCT Oil is a modular source of MCT (Medium Chain Triglycerides) for patients unable to digest or absorb conventional fats. Consumers should consult with their physician on the use of this product. Refer to <http://www.novartisnutrition.com/us/productDetail?id=593&source=summary> for more information. NOT to be used by MCAD-ers. Some Families have found an **alternate MCT Oil through Sound Nutrition**. It is called original Thin Oil and is basically the same quality as the other Oil. It is less expensive by the case (12 16.7, 500ml bottles) - it costs about \$7 a bottle. To order just call **Sound Nutrition at 1-800-437-6863**.

'Fundraising' Information

If you'd like to participate in a project that would help with copying and printing costs for our newsletters/Family Packets (although the donations would NOT be tax deductible), please read our Jan 2004 issue online ~ individual members of our Group have several projects going on with Pampered Chef, Tupperware, iGive, and PartyLite Gifts.

VLCAD Email Network

Gina Revinski (Brett, VLCAD) is looking to start an FOD subgroup for VLCAD families. If you are interested in **networking with other VLCAD families around the world please email Gina at ginamjb@optonline.net** or call her at (845) 928-9574.



question &
ANSWER



Q: Does anyone have a suggestion for helping school teachers and others that take care of our FOD children, what to look out for (as far as our children getting sick etc) so they can contact us sooner rather than later?

A: One of our Parents shared with us a form (see below) she created to help her child's teacher determine if he/she should call the parent in case of emergency. The form helps the teacher base the decision on observations ranked from early, more serious, and acute, as well as the actions that should be taken to get her child help as soon as possible. This form may be specific for TFP but you can individualize it for your own child's disorder and how he/she behaves when starting to feel sick or low on fuel.

Behavior and signs to watch for when affected by TFP (Energy demands outstrip supply, body unsuccessfully tries to burn fat, creating toxins, leading to high acid/ammonia levels, and hypoglycemia. Sometimes poor behaviors precede a low blood sugar.)

<p>Early ("Brownout"):</p> <p><i>Verbal:</i> Becomes mostly non-verbal. Can become whiny, difficult to satisfy. Cries easily over simple problems. When Mom is not present he is more likely to just become very quiet and non-participative.</p> <p><i>Cognitive:</i> May act like he is only running on 4 cylinders instead of 8. Slower to respond. May gaze off into space.</p> <p><i>Physical:</i> Tired, usually wants to be held and lay his head on shoulder and/or lie down and take a nap. Weaker than normal. Head may list to one side. Usually has poor eye contact. Sallow, sunken look in eyes, slight under-eye puffiness.</p> <p><i>Social:</i> May be anti-social. May hit, scream, or be aggressive toward others.</p> <p><i>Eating:</i> Fickle. May want to eat, may refuse to eat/drink. Doesn't really know what he wants.</p> <p><i>Glucose levels:</i> Below 80, however we have seen problems even in the low 80's.</p>	<p>Action required:</p> <p>-Should drink 3-4 oz. of either Portagen, white grape juice, or apple juice. Pedialyte or Gatorade okay to try. Portagen is first preference if available because it keeps (Child's) blood sugars more even. If he refuses or is unable to drink because he is too tired, fluid should be given immediately through g-tube, whether he is asleep or not. He can handle about 2 oz. fairly quickly, and then another 2 oz. after 10 minutes.</p> <p>-If (Child) does not seem better in 20 minutes, check glucose levels with finger stick. Note: Glucose levels are not always the best indicator that there is a problem with energy processing in TFP, but can be helpful at times. General observations are best.</p> <p>-May need to rest, lying down or sitting quietly with a book, for a minimum of 20 minutes.</p> <p>-Notify Mom. Cell: _____ Home: _____ Dad's work: _____</p>
<p>More Serious: (Any of the following)</p> <ul style="list-style-type: none"> -Glucose in low 70's > 60 minutes despite above efforts -Lethargic -Stomach hurts (usually in liver area) -Urine may seem very concentrated and/or brown -Diarrhea in addition to any other symptoms -Brownout complicated with other illness (cold, flu, infection) -Gaggy/nauseous – Unable to tolerate fluids -Complaining of leg pain 	<p>Action required:</p> <p>-In addition to the above actions, he must lay down. Continue to deliver 2 oz./15 min of Portagen or Apple juice as tolerated.</p> <p>-If Mom or Dad cannot be reached, and (child) does not feel and look better within 60 minutes, call (Pediatrician). Call 9-1-1 if he instructs, or if he cannot be reached, and follow emergency protocol. – (Child) may be taken to (Hospital).</p>

