Happy New Year to everyone! The Organic Acidemia Association [OAA] and the FOD Group are in the process of asking Speakers to address our Families and Professionals at the Red Lion Hotel on the River [general hotel info] in Portland, OR for our 2012 FOD/OAA National Metabolic Conference on JULY 20-21, 2012. Our Premier Sponsor is the OHSU - Oregon Health & Science University Department of Medical & Molecular Genetics ~ THANK YOU! We are also soliciting more sponsors and donors, setting up an online Registration form and the Hotel online reservation page. There will be NO Registration fee for Families. However, Professionals will be charged $50 per person.

Attendees are responsible for making their own Hotel reservations. Prices will be in effect 2 days before and after the conference dates of July 20 and 21, 2012.

Phone Hotel Reservations: If you call 503.283.4466 be sure and state your reservation is for the FOD/OAA National Metabolic Conference so you will be given the reduced conference rate - starts at $99 - $114 depending on room size. If you have special needs (i.e., refrigerator, microwave, crib, etc) please call and ask about availability and/or extra cost.

Red Lion Hotel on the River is 15 minutes from the Portland International Airport and there is complimentary on-call airport transportation, as well as other ground transportation services. Ask about the Hotel's airport service if you call for a reservation.

Travel: Attendees are responsible for making their own travel reservations. Speakers will work through Deb and Kathy for both Hotel and Travel arrangements.

Meals: We will be working with the hotel chef to provide a wide variety of food offerings for Friday and Saturday breakfast and lunch and the Friday night Reception for all Registered attendees, as well as lowfat and low protein options to accommodate our Families' needs. Once we have the Registration Form online (@Feb 1) you can state if special food restrictions are needed and the Hotel chef will try and accommodate as much as possible.

Registration Form: Will be online soon - Only Registrants will have access to our Conference Breakfast and Lunch on Friday and Saturday, as well as the Friday night Reception for Families, Professionals, Exhibitors, and Speakers. All individuals older than 5 yrs old MUST be registered.

Child Activity Room: We hope to provide an activity room for children - however, each family will be responsible for providing someone to supervise their child (i.e., grandparent) and that person will also need to be Registered. Volunteers will help in the room to provide drawing activities, games, videos etc. Children will be allowed in the conference sessions IF they can do so quietly. Otherwise they will need to be removed from the session. Older children/teens are encouraged to attend sessions - every child over AGE 5 will need to be Registered if they plan on having the Friday and Saturday Breakfast and Lunch and the Friday night Reception.

Please also continue to create awareness of FODs with your family, friends, and medical professionals, as well as create your own ways to raise funds, via ‘Family Fundraisers,’ so we can continue to spread the word about FODs via our website, Conferences, speaking at hospitals, and other various ways that allow us to offer all of our services free of charge. Also, when buying online please remember when you use the iGive link on our site, the FOD Group gets a percentage of your sale. We also earn funds by using GoodSearch as a search engine, or using the Donate button on our site or on our Facebook Cause page. You can also order your very own embroidered or screenprinted FOD polo shirt, cap, or any other item of various colors from the same embroidery company that I purchased our Speaker shirts from the 2010 Conference! They have our logo on file.

Families ~ We welcome ALL new or updated Family Stories and pictures and we encourage Families dealing with the less common FODs (i.e. HMG, GA2, Carnitine Uptake Defect, TFP, CPT 1&2 etc.) to share their experiences. We’re also always looking for more low fat recipes, poems, ‘Silver Linings,’ pictures, and ‘Reach for the Stars’ accomplishments of our kids/families.

Professionals ~ we need to hear from you too! New Medical, Research, Nutritional, Counseling/Coping, etc articles are always appreciated.

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

‘We Are All in This Together!’

Take care… DLG
It is wonderful that one of our moms, Stephanie Harry, has written a paperback book that will help educate many about Fatty Oxidation Disorders and how it feels to be a child living with a long chain disorder. The book comes out of her experience with her son, Christopher, whose story is on page 3. Please read Stephanie’s information below and if you receive one of the books be sure and send her much deserved positive feedback for creating awareness of FODs! DLG

I am so excited to share that after two years of thought and preparation My Special Body, a children’s book for kids with LCHAD, VLCAD and TFP is completed and in print. The idea for this book came in the first year of my son’s life, as I was trying to brainstorm how to talk with him about his metabolic condition. Although art is something I love and am passionate about (I am a potter), writing and illustrating a children’s book was definitely new and challenging! My hope in creating this book was that young children would have a safe place to explore their condition and that parents would have thoughtful questions to engage their kids around their diet. Obviously we as parents of FOD children think about diet and food everyday, but it can be challenging to know what to say and when to say it. I think books are a great way to talk with children about important and difficult topics, because they know that when they are done with the book they can have a certain level of space from the issue at hand. Also, I wanted Christopher, my son, to be able to read about another character whose body was created like his so as to create a sense of normalcy around his condition.

My Special Body is specifically geared toward 3-6 year olds with LCHAD, TFP and VLCAD. The book showcases Andy, who shares with the reader about his “special body.” He shares his condition’s name, the foods that are good for him, the difference between medium and long-chain fats, the importance of MCT oil and medical formula in his diet, how to read nutrition labels, and how to sort through which foods he can eat. The book has engaging questions on the left, with the main text on the right. There are also activities in the back of the book to continue conversation about diet as your child gets older. I do want to note that this book discusses a lot about the importance of MCT oil in the diet and medium chain fats being good, so unfortunately this book would not be appropriate for children with MCAD.

The book was originally published by a grant. We were excited to provide initial copies of the book to families at no cost through this grant. With the grant running out we are seeking to make this project more sustainable, therefore we will have to start charging for copies of the book. We are asking for a $10 donation per book. This price includes shipping (if you are outside of the United States and Canada it would be helpful if you could send $3.00 extra to compensate a little of the shipping cost). The small profit of each book (approximately $3.00), we are discussing how to best distribute. I will keep you posted on these logistics, but our hope is to create a fund to do future book projects for other FODs and use any surplus toward FOD research. I do realize that in these economic times it may be challenging for some families to buy a book. I do not want finances to prevent anyone from having a copy, so if you feel like you cannot cover the cost please e-mail me personally and I will make sure that you get a copy for free!

Our hope is that in the next month we will have an order form set up on the following web sites: http://southeastgenetics.org and http://genetics.emory.edu/nutrition. So you should be able to go there, find the book, fill out an order form, and send a check or money order to the address given. I have also talked with Deb Gould about setting up a link through the FOD website and she has graciously agreed! In the mean time if you would like copies of the book feel free to go ahead and e-mail me, I will get you more info about where to send payment and make sure that you get your copies! If you gave me your address to get a free copy over the holidays they have been shipped so they should be there any day! I would love to hear from you about what you think! My e-mail address is srharry374@hotmail.com. Lastly, I will be at the conference this summer in Portland! So I will make sure to bring extra copies there too!

Much love! And I am so thrilled to be able to share this with you all! The e-mails I have received so far from families have been so encouraging!

Sincerely,
Stephanie Harry
srharry374@hotmail.com
Five years after our marriage, my husband Ryan and I got pregnant and were excited to start our family. I suppose many couples imagine what childbirth and rearing might look like, and we were no different. We spent months going to Bradley Method classes hoping to have a natural childbirth, and spent quite a bit of time reading and talking about the changes that would occur in our lives. As we waited for the birth of our child there was much excitement, anticipation, hope and expectation about what lie ahead. We imagined that after the birth of our child we would gather in our birthing room, drink sparkling cider with our friends and give a toast to our new child.

Much to our surprise Christopher Ryan Harry was born on August 11, 2008 through emergency c-section. My liver and kidneys had begun to fail, so in an attempt to save my life they delivered my son. Christopher did quite well in the NICU and Ryan often talks about how he was one of the largest babies there (at 18 inches long and 5lbs 3ozs). I, on the other hand, was fighting for my life. My kidneys had completely failed, my liver was not working and my body went into DIC. I was put on life support, where I stayed for eight days in a coma. During that time I was given a product called factor seven. It helped my blood to start clotting again, and slowly I began to recover.

The first time I saw Christopher was 10 days after he was born. The amazing NICU staff and my ICU nurses worked hard to make it possible for Christopher to come down and see me the day after I was taken off of life support. Although I do not remember this, the pictures and video that was taken at this time still brings tears to my eyes.

The first day I remember seeing Christopher and his first year of life, are far from what Ryan and I could have ever imagined for our family. Soon after I awoke from life support we were told that we couldn’t have any more children, that I couldn’t breastfeed and that our child had LCHAD. Ryan went on medical leave from his job for three months to help care for Christopher and I. When I left the hospital I was still fighting an infection in my bloodstream and was trying to re-learn to walk. In the midst of all of the challenges of our birth experience and trying to acclimate ourselves to the constant feedings of our child, Ryan and I quickly saw that we had so much to be grateful for. We were so encouraged as family, friends and neighbors offered their time, care and support. Although Christopher couldn’t receive the initial doting we had hoped for, he received plenty as the year moved forward.

Christopher continues to grow and is doing very well. So far he has been able to by-pass hospitalization due to metabolic crisis and we are amazed at how he is thriving. His joy for life is what stands out to me the most. If he finds something funny he doesn’t just laugh and move on to something else, he is on the ground rolling in uncontrollable laughter. He loves music and will sing while doing a tambourine march around our house. It is hard to know how to capture the vibrancy of a child in words, but he shakes your soul at the core and makes you smile.

We are strong believers that he will be his best advocate growing up, so we are trying to teach him as much as we can early on about his condition and food choices. He loves singing the “Fat Song” which teaches him how to spell the word fat so he can recognize it on food labels. We also try to talk about what is “Christopher Friendly” and what is not. His favorite question these days is, “Is that Christopher-Friendly?” I always try to put more emphasis on what he can eat rather than what he can’t because there are so many yummy foods that are good for him!

I think the biggest challenge for me right now is balancing the knowledge of his condition and diet management, with enjoying him and who he is at age 3. Although Christopher has stayed out of the hospital, his blood work has led to several dietary changes over the last three years. Sometimes this has meant adjusting daily consumption of fat, other times this has meant adjusting MCT oil, and still other times this has meant changing his cornstarch intake. I often feel like a “fat-o-meter” and “energy-monitor” trying to make sure that all of his dietary needs are met. All the while Christopher is bopping around me, smiling and enjoying life. I often find myself asking, “Am I enjoying life enough with him? Or do I spend too much time worried or discouraged about the future?” Balance is such a difficult thing to obtain.

Thank goodness that I have Christopher to keep me in line. When I get all too serious or frustrated with his condition, he runs up to me in his fireman hat and jacket and says, “Mommy you on fire?” And off we go, putting fires out and rescuing his favorite stuffed animals from the flames in his bedroom! And as I sneak a few kisses in from my fireman, I feel so grateful to have such an amazing son and wonderful family.

Stephanie Harry
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Family Stories

Cameron’s Story ~ CPT 2

The Day Our Lives Were Changed Forever By CPT 2

I recently returned to school to complete my degree and had to write a memoir. I thought that I would share it, in the hopes that it may encourage or help someone in the FOD Group.

When you are searching for answers you may not always like what you find. You may want to turn back the hands of time and take it all back. You have two choices, empower yourself, accept it, educate yourself, and move forward with confidence or you can live in denial. When my son, Cameron, was just four years old I found that answer that I was looking for—he has CPT 2. In simple terms, Cameron is missing an enzyme that helps him convert fat from food he eats into energy.

When I finally heard the words “Mr. Jenkins, we know what’s wrong with your son,” I breathed a sigh of relief. You may think this sounds strange, but I had already come to terms with the fact that Cameron was sick and there wasn’t anything I could do. I had come to terms with the fact that what was making Cameron sick might take his life. Finally knowing what we were dealing with, I didn’t feel so helpless anymore. This thing that was making my son sick finally had a name.

I am that guy that likes to know what he is dealing with. So I went on the internet and started doing research about CPT 2 because knowledge is power, right? It’s amazing how CPT 2 and a lot of the other fatty oxidation disorders have only been known about since the nineteen seventies [and several from just the 1990s]. What amazes me more is how little is known about these disorders and how many doctors have never even heard of them. When I take Cameron to the hospital I have to explain to the doctors and other staff what CPT 2 is because they have never heard of it.

When you hear about someone having an illness or a rare disorder like CPT 2, you think to yourself, “Oh my goodness that’s horrible. How do they deal with that?” I am here to tell you how we deal with that. We deal with it by taking it one day, one minute, one fat gram at a time. This disorder has actually made everyone in our family a better person, and made our family better. We eat healthier because of having to restrict Cameron’s fat intake to 22 grams a day. Most importantly however, because of CPT 2 and Cameron’s battle, my wife and I have both developed an interest in the medical field. My wife, Amy, returned to school to become a medical assistant and wants to get a job at the Children’s Hospital when she graduates. I returned to school to finish my degree and changed my major to health information systems.

When you’re searching you may not always like what you find. But what matters is what you do with it when you find it.

James D Jenkins
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Ella Grace’s Story ~ MCAD

Our dear Ella Grace was born in March, 2005. Little did I know that the birth of my firstborn daughter was going to challenge me in ways I had never been tested...both physically and emotionally. Naively, I never entertained the possibility that we would have anything but the perfect first time parenting experience.

Early in my pregnancy I saw an episode on the Today show about supplemental newborn screening. Casually, I decided to order the testing kit, as it was inexpensive and performed by Baylor, a local hospital. It seemed like the proactive and responsible thing to do, yet I can’t say I really knew much about it. Little did I realize how vital that test would prove to my daughter’s life.

Several weeks after Ella Grace was born, our pediatrician called to inform us that one of her screenings came back positive. Although I was surprised, I didn’t worry too much, as no one else seemed panicked. We met with the metabolic geneticist nearly 6 weeks after Ella Grace’s birth and learned she had something called MCAD. I frantically searched for information, learning everything possible. Fear engulfed my world after learning more about the condition. What if she had a crisis? Would I know what to do and when to go to the hospital? Day to day I felt vulnerable and very uncertain about my daughter’s care. I questioned everything, including the doctors. Treatment varied and answers weren’t certain. Reassurance was out of the question. Nighttime was most difficult, as we fed Ella Grace around the clock, yet worried that we’d miss a feeding and she would be another statistic.

[Cont’d on pg 5]
Routine visits to the doctor left me frustrated, as answers to my series of questions were vague. And, a part of me was angry, reflecting on the fact that Ella Grace’s diagnosis and first meeting with the doctor came several weeks after her birth – clearly the possibility of a metabolic crisis had existed.