<p>Acute:</p> <ul style="list-style-type: none"> -Vomiting with glucose in low 70's > 1 hour. -Cannot wake him -Seizure-like twitching of arms or head -Eyes rolling into back of head -Lips look blue -Respiratory failure -Heart failure/cardiac arrest 	<p>Action required:</p> <ul style="list-style-type: none"> -Start CPR if needed, have someone call 9-1-1 immediately, and follow emergency protocol. -Notify parents. -(Child) may be taken to (Hospital) <p>Approved by Doctor _____</p> <p>Date _____</p> <p>Child's Name _____ Disorder _____</p>
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NBS Update

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

There are MANY Families out there doing great things to raise awareness and funds for expanded NBS. One unique fundraiser that was brought to my attention a few months ago was called 'Katie's Ride' in memory of Maureen and Rick Boiros' daughter, Kathleen Celeste Boiros ~ Katie. Katie was born Oct 24, 2003 and died suddenly 8 months later due to an undiagnosed FOD. Katie's aunt, Pam Boiros, and many family members and friends participated in the June 13th Memorial Ride through scenic Massachusetts' back roads (<http://katiesride.net4world.net>). They also held a silent auction, raffle, climbing wall, face painting, and a ride on Mt Wachusett's ski lift! **This event raised \$10,000 for Women and Infants Hospital in Providence, Rhode Island, and also raised awareness of the importance of infant screening and the existence of these metabolic disorders. This was and will continue to be a very loving and touching way to keep Katie's light and memory alive!**



Pharmaceutical Update



Sigma-Tau Pharmaceuticals Remains the World Leader in Carnitine and Metabolic Disorder Research

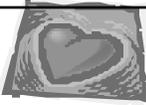
"At Sigma-Tau Pharmaceuticals, we believe quality of life is precious and no patient should be overlooked." This is the mantra of this pharmaceutical company dedicated to the development of orphan drugs for small numbers of patients whose pharmaceutical needs might not otherwise be met.

In 1985, Sigma-Tau received FDA approval for Carnitor® (levocarnitine) for the use of certain inborn metabolic disorders – and over the years Carnitor has helped many children with these rare disorders live a better life.

It is important to remember that Sigma-Tau is the worldwide leader in carnitine and metabolic research and ranks among the top 25 niche companies in the world in patent activity. More than 20 percent of the company's revenue is devoted to clinical research, a rate that exceeds the industry average.

Payer coverage and reimbursement for Carnitor® (levocarnitine) will depend on the patient's prescription coverage plan. Except in cases of specific policy exclusions, most insurance companies provide coverage for Carnitor® (levocarnitine). For information on reimbursement for Carnitor® (levocarnitine), or to find out if Carnitor® (levocarnitine) is covered by your prescription plan, please contact the Carnitor® (levocarnitine) Reimbursement HotLine at (800) 490-3262 or email us at CarnitorReimbursement@PharmAnalysisGroup.com. For more information about Sigma-Tau Pharmaceuticals and Carnitor,

please visit www.carnitor.com.