Years later, my story has changed. Looking back, I recognize how much MCAD affected my learning curve of parenting. Fear lurked around every corner and wreaked havoc on my confidence. Years later, I have learned that the worries have their place, but they no longer control our life. MCAD is manageable, if detected. Ella Grace leads a normal life – she loves playing soccer, enjoys swimming, running and anything active.

Our story is about being very fortunate. We are fortunate her condition was discovered through supplemental newborn screening because, at the time, Texas did not routinely screen for MCAD. We are fortunate because in six years, Ella Grace has only been hospitalized one time. We have learned to be vigilant when she is ill, monitoring her a little more closely than our second daughter, Evelyn, unaffected by MCAD. We are fortunate because Ella Grace did not have a metabolic crisis the several weeks before our first meeting with her metabolic doctor. We are fortunate because, most importantly, we did not lose our child, as many others have before learning their child was affected by MCAD.

And so, I am beyond thankful. I am so incredibly grateful for the parents that have walked a difficult journey before us, losing their precious child yet having the strength to make sure others were diagnosed properly. My very humble thank you...

Beth Meyer
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Austen’s Story ~ GA2/MADD

I share the same story with many of you. The normal pregnancy, the c-section birth, the sight of that precious, little face and the feeling that finally, all was right with the world.

And then the shock.

My previous pregnancy, eight years back, was difficult. I had conceived triplets through the en-vitro process, after trying for years to get pregnant. I lost a baby after my first attempt so I was off my feet for this whole pregnancy and hospitalized four weeks before their birth at thirty weeks. Although they were preemies and had to stay in the NICU for five weeks before coming home, they had few problems once they were home and were all good nursers after they got the sucking reflex which happens at about thirty two weeks of age. I nursed all three of them for the first six months and when they went onto solids my kids were great eaters!

I was surprised that Austen had no interest in breast feeding and although the nurses said it was not an uncommon thing for a baby not to eat for the first few days, I remember being very concerned about his crying. He wailed like only a hungry baby could. When he was taken from my room so I could get some rest, I could hear his cries in the little nursery down the hall, louder than all the others. The second day was like the first, now I was worried. The staff seemed indifferent. He still wouldn’t eat, but he was clearly unhappy. That evening lying next to my bed in his little bassinet, his wailing forced me to get out of my bed to pick him up. The nurses were all busy and because it was a Friday there was lot’s of visitors and noise and rejoicing the birth of all the new babies.

Austen was eventually revived. The nurses said he was without oxygen for twenty minutes. I think it was longer because no one came to my room to tell me anything and I was very hoarse from wailing. He was transported by ambulance to New England Medical Center where he was stabilized from the seizures, put under oxygen and once again I had a baby hooked up to wires and tubes, but there was no excitement this time. He had suffered serious brain damage and on the third day of his stay in the NICU, I was asked to sit down with the doctors who had been following him.
Dr. A, the metabolic doctor who had been spending a lot of time with him, told me that he had a serious genetic defect. She called it Glutaric Acidemia type II or otherwise known as Multiple Acyl Co-A dehydrogenous Deficiency (MADD). She told me that it was a fatal disease and explained that he could not metabolize fats or proteins. She said that no other child born with this disease had lived for longer than six to eight months. She was patient with me when I cried. She said that she wanted to send a muscle fibroblast to a doctor in Iowa who would confirm the disease. She said that he could be brought home once he was taken off the tube feeding him through his nose and suck on his own. He should be given the best life possible in the next few months. It was best “not to resuscitate” should he stop breathing again, because of all the damage that he had already sustained. He was not to drink breast milk, too much fat and protein, so he needed a special formula.

Up to this point, I was in shock and depressed and felt very alone and defeated. But as I spent days with him watching as he came out of his long sleep and when he finally looked up at me and took formula from a bottle, something changing in my attitude. On the day we were told we could go home I became an angry, assertive woman, a person I had never been in my whole life. I demanded an apnea monitor to have at home in case he stopped breathing, I insisted that I would breast feed him and I didn’t want the formula, (after all if he was to be given the best life in six quick months, shouldn’t he be allowed to breast feed?)

He went home and I pumped and he learned how to nurse. I got in touch with the FOD (Fatty Oxidation Disorder) Family Support Group and got a home computer (one of my smarter purchases!). Austen got bigger and gained weight and I became more determined not to let the diagnosis defeat us. We got the confirmation from the fibroblast that he had “2% of controls…as low as enzymes get.” It was a mitochondrial disease and there was no cure except to continue with the carnitine and B2 supplements that were supposed to sustain him. I read Dr. Andrew Weil’s book, “Spontaneous Healing” and was particularly affected by the chapter that addressed malfunctions in DNA. He says that it can be reversed through diet especially by ingesting natural enzymes. I changed my diet radically after going to see a naturopathic doctor who put me on a meat-free, dairy-free diet which included mostly raw fruits and vegetables (both Austen and myself still follow this diet). I didn’t stop there, I sought out a Native American shaman, a Catholic priest who was a faith healer, a chiropractor…and a second opinion from another metabolic doctor.

From the first time I met Dr. K, I knew that we would be in good hands. He believed in treating Austen the individual, not the disease. His approach was much more hopeful and that is what I needed to keep going, especially since I was a single parent at this point. He marveled at his weight gain and cognitive abilities, although he was delayed, he seemed to be progressing. The one concern was that his head circumference had come to a halt. Dr. K prescribed CoQ10 and in the next few months after starting it, his little head started growing again, although he is still considered to be microcephalic. When we started with solid foods each meal was traumatic, because he would throw it up from reflux. I had to clean up the mess and start all over again, because I knew if he didn’t eat we would end up in the ER. He came to recognize that he had no choice in this food business, he had to keep it down!

I weaned him very gradually off of the barbiturates that he had to take for seizures and he has never had another one that I know of. Early on I recognized that Austen had a severe visual impairment and hooking up with Perkins School for the Blind got us involved with the infant toddler program, preschool, the ‘Lower School’ and last June he graduated to the ‘Secondary Program’. Social skills, PT, OT, sensory integration, mobility, music therapy, gym, arts, swimming and of course academics are only part of the total program. We have a skilled health care clinic on the grounds and we have been very lucky to live so close to the school and be part of this wonderful community.

Today, at fifteen years old, he is of normal height and weight, a handsome devil who is devoted to his older siblings, Nathan, Sasha and Taylor, and his loving stepfather, Joe. It has not been an easy road. We have had many bumps and starts. He stopped sleeping, (night and day) at about age three. Several trips to the ER after a vomiting bouts, severe sinusitis (now controlled with Periactin), incontinence still to this day…it hasn’t been easy. He has many food allergies which have been hard to decipher since he can’t really tell us ‘where it hurts’ and displays his discomfort through tantrums or negative behavior, although once we got the wheat, eggs, nuts, etc. out of his diet we have seen much less confusion and better spirits. The change in diet and recognizing that his behavior was on the autistic spectrum, (and getting the diagnosis of asperger’s syndrome), has afforded me with much more knowledge of how to help my son deal with this world that he doesn’t really understand, and help others to understand his world.

I feel strongly that the choices I have made for my son have been the right ones, but I could not have done it without the help of all the wonderful people I have met along the way and who now share a part of Austen’s world, and without them, I might not have made some of the connections that have made such a big difference in the quality of my son’s life. Thank you Deb, and all my FOD friends around the world.

Gwen Abele
Boston
gwenabele@hotmail.com
SUBJECT RECRUITMENT for FOD RESEARCH

APPROVED: Jun. 13, 2011

We are conducting a survey among patients diagnosed with either mitochondrial trifunctional protein deficiency or long-chain 3-hydroxyacylCoA dehydrogenase deficiency (TFPD or LCHADD). Our survey can be accessed at the following url: https://octri.ohsu.edu/redcap/surveys/?s=e1745a.

This study aims to determine which features of these deficiencies may be related to vision loss. We also hope to determine the frequency of certain changes in the DNA in genes that code for trifunctional protein (TFP) in the United States. The investigators of this study are Melanie Gillingham, PhD, RD and Cary O. Harding, MD.

This research study consists of two components. The first component is an online survey to be filled out by the subject or their parent/guardian. The second is a request to review medical records. Participation in either portion of this research study is voluntary. Medical record release is not required for participation in the survey portion of the study.

The online survey component of the study consists of questions about your medical history, and questions from the National Eye Institute Visual Function Questionnaire. There are a maximum of 88 questions, depending upon your answers. We expect that the survey will take no more than 1 hour of your time.

This study was approved by the Oregon Health & Science University. If you would like more background information about this study, please refer to the FODsupport.org newsletter published in January 2011 available online at http://www.fodsupport.org/documents/Jan2011issue.pdf. This issue of the newsletter contains a lay review entitled What causes retinopathy in long-chain 3-hydroxyacylCoA dehydrogenase deficiency?

If you have questions about this study at any time, please call Autumn Fletcher, study coordinator, at (503) 418-0109 or send an email to fletchea@ohsu.edu.

Another FOD Research Opportunity

A new study looking at the effects of 4 months of an odd-chain fatty acid supplement, Triheptanoin, to treat muscle pain and weakness, heart function, and energy problems of fatty acid oxidation disorders is being conducted at OHSU and the University of Pittsburgh. If you have CPT2, VLCAD or LCHAD or TFP deficiency and are 7 years old or older, you may be eligible to participate. Participants must come to OHSU or University of Pittsburgh and stay at the Clinical Research Center for 4 days on two different occasions. Participants may be asked to take medium-chain triglyceride (MCT) or triheptanoin (the study supplement). The chance of being asked to take the study supplement is 50%.

For more information, please contact Melanie Gillingham, PhD at (503) 494-1682 or email gillingm@ohsu.edu (eIRB 7140)

[More information on page 8]
SUBJECT RECRUITMENT for FOD RESEARCH

Lay Language Protocol Summary

Principal Investigator: Melanie Gillingham, PhD, RD
Study/Protocol Title: Triheptanoin Treatment of Long-Chain Fatty Acid Oxidation Disorders
IRB#: 7140

Please answer all of the following questions using lay language, similar to the language used in a consent form. Please number your responses.

1. **Purpose:** Humans eat long-chain fat in their diet and use it for energy during exercise and during periods of fasting. Patients with long-chain fatty acid oxidation disorders cannot use dietary fat for energy. They sometimes develop muscle breakdown, and severe pain with exercise or illness. They can also develop a heart that does not function properly. These patients are tired and expend less energy than people who do not have a long-chain fatty acid oxidation disorder. However, they can use a supplement oil called medium chain triglyceride or MCT. This study will determine if a new experimental oil called Triheptanoin can decrease the muscle pain and increase the heart function and the amount of energy in patients with long-chain fatty acid oxidation disorders.

2. **Recruitment:** Patients with a long-chain fatty acid oxidation disorder will be recruited through our clinic, past research participants, a patient support website, and recruitment letters mailed to physicians around the US. We will enroll 16 subjects at OHSU and 16 subjects at the University of Pittsburgh, age 7 to 40, with a disorder in fatty acid oxidation.

3. **Procedures:** Subjects will be admitted to the clinical research center for 4 days. They will collect all their urine for 24 hours. Heart function will be measured using ultrasound an electrocardiogram (ECG). For this test, the patient lies still on a bed and a probe is placed on their chest. The motion of the heart will be measured by magnetic resonance imaging/spectroscopy (MRI/MRS). For this test, the patient lies in the magnetic field of the MRI machine in the Advanced Imaging Resource Center (AIRC) for about 45 minutes. The amount of muscle and fat in the whole body and inside the liver and muscle will be measured by MRS and by dual X-ray absorptiometry (DEXA). Subjects will walk on a treadmill for about 45 minutes. The amount of Calories they use, their heart rate, and if they burn fat or carbohydrates will be measured. Blood samples will be collected before and after exercise. A meal test will be used to determine how much fat they burn. The subjects will drink a liquid breakfast with a stable isotope labeled fat in the breakfast. Breath and blood samples will be collected before and after the meal. The amount Calories burned by each subject will be measured when they are at rest on a bed by indirect calorimetry. The amount of Calories burned by subjects when they are doing their routine daily activities will be measured at home by doubly labeled water. All of these tests will be done at baseline. Then, subjects will be randomly assigned to consume MCT (current standard of care) or triheptanoin at 20% of their estimated Calorie needs for 4 months. The subject and/or the parent will be taught how to use the supplement oil in their diet for cooking and baking. The subject will be sent home and the oil will be shipped to their home. The study coordinator will call the subject or subject’s guardian each week to monitor the subject’s diet, potential side effects and assist with diet planning. At the end of 4 months, all of the baseline tests will be repeated.

4. **Survey Instruments:** Subjects will record what they eat for three days. They will write down what foods they eat, how they were prepared and how much they ate on a form provided to them. Subjects will complete the 3-day diet record 2 times during the study.

5. **Triheptanoin is experimental oil. It is a clear, odorless oil that can be mixed with foods and used in cooking. Almost all oils are made from even chains of carbon molecules. Triheptanoin is different because the carbon chains are odd in number. The co-investigator of this study at the University of Pittsburgh, Dr. Jerry Vockley, holds an IND for the prescription, and use of triheptanoin in humans (IND 106011).**

6. **Data Analysis:** The change in exercise ability, heart function, Calories used and body fat after 4 months will be compared between subjects randomized to MCT versus triheptanoin.
Update from Washington DC
[printed in Oct 2011 OAA Newsletter]
By Jana Monaco, IVA/OAA parent
[Thank you Jana for being a representative for many Families that live with a metabolic disorder, including our FOD Families!]

It has been 10 years since my husband and I came to realize the meaning of newborn screening and how important it really is—both to a child and a family. Our son Stephen was diagnosed with Isovaleric Acidemia at age three and a half, because this disorder was not routinely screened for at the time of his birth. If it had, his and our life would be extremely different. It would have been full of typical, wonderful and amazing milestones that we had hoped to experience with Stephen. Instead, it has been 10 years of continuous grieving of what we lost and continues to lose; learning and accepting the severe disabilities, seizures, surgeries and other complex health issues that are a direct result of the lack of expanded newborn screening for Stephen. Every year is another year of lost opportunities and memories taken away from us.

As we celebrate Stephen’s 14th birthday on October 26th, I reflect on the past 10 years and its whole new meaning is that one that has helped to give some sort of solace, however small, to our personal experience and loss. A multitude of initiatives have taken place allowing us to use our tragic experience to help change newborn screening.