Love Messages

Jaan and Tim Aalberts

Luke - Birth June 7, 1994 Death May 1, 1995

Sandy and Howie Aitken

Kristopher - Birth Dec 22, 1997 Death Dec 29, 1997

Jeanne and Mark Barilla

Michael - Birth Feb 2, 1990 Death Nov 25, 1990

Jodi and Wayne Barnes

Amy - Birth Feb 20, 1995 Death Sept 27, 1995

Baby Barnes - Death in-utero Oct 7, 1999

Delane and Althea Becker

Warren - Birth June 9, 1987 Death Feb 4, 1990

Sue and Jim Berneski

Michael - Birth Dec 28, 1992 Death Jan 3, 1993

Jennifer and Bill Boucher

Alyssa - Birth Nov 18, 1999 Death July 22, 2000

Jacque and Mike Bradford

Eric - Birth Aug 22, 1990 Death Aug 26, 1990

Cynthia Brown

Miranda - Birth Death April 3, 2004

Joseph and Barbara Brown

Amber - Birth June 18, 1989 Death May 17, 1990

Barry and Julie Bryson

Skyler - Born November 25, 1989 Death Aug 25, 1990

Cardien Grootaert - Callens

Liese - March 2, 1988 Death Aug 25, 1990

Tom and Lynn Camino

Stephanie - Birth June 28, 1995 Death Feb 6, 1996

Claudia and Antonio de Carmo

Bruna - Birth July 22, 2003 Death Oct 31, 2004

Mark and Karen Carpenter

James - Birth May 7, 1985 Death Dec 6, 1986

Jenny and John Carroll

Sarah - Birth March 4, 1992 Death Sept 1, 1992

Mark and Diane Casey

Matthew - Birth Apr 15, 1974 Death Jan 13, 1975

Jennifer - Birth Oct 19, 1975 Death Nov 13, 1979

Lori - Birth Aug 31, 1980 Death July 1, 1984

Tammy and Roger Clark

Jenna - Birth Feb 17, 2002 Death Nov 22, 2002

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

Valerie & Chris Ciachette

Benjamin - Birth Jan 12, 1987 Death April 18, 1987

Toni and Mark Cline

Kasie - Birth June 6, 1990 Death March 10, 1991

Sandy and Jon Cooper

Noah - Birth Oct 5, 1997 Death June 30, 1998

Martin and Kathy Davis

Mary Katherine - Birth June 27, 1996 Death Nov 7, 1996

David and Amy Deshais

Megan - Birth Feb 11, 1991 Death July 7, 1991

Doug and June Evenhouse

Marie - Birth Dec 15, 1985 Death Nov 19, 1986

Carolyn and Terence Finn

Emily - Birth Feb 13, 2002 Death April 3, 2004

Andrea and Phillip Franklin

Brandi - Birth Dec 2, 1986 Death Jan 1988

Lance and Dawn Goldsmith

Joel - Birth Feb 15, 1990 Death Nov 16, 1990

Deb and Dan Gould

Kristen - Birth Oct 6, 1983 Death July 21, 1985

Shelly and William Grabow

Caleb - Birth Sept 14, 2001 Death Sept 27, 2001

Noah - Birth Nov 18, 2003 Death March 23, 2004

Brandis Greichunos

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

Jeannette and Keith Guillory

Dominique - Birth Jan 21, 1997 Death Jan 23, 1997

Nicole and Chris Gulinello

Alec - Birth Feb 21, 2001 Death Aug 24, 2001

Michael and Nicole Gumiel

Michael - Born March 28, 1998 Death April 4, 1999

Carol and John Hall

Sarah - Birth June 8, 1998 Death July 30, 2000

Robin and Vince Haygood

Ben - Birth Feb 19, 1998 Death Aug 8, 2000

Ralph and Angie Hedrick

Chelsea - Birth Jan 11, 1995 Death Apr 3, 1996

Nikki and Toby Hiatt

Reece - Birth Aug 1998 Death April 18, 1999

Pauline and Bill Hill
Rosemarie Rees - Birth Apr 15, 1976 Death Dec 23, 1999

Amy and Mathew Hoffman
Grace - Birth Jan 25, 2001 Death Oct 25, 2003

Brad and Kim Holmes
Brittany - Birth March 20, 1991 Death Feb 21, 1997

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

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

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

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

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

Brian and Patricia Karhu
Patrick - Birth July 15, 1996 Death July 28, 1997

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

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