The Secretary’s Advisory Committee for Heritable Disorders in Newborns and Children commenced seven years ago to address the disparity of newborn screening across the nation. Laws are now in place across the country in all 50 states expanding newborn screening to include all 29 disorders recommended by the American College of Medical Genetics and endorsed by the advisory committee. With the addition of SCID to the ACMG panel a year ago as recommended by the Secretary’s Advisory Committee, states are assessing their programs and gradually including it on their newborn screening panels. The most recent condition added to the recommended panel by the Secretary’s Advisory Committee, just last month, is Critical Congenital Heart Disease. Screening for this group of disorders with pulse oximetry will now challenge states to further assess their programs as they must consider adding it to their newborn screening panels. I am proud to have served a four year term with the committee and participated in the vote for each of these disorders. Though my four year term ended in January 2011, I continue as a member of the committee’s Education and Training Subcommittee. This subcommittee has been successful in influencing the decision of the American Academies of Pediatrics, Obstetrics and Gynecology and Family Practitioners to include education on newborn screening and inborn errors of metabolism for their physician training material and to find ways to incorporate it into their patient education material.

The advisory committee was tasked to identify a newborn screening “clearinghouse” by the Newborn Screening Saves Lives Act. It would serve as a “go to place or one-stop shop” for in-depth educational information for professionals and the general public regarding newborn screening. The Genetic Alliance was chosen to fulfill that goal and it successfully launched the site, “Baby’s First Test” last month. These are just a few recent accomplishments of the advisory committee in its efforts to fulfill its charter and mission for newborn screening. With the celebration of its 25th meeting last month, Dr. Rodney Howell, the chair of the advisory committee since its inception, completed his extended term of this committee. His passion and commitment to children’s health and newborn screening along with his years of knowledge and experience as a physician have guided him in leading this committee in a most reputable and distinguished manner in achieving its goals. Dr. Howell has been a prominent figure with the committee and will certainly be missed. At a recent dinner hosted by Dr. Howell in Washington to celebrate the 25th meeting of the committee, his time with the committee and its achievements, I had the honor of presenting Dr. Howell with a beautiful photo book full of kids with different organic acids, created by our own Raymonde DeGrace, and share a few words of our appreciation to him.

[Cont’d on pg 10]
Dr. Joe Bocchini, already a well respected and valued member of the committee, was selected as Dr. Howell’s successor. Following day one of the committee meeting, many were in attendance at Genetic Alliance’s 25th Anniversary Gala to celebrate 25 years of its hard work and achievements in the world of genetics. It was a privilege to be a part of its innovation theme this year and be included in its video collection of innovators viewable on YouTube.

Exciting and important work supporting the process and further progress in newborn screening continues and I am honored to represent the voice of the consumer and OAA in Washington. Most recently, I presented at the recent committee meeting on the consumer perspective and role with the committee and was able to raise the issue of reimbursement for medical formula and foods at the US Conference on Rare Diseases and Orphan Products in Washington, sponsored by NORD. The work never ceases, but progress is being made. Babies are being diagnosed at birth and receiving the necessary treatment for their disorder.

Children like my daughter Caroline, now nine, are living out their hopes and dreams despite their disorder, thanks to that early detection. Education and awareness of these disorders and newborn screening is greater than ever. More importantly, babies’ lives are being saved! As my family and I continue to cherish each day with Stephen through bittersweet eyes, we rejoice with his and Caroline’s life and the testimony to newborn screening that they represent. We celebrate each and every achievement to enhance newborn screening across the country.

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**Educational Advocacy Corner**

**Section 504 Helps Kids with Metabolic Disorders**

Beth-Ann Bloom, MS, CGC
Genetic Counselor

_Beth-Ann is the parent of a young adult with a disability and has enjoyed over 30 years of working with families dealing with metabolic and other genetic disorders [This was a posted document on facebook]_

With school starting again this is the perfect time to become familiar with how to help your child in school. Special education helps children whose metabolic disorders have caused serious complications, but many children who were diagnosed early and are doing well still need some extra help and protection in school. Federal law protects the rights of children with metabolic disorders. It guarantees reasonable accommodations and saves parents the need to go from teacher to teacher pleading for “favors” to keep their children healthy and in school.

Section 504 is a law that can help people with metabolic disorders. It protects them from discrimination. This US law gives people with disabilities the same chance as other people. The 504 law can help children in school.

Section 504 of the Rehabilitation Act of 1973 gives rights to people with disabilities. The law says that any group that gets money from the US government must be fair to people with metabolic disorders like OA and PKU. Public schools, cities, park programs, etc. must include people with disabilities.

504 says that public schools have to include all children. They have to make reasonable changes to make it possible for all children to join in and learn. These changes are called accommodations.

Children with health problems including metabolic disorders can get help from 504. Parents can ask for a 504 plan. It is a list of the accommodations the school will make. These changes will allow the child to be part of the school. These changes will make it easier for the child to learn. These changes are a civil right.


To get a 504 plan the child must qualify. The rules are different in every school district. To start write a letter to the principal asking for a 504 plan. There is a sample letter at the end of this article.

Ask your child’s doctor to write a letter. The letter needs to say why the child qualifies for a 504 plan. It needs to give the name of your child’s disability. It needs to list which part of the body is affected. It needs to say which life function is changed. It can suggest accommodations. There is a sample doctor’s letter at the end of this article to guide physicians who may not be familiar with 504 plans.

The law says your child can have a 504 even if he or she is doing well. If a special diet or treatment is working, the 504 can make sure it keeps working.

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[cont’d on pg 11]
School staff will tell you how they will decide if your child can have a 504 plan. You have rights. If they don’t agree to write a 504 plan, you can appeal.

A 504 plan is NOT special education. It gives help in regular classes. There are no special teachers or funding for 504.

Here are some of the kinds of accommodations children with metabolic disorders can get:

School staff will send a note home daily about what was eaten at lunch
Teachers will have training about the metabolic disorder
Child’s cafeteria lunch will meet special dietary requirements (Same cost as regular cafeteria meal)
Can keep water bottle at desk all the time
Can go to the bathroom at any time
Can have indoor recess when it is too cold
Grades not lowered if work is late when sick or at a clinic appointment.

All the teachers need a copy of the 504 plan. Other school staff like the nurse and the bus driver needs a copy of the plan.

Learn who is in charge of the plan. This person is often called the 504 Coordinator.

If the plan is not followed, tell the 504 Coordinator. If the problem continues, advocate for your child. Write to the coordinator. Ask for a meeting. Ask for help from a friend or another parent. Contact a parent center.

If your child needs more help, ask the teacher or the 504 coordinator. Review the plan at the start and end of every school year.

Children with metabolic disorders can do well in school. Section 504 helps them do their best.

Sample Letter Asking for 504 Plan

Date__________
Dear Principal_____________

My child, __________________________, Date of Birth ___________is/will be a student in grade_________ at __________________________ School. My child has a medical diagnosis of ____________________. This condition can have serious impact on major life functions including learning.

I am requesting that a 504 Accommodations Plan be written for my child. I will ask my child’s clinic to send a letter explaining the condition. Please contact me this week with information about the process used at the school for writing an implementing a 504 plan.

I can be reached in the following ways:
Home phone: ____________________
Cell phone ______________________
Email _________________________
Other: _________________________

I look forward to working with the school staff in writing this accommodation plan that will allow my child to be a fully included member of the school community.

Sincerely,

__

Sample Letter Asking for Doctor’s Help to Get 504 Plan

Date__________
Dear Dr_________________,

I need your help so that my child_________________Date of Birth_________ can be considered for a 504 accommodation plan at school. A doctor’s letter is usually the first step in getting the plan. It would be very helpful if your letter includes the following information. Can you please send a copy straight to the principal and another copy to me?

Thank you again for all you do to keep my child healthy. I appreciate your help in making sure things work well at school too.

Sincerely,
Dear Principal_________________,
I am the treating physician for Child’s name_________________ Date of Birth______________who is/will be in the ____________grade at _________________School. A 504 accommodation plan has been requested for this student.

Child’s name ____________ has a diagnosis of_______________________________. This disorder is characterized by (brief clinical description).

Name of disorder___________________ substantially limits this child’s learning and performance of major life activities. The major life activity (ies) most impacted by the disorder include (as many as relevant from list).

- Caring for one’s self,
- Performing manual tasks
- Eating
- Sleeping
- Walking
- Standing
- Seeing
- Lifting
- Hearing
- Bending
- Speaking
- Reading
- Breathing
- Concentrating
- Learning
- Thinking
- Working
- Communicating

Name of disorder___________________ significantly affects the following systems in this child’s body: include (as many as relevant from list).

- Neurological
- Musculoskeletal
- Special sense organs
- Respiratory, including speech organs
- Cardiovascular
- Digestive
- Genito-urinary
- Hemic and lymphatic
- Skin
- Endocrine
- Normal cell growth
- Immune system function

__
Child’s name’s _____________disorder has been mitigated by medical treatment. It is essential that this medical treatment continue during school. Although we try to provide as little disruption to the student’s day, sometimes it is unavoidable. The positive impact of the mitigation should not interfere with the student’s eligibility for 504 accommodations.

Please let me know if you need any additional information from my staff or me in order to find this student eligible for a 504 plan.

While I understand that the school will write the plan I would like to suggest that these accommodations be the minimum provided.

Your suggestions could include accommodations in these areas and others too:

- Access to health treatments during the day
- Dietary accommodations
- Staff training
- Access to water and the bathroom without permission
- All infection control practices needed to protect this student
- Special transportation
- No penalty for absences due to illness and medical care
- Accommodations for physical education and recess
- Peer education
- Inclusion in extra-curricular activities and field trips

Sincerely,
_____________________M.D.
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?’

One of our LCHAD dad’s, Scott, wanted to share some success experiences of his teen son, Doug - he's been wrestling for years and despite being challenged by his disorder, he strives for success in his sport and in life!

Here's an inspiring story that was recently written about Doug’s wrestling endeavors - way to go!

Shawna Cummings (MCADD), 8 yrs old, raised over $9000 for the Make-A-Wish Foundation at the 2nd Annual Shawna’s Walk For Wishes. This is the second year for the walk and we hope that next year we can raise double that amount. This year Shawna also had another big accomplishment— Shawna along with her sister, Seirra, and brother, Zachary (PDHD), received their yellow belts in Tae Kwon Do.

Melissa (NY)- Mito(PDHD) and MOM to Zachary- 6 yr- Mito (PDHD)
Shawna- 8 yr- MCADD
Seirra - 10 yr- unaffected

This is Juan.
He is wearing a River Plates tshirt, the football team which he loves, although as a football club this was not its year, definitely. Juan finished his secondary school this November and he already begins his music studies in 2 different universities. In one of them he will study “Musical Composition” and in the other one “Musical Arts.” He wants to become a classical music composer, so he has to keep on studying with a Professor and

continue his German lessons, because he has plans to go to Austria or to Germany.

As you can see, he is well. Hoping you are all healthy too. We wish a better 2012 for all the world!

A KISS from:
MARÍA MARTHA,
Mother to Juan, 17 years and a half, VLCAD, Buenos Aires, Argentina
Love Messages

Lisa Poon and Kevin Tsoi

Natasha...
Oct 1, 2009 ~ December 19, 2011

Born in Hong Kong, Natasha had a metabolic crisis when she was about 22 hours old. She was diagnosed with CACT shortly after through blood tests.

Her father is Kevin Tsoi and she has an older brother, Matthew, who is a carrier.

Thank you for this FOD Family Support Group and please let us know if there is information we could share to help raise awareness about this particular type of FOD.

With love and blessings,
Lisa Poon

Condolences...

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

‘Healing is not forcing the light to shine but removing the obstacle that blocks it’
~ Tyler Woods

It is with great sadness that am forwarding the announcement of the untimely death of a good friend and colleague, Dr. Paul Fernhoff.

Dr. Fernhoff was an active advocate for newborn screening and contributed much to the Georgia newborn screening program and to national and international newborn screening and genetics activities. He was the principal investigator of a multi-year contract between Emory University and the State of Georgia to follow up for all infants with abnormal metabolic screens, confirm their diagnosis and initiate appropriate short and long-term management. Dr. Fernhoff was the medical director of the Emory Lysosomal Storage Disease Center, and the principal investigator on over 20 studies funded by commercial pharmaceuticals to examine the effectiveness of enzyme replacement therapy and other enzyme enhancement therapies for children and adults affected with lysosomal storage disorders. He was also the Medical Director of the Atlanta Jewish Genetic Gene Screen program and the Pediatric Program of Hospice Atlanta, one of the largest children’s hospice programs in the country. Dr. Fernhoff was active in local and national activities assessing the ethical implications of introducing genetic technologies, including newborn screening, into public health programs, and he was a visiting scientist at the Center on Birth Defects and Developmental Disabilities at the CDC, working closely with colleagues to study the origins of birth defects and developmental disabilities and assessing the long-term outcome of genetic screening programs.

He will be sorely missed!
Dr. Brad Therrell

Funeral Guestbook

Dr. Paul M. Fernhoff, age 65, Atlanta died September 19, 2011. Dr. Fernhoff was the Associate Professor of Human Genetics and Pediatrics and the Medical Director of Human Genetics at Emory University Hospital. Survivors include his wife, Dr. Deborah Finkelstein Fernhoff; daughter and son-in-law, Drs. Shana and David Cohen, Atlanta, GA; son, Dr. Nathaniel Fernhoff, San Francisco, CA; granddaughter, Talia Cohen. A graveside service will be held on Wednesday, September 21, 2011 at 10:30 am at Crest Lawn Memorial Gardens with Rabbis Adam Starr and Ilan Feldman officiating. Arrangements by Dressler’s Jewish Funeral Care, Atlanta.
We are still seeking a Family Volunteer to sit at our booth in New Orleans, LA for the Genetic Metabolic Dietitians International (GMDI) Conference on April 19-21, 2012

Conference Info

We are sharing a booth with the OAA (Organic Acidemia Association) - I'll be sending a banner and brochures etc to share with the professionals to create awareness of our Group and its free services.

If anyone can volunteer for 1 day or 2 days please let me know. If no one is available, I will let the OAA have our seat and they will have another parent use our ticket. The ticket covers meals while there, but it does not cover attending the sessions—that's another separate fee.

Please let me know in the next several weeks if anyone would like to volunteer! Email Deb

~ NEEDED FOR THE JULY 2012 ISSUE ~

Medical Update ~ Please

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

FAMILY STORIES & Pictures for KidsKorner

The ‘Silver Linings’ of FODs ~
What is your ‘Silver Lining’?