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

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

Sondra Koehn
Darcy - Birth Aug 10, 2000 Death March 19, 2002

Jamie and Tom Lazzaro
James - Birth Dec 8, 1996 Death Aug 13, 1997

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

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

Darlene and Larry Lopez
Marissa - Death Feb, 1999

Heather and Phillip Marsella
Toni Marie - Birth Oct 8, 1990 Death March 22, 1991

Ron and Paula Matthews

Randy and Misty McDonald
Jeremiah - Birth April 3, 1991 Death Dec 15, 1991

Christine and Mark McFarland
Erin - Birth Aug 26, 1988 Death Jan 21, 1989

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

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

Lori and Jeff Michaud
Jordan - Birth Feb 19, 1997 Death March 21, 1998

Simone and Michael Miller
Michael Dylan - Birth Aug 24, 1991 Death Aug 24, 1991

Mike and Sheryl Mulhall
Justin - Birth April 22, 1990 Death April 22, 1990
Nathaniel - Birth Aug 15, 1991 Death Aug 18, 1991

Verna Parker
Charles - Birth Oct 24, 1988 Death Oct 26, 1988

Diana and Kevin Patterson
Trevor - Birth Aug 30, 1997 Death Mar 4, 1998

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

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

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

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

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

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

Brian and Cheryl Rosenberger
Kylie Ann - Birth Feb 7, 1990 Death Feb 11, 1990

Jani ce and Steve Rowland
Josey - Birth Sept 30, 1996 Death Oct 28, 1998

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

Jackie Shears
Timothy - Birth Oct 3, 1991 Death Oct 4, 1992

Lisa and Scott Sleezer
Emily - Birth Mar ch 5, 1998 Death June 18, 2001

Leah and Paul Sofranko
Kyle - Birth Feb 7, 1988 Death Feb 5, 1989

Rhonda and Matt Southard
Trace - Birth May 2, 2000 Death Aug 26, 2000

Janna Sowers
Kelsie - Birth April 23, 1993 Death April 23, 1993

Anne and Gary Stitt
Sydney - Birth Oct 13, 1995 Death May 20, 1996

Lisa and Doug Tennyson
Sammy - Birth Aug 23, 2000 Death Dec 21, 2001

Rick and Stephanie Thomas
Trina - Birth July 1977 Death Jan 14, 1978

S. Elizabeth & G. Douglas Turman
Philip - Birth April 6, 1994 Death April 8, 1994

Darren and Karen Wade
James - Birth Jan 15, 1996 Death Aug 31, 1996

Sirpa and Jay Waananen
Nora - Birth Mar 29, 2001 Death Aug 9, 2001



Jenni Wagoner
Lauren - Birth Oct 26, 1993 Death Nov 13, 1999

Richard and Amy Warner
Andrew - Birth May 1978 Death Nov 18, 1979
Scott - Birth May 1983 Death April 25, 1985

Denise and James Westman
Benjamin - Birth March 11, 1987 Death Dec 20, 1988
Beau - Birth July 10, 1991 Death Feb 19, 1995

Mike and Darci White
Brett - Birth June 14, 1993 Death June 17, 1993

Karen and James Whiteside
Caitlin - Birth Aug 12, 1996 Death Dec 19, 1996

Lori and Dean Williams
Brennan - Birth June 1, 1999 Death June 6, 1999

Christi and Ronnie Williams
Preston - Birth Mar 11, 2000 Death Mar 15, 2000

'I existed from all eternity and, behold, I am here: And I shall exist till the end of time, for my being has no end.'

-Kahlil Gibran, "Anthem of Humanity"

Condolences

We were very saddened to hear of the death of 15-month-old Bruna de Carmo (Unclassified FOD, Portugal) on October 31, 2004. Bruna's parents, Claudia and Antonio, appreciated everyone's prayers during Bruna's courageous fight to live. Our deepest thoughts and prayers go out to the entire de Carmo Family at this time of saddened hearts.

All of our FOD children will ALWAYS be with us in our hearts!



**** Greetings from NICHCY, the National Dissemination Center for Children with Disabilities.**

We're writing to let you know about a new information service we've launched called eNews. If you're looking for information and assistance on disability and education issues, then eNews is made for you. Literally. It's not a newsletter sent to the masses - you tell us what type of information you're looking for, and we'll send that type of information straight to your e-door. Want to sign up? Want to tell us what you need? Want to first learn more about eNews? You can do all that at this link:

<http://www.nichcy.org/SurveyIntro1.html>. We look forward to hearing from you. Come one, come all! eNews is free and available to everyone who's signed up. NICHCY staff

Welcome to new babies!

Leslie and Robert Zemeckis welcomed their new son, *Rhys Charles*, in September 2004. Big brother, **Zane** (MCAD) is thrilled!



Kids Korner



Dakota Eads (Unclassified)



The McConnell Family: Mike and Nancy, Meredith, Michael, and Morgan (LCHAD)



Jake Bray (LCHAD)



Kloey Cole (sister to David, MCAD)



Karli Lafferty (MCAD) and sister, KoKo



Adam Fulton (teen LCHAD)



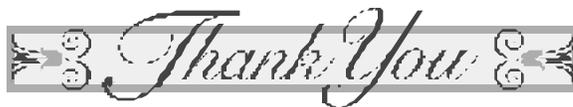
Family & Professional

Family Donations: Leslie and Robert Zemeckis, two donations in honor of Zane (MCAD) and their new son, Rhys Charles. Darlene Carson in honor of her granddaughter, Hayley Stuve (MCAD). Betsy and Eric Furler in honor of Henry (Unclassified FOD). Jean and Chris Robeson in honor of Steven (still testing). Sheri and Derek Sandstrom in honor of Carter (MCAD). Kim and Nick DiPaolo in honor of Gareth (MCAD). Louise Ramos in honor of Joseph (still testing). Pam Wilson in honor of Josh and Jacob (both SCAD).
iGive Donations (by shopping online) by Kim DiPaolo, Jennie Heiner, Anna Taiser, and Cindy Wilt. Terry and Patricia Kalhar, Janet and Thomas Lindsay, Staff at Harvey W. Scott Memorial Library of Pacific University, Julie and Jon Larson, and Jan and Daryl Maloch in memory of Matthew Christerson (adult VL CAD). Hugo and Mary Holm in memory of their grandsons Matthew Christerson (adult VL CAD) and Eric Bradford (Undiagnosed MCAD). Stephanie and Andrew Plaisted in memory of Drew (undiagnosed MCAD).

Thank you to all that have bought products from companies on the internet that support the iGive program of donating a certain percentage to Groups like ours. Please remember, however, that **donations are NOT tax-deductible because we are not a nonprofit organization.** For more info on the iGive program, visit <http://www.iGive.com/html/refer.cfm?causeid=24970>.

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

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



Thank you to Erika Wallace - erikawallac epa@yahoo.com (Mailing Lists), Mary Lingle - Mcartwrite@aol.com (Web Page) and Brian Gould - briangould@triad.rr.com (newsletter) for all your hard work. Special thanks to Sigma-Tau Pharmaceuticals, Inc. for their continued financial support.

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



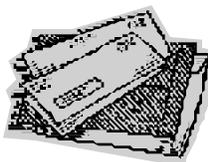
Reminders

Families - Please send TYPED stories by JUNE 1, 2005 To be listed on the FAMILY LIST, please return the SIGNED Family Questionnaire or hand-write your information as seen on the current Family List and sign and date it. Continue to spread the word about FODs and the need for screening -- it will SAVE LIVES!

Professionals - Please let us know about your research and/or clinical work with FOD Families. Send articles by JUNE 1, 2005. Also, please return to Deb the Professional Questionnaire even if you are already listed on the printed Professional List.

'When the power of love overcomes the love of power, then there will true peace.'

- Sri Chin Mā GOsh-



Communicate With Us

Please **ADD** me to your mailing list
 Family Professional (**please circle one**)
 Name/Address or Address Correction (**circle one**)

Please **REMOVE** me from your mailing list:
 Name/Address:

Please include ideas for future issues or your questions Page 17