URGENT NEED for Medical Professionals

With more Families being identified with an inborn error of metabolism (through expanded newborn screening), our Families will need ongoing Clinical Care from knowledgeable and caring professionals. In addition to our Newborn Screening Advocacy by many of our Families, our Group is hoping to also bring awareness to medical schools and other medical organizations and facilities the need for educating and training new Professionals (physicians, metabolic nutritionists etc) in the field of Medical Genetics and Metabolism to treat our children, as well as our FOD adults. We are also raising funds for Clinical Training. [see our website for the donation box]

Once we raise enough Funds we will be able to offer grants to US Clinical Training institutions. We NEED your help NOW and in the FUTURE so our children will thrive and grow into adulthood with the best of ongoing care!
**‘Heart Healthy’ Recipes**

### Emergency Food Kit for Car

Making an emergency food kit for the car has been on my to-do list for the last three years...but has always gotten put down low after all the things that need to get done (you know how it goes). Well, yesterday Christopher and I went hiking and got to the woods and realized that I left his snack with his MCT oil and cornstarch at home. I panicked. It was one of those situations that it took us so long to get there (because we spent half the morning in time-outs) and I knew if I tried to get him back in the car and travel 15 mins to the store etc. we might as well go home. I opted for buying gatorade at the state park store and used all the snacks I had packed for him...and tried to do more mellow activities on our hike (ie., throwing rocks, drawing in the sand). I wonder if I made the best decision by pushing the situation onward...he seemed fine...but I vowed after all the anxiety that I had over everything I was going to make emergency food packs to keep in the car. After making the packs I have realized how much less stress I have on a day to day basis worrying, "Did I pack enough extra food?" And I don't have to limit our schedule quite as much because I didn't pack enough snacks...it feels very freeing...so I wanted to share with everyone in case there are others who haven't done this yet.

**Here is what is in my "kit" or box in the car:**

1. A bottle of coconut water (we recently have been advised to combine this with cornstarch before activity—coconut is not for MCAD)
2. Small bag of cornstarch
3. Fruit rolls
4. Lunch size applesauce
5. Pretzels
6. 2 Cliff-kid bars (obviously a little higher in fat than I'd like but at least substantial)
7. 3 small boxes of raisins
8. When I order my next batch of PB2, I am going to get small packets of this to throw in with crackers
9. A long awaited tiny first aid kit (Did you know tampons are great for nose bleeds...had to throw that in there!)

Well, thanks to whomever originally thought of the idea...and I hope that this can be helpful to others as well. We have enough on our plates to stress about! Who needs to have these moments as well!

Love ya'll
Stephanie srharry374@hotmail.com
Christopher 3 LCHAD

### Yummy Chocolate Banana Cake

For my daughter Viola’s first birthday, we made a frozen layer cake with meringue rounds and blueberry/raspberry sorbet ice cream in between. We topped with something similar to Marshmallow Fluff and sprinkled with candied violets (to make it Viola’s cake :-). For her second birthday, we made an oatmeal cookie style layered cake - in essence, just VERY big oatmeal cookies to stack - and the filling was loads of finely cut fresh fruit and low fat greek yoghurt whisked with honey and vanilla essence. We topped with beautifully arranged strawberry slices. I hope this can provide some inspiration!

All my best
Fie Lundsgaard Olsen, Copenhagen, Denmark fielundsgaardolsen@gmail.com
mom to Viola, 2 years VLCADD mom to Viola, 2 years VLCADD

### MCT Butter

1 tbsp corn starch
3/4 cup skim milk powder
1 tbsp lemon juice
3/4 cup water
1 cup MCT oil
Few drops yellow food colouring

Sift dry ingredients. Slowly pour in lemon juice water and oil. Mix well. Cook over medium heat, whisking gently until mixture thickens. Add yellow food colouring for colour if desired. If mixture is not smooth, mix in blender until smooth. Pour into glass jars or containers. Freezes well, but needs to be reblended when thawed out. Keeps in fridge for about two weeks.

Michal Peters m2010peters@gmail.com

### White Mountain Frosting (From Betty Crocker Cookbook)

Makes 24 cupcakes (Bake 20-23 min.) (1 cupcake= 2 t. MCT oil)
Makes 66 gmsize cupcakes (Bake 15 min.)

For no fat frosting: Use Fat free frozen whipped topping, sweetened non-fat sour cream, or White Mountain Frosting.

White Mountain Frosting (From Betty Crocker Cookbook)

1/2 cup sugar
1/4 cup light corn syrup
2 tablespoons water
2 egg whites (1/4 cup)
1 teaspoon vanilla

Combine sugar, corn syrup and water in small saucepan. Cover; heat to rolling boil over medium heat. Remove cover and boil rapidly, without stirring, to 242 F. on candy thermometer (or until small amount of mixture dropped into very cold water forms a firm ball).

As mixture boils, beat egg whites until stiff peaks form. Pour hot syrup very slowly in a thin stream into the beaten egg whites, beating constantly on medium speed. Beat on high speed until stiff peaks form; add vanilla during last minute of beating.

Fills and frosts two 8- or 9-inch layers or frosts a 13x9-inch cake. After frosting, add candy sprinkles to make it colorful.

Diane
Stephen, age 8, LCHAD/TFP
3 girls, ages 12, 15, 17, unaffected
dnielsen6@gmail.com

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**Emergency Food Kit for Car**

- A bottle of coconut water (recently advised for MCAD)
- Small bag of cornstarch
- Fruit rolls
- Lunch size applesauce
- Pretzels
- 2 Cliff-kid bars
- 3 small boxes of raisins
- A first aid kit

**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 tsp cinnamon
- 1/4 cup liquid egg substitute or 4 egg whites
- 1/4 cup Safflower oil (or MCT oil...VLCAD or LCHAD only)
- 1/4 cup water
- 1 tsp vanilla
- 1 cup plain nonfat yogurt or buttermilk
- 1 banana, mashed

**White Mountain Frosting**

- 1/2 cup sugar
- 1/4 cup light corn syrup
- 2 tablespoons water
- 2 egg whites
- 1 teaspoon vanilla

*(As written in Betty Crocker Cookbook)*
We had a meeting where a neuropsychologist spoke about ADD issues. He recommended the book, *Smart but Scattered*. I've started reading it and so far I think it'll be a great help. I also had our Dr set up an appointment with a pediatric neuropsych to get a base evaluation.

It was suggested to have the evaluation done just in case they ever had an early crisis.

It's also to check their progress and development.

Check it out, my local library had a copy.

Stacy, MN
Hazel & Pearl, MCAD  styew@yahoo.com

I highly recommend that you read the Ferber Book *Solving Your Child's Sleep Problems* but make sure it's the latest edition. He's revised it to include elements of co-sleeping and is less harsh than his original book. The book really helped me understand what the current sleep theories are, and what are our sleep associations, and it gave me a better understanding of sleep issues we may deal with. You can also read Elizabeth Pantley's *The No Cry Sleep Solution*.

Natalia Olivera  archnatalia@gmail.com

Articles on Autism by Dr Robert Naseef, a dad and psychologist


Sensory Integration Disorder: A great book is *The Out of Sync Child* by Carol Stock Kranowitz
Sheri Sandstrom  sheri@epix.net

I think this is a great article about diet and supplements not only for kids with autism but for those of us with metabolic disorders as well.

http://nourishinghope.com/2011/09/one-of-the-most-comprehensive-studies-on-nutrition-for-autism/
Gwen
mom to Austen age 15 MADD
Boston
gwenabele@hotmail.com
We sent Gwen (MCAD) off to college about 3 ½ hours from home. Yes, I worry, BUT...she’s been through 2 years now, and so far, so good. We DID go and speak directly with the campus medical director, and supplied them with protocol letters and contact info.

Gwen wears a medical ID, so if there’s a problem, the emergency responders will know she has MCAD. She also made an effort to talk with her roommate(s) and friends about it, so they would know what to do if she got sick.

Some things to consider:

1. Do the freshman dorms have A/C? Some on Gwen’s campus did not, and we had to provide a letter of medical necessity for her to live in one that did.
2. Does the cafeteria/meal plan allow for healthy meal alternatives (at EVERY meal)?
3. What is your emergency plan if there is a problem? Will you or a family member be able to get there ASAP? You may also want to check out the local hospitals/specialists.
4. Is off-campus living an option for freshman? We found that Gwen eats much better when she’s doing her own meal prep (and the bonus is that it’s cheaper as well!)
5. What kind of emergency response does the campus have? Is there a campus EMT service? Or is it city? And what is the response time?
6. Size of the campus. Students will do a LOT of walking...and when it’s hot (classes start mid-august), are there places for them to get in the shade/drink water/have a snack/cool off between buildings or parts of campus?
7. Is there a campus shuttle bus? This can be especially important on days when your student may not feel quite up to all the walking.
8. The maturity level of your student – are they willing (and able) to make decisions about their meals/activities/friends/living style that will be healthy for them (even when those choices may be hard or unpopular)?

As for cutting class...students choose their own class schedules...what that means is that if they have trouble getting going in the morning, they can schedule their classes so that the first one is at 10 or 11 or 2. Most classes allow 1 or 2 absences before the student’s grade is affected, and many of the professors are willing to work something out if the student has legitimate medical issues that cause them to miss more class than is allowed.

Hope this helps. Choosing a school is an exciting thing for students...enjoy it!

Hope
Mom to Gwen (20 yrs) MCAD  hopek@considertheheavens.com

•   •   •

We home school, which makes food choices and timing much easier. It is really the only option for us, given the constant fatigue and need to eat. But many of our snack items are "packable," especially if your school is flexible with snack times/opportunities. My daughter is gluten intolerant and simply dislikes the taste, texture or something about many other foods. While she does not have an FOD, she doesn't do well with much fat, and sometimes when she is sick she goes fat free (you can see why they thought it was an FOD). We try to rotate foods to keep them interesting, keeping corn chips, Pirate Booty (or similar low fat puffed corn/rice/cheese product), brown rice crackers, apple slices, applesauce pouches, meal replacement drinks (she likes Orgain), rice cakes, and rice in our carb arsenal. For protein, we rely heavily on 1% milk, and some lean meats. We have found that combining some fat (if allowed), protein and complex carbs yields the longest lasting energy and blood sugar maintenance.

Rosie
Near St. Louis
mom to Sarah, 13, carn. def. + ???mito
and 4 similar kids
jimrosiesarah@gmail.com
Q: My son has been doing pretty well at drinking but it seems like more times than not, he still has that fishy smell. They already reduced his carnitine dose, so I thought it is odd that he is continuing to have this issue. Any other suggestions?

A: The fishy smell is trimethylamine (TMA), which is a product of carnitine. It’s made in the gut. Roughly 15% of a carnitine dose is likely to be absorbed; the gut flora get the rest. Some of them will make the TMA, which can be further metabolized in the bowel, or absorbed and metabolized by us to TMA oxide, which is odorless. There are other sources of TMA, including choline, which is present in fish and other seafood. So if a persons’ ability to deal with TMA is exceeded, the first thing you might notice is a fishy smell to the diapers. If the liver’s ability to cope is exceeded, the TMA will come out everywhere — all the pores, the urine, the lungs/breath, etc. The odor will be worse if the person is wearing clothes which trap the TMA —caps in winter, for example, instead of allowing it to dissipate as quickly as possible.

Changes in diet are of course a big factor; likewise changes in carnitine intake. Alterations in gut flora are another big factor. We often hear about transient TMA events in some of our carnitine patients, associated with viral GI upsets. Some broad-spectrum antibiotics will suppress some of the microbes that make TMA; nowadays I would be inclined to try a probiotic preparation first, to see if that would alter to gut flora favorably. (The probiotics are typically aerobic organisms-they use oxygen. The microbes that make TMA and other very smelly substances in stool are typically anaerobic-they flourish only where there is little oxygen.) Lowering the carnitine dose is usually only needed temporarily for someone who has had no problems most of the time.

Ages ago, when I was at Duke, we developed a simple underarm deodorant that would trap the TMA on site, instead of allowing it to waft away from that location. When the user took a shower at the end of the day all the TMA would be washed off, so the fishy odor was quite strong in the shower, but not so much during the day.

The enzyme trimethylamine oxidase, which makes TMA oxide in the liver, uses a riboflavin cofactor. I wonder if a mild shortage of riboflavin might account for some of the occasional TMA problems we encounter. There is a genetic deficiency of TMA oxidase as well. It is quite variable—some people have intermittent troubles, others have a major problem all the time, even when they avoid all obvious dietary sources of choline. This genetic form of TMAuria is quite rare. A similar disorder, even rarer, is dimethylaminuria (DMAuria). It will produce the same odor, from the same dietary sources.

I think two labs in the US measure TMA (in urine) routinely-ours, and Dr. Van Hove’s in Denver. DMA would also show up on the same test. It’s not necessary to measure it, of course, in someone who is on carnitine-we know what is causing the fishy smell.

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Mail: Division of Clinical Genetics
Slot 512-22
Arkansas Children’s Hospital
One Children’s Way
Little Rock, Arkansas 72202-3591

Pharmaceutical Update

The liquid and tablet generic drug brand for Levocarnitine was approved for distribution by the FDA several years ago. Please note that this generic drug form by Rising Pharmaceuticals, Inc. (as well as Sigma-Tau Pharmaceutical’s brand name Carnitor®) needs a Prescription from the Doctor. Hi-Tech Pharmacal, Inc also makes a generic drug oral solution and tablet form of levocarnitine.

Please also note that the generic DRUG form of L-carnitine is NOT the same as the over-the-counter carnitine supplements often bought at healthfood stores — those products are NOT regulated or approved by the FDA to be used for metabolic disorders (read the article on our Pharmaceutical page for further information). The term 'generic form of a drug' should NOT be used interchangeably with the term 'over-the-counter supplement.'
Kids Korner

Maggie
Almost 3 yrs old
VLCAD
Kentucky

Brody
1 yr old
VLCAD
Virginia

Slayer
9 mos old
MCAD
Oklahoma

Drake
14 mos old
MCAD
Utah

Kinley
19 mos old
MCAD
Baton Rouge, Louisiana

Landon
Non-cardiac
VLCAD
Arizona

Connor
2 yrs old
VLCAD
Oklahoma

Please note that we also have an FOD KidsKorner/Adults Gallery and other Pictures on our homepage. To submit a pic please email Deb.
**DONATIONS**
[since our July 2011 Newsletter]


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 www.fodsupport.org./Donate.htm


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 501c3 Tax ID # is 83-0471342.

**Reminders**

**Families - Please send TYPED** (preferably in word document) stories etc, by **July 15, 2012 to Deb.** 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, summaries, etc by **July 15, 2012 to Deb.**

‘Walking with a friend in the dark is better than walking alone in the light’

~ Helen Keller

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**Communicate With Us**

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

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