

fatty oxidation disorder communication network

FOD Support



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From the Editor



It's hard to believe another year has gone by so quickly ~ I hope all of you had a blessed and healthy holiday season and HAPPY NEW YEAR!

We are quickly moving into high gear with Kathy Stagni (OAA Director, www.oaanews.org) to plan for our National Metabolic Conference hosted by Dr Charles Roe and the Institute of Metabolic Disease in Dallas, TX ~ the dates will be Friday, June 23 and Saturday, June 24, 2006. I will be mailing Registration Forms to US and Canadian FOD Families and Professionals in February or March, but we will also have it on the website sometime this month. We are waiting to get confirmation on some of our Speakers. The FOD site already has some Hotel information on the homepage, but more will be posted in the next few months. In addition to receiving the Registration Form in the mail, I will be sending an updated Family ad Professional List ~ it's been a few years since we've done that (due to low funds).

We are still trying to line up our speakers/topics, but we do have a few confirmed ~ all speakers and topics should be set by February or March. One way to stay updated on Conference news, as well as for networking with other Families, is to sign up for our Email List on our site. 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.

Our Awareness Bracelets (advocating Expanded Newborn Screening) have sold well on the website ~ I will be bringing more to the Conference so both FOD and OAA members can purchase one (or more!) in June! Additionally, with your Conference Registration Fee (\$50.00 per person) you will receive a special yellow and blue FOD t-shirt ~ it would look even better while wearing the FOD/OAA

bracelet! Further information (on sizes etc) will be on the Registration Form. If you'd like to purchase the bracelets from the site (right sidebar link) ~ Please read the entire page before you order ~ you can pay using a credit card via the FOD PayPal link or you can write a check out to FOD (not tax-deductible) or to OAA (tax-deductible ~ we are doing this project with the Organic Acidemia Association).

Please note that at times (usually 5-7 days a month) I am away from Greensboro ~ Dan works at Michigan State, so we are commuting until next August when Brian graduates from high school and starts up at MSU as a freshman. If I don't get back with you right away, that's why ~ and I will email/call when I return to NC. In any case, if it's very important you can contact me in MI at 517-381-1940.

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.

We also have some Professional Medical and Nutritional information in this issue ~ Thank you to all our contributors. Some of those articles will be posted on our website ~ we always welcome new Medical articles (in pdf form).

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...

Take care... DLG



'We Are All in This Together!'



Family Story

Andrew's Story ~ LCHAD, Canada

Our son Andrew Ouellette was born on March 5, 2001 six weeks premature. As most, we were two extremely happy and proud parents. Little did we know, our little Andrew was suffering from a very serious, but silent disease.

In his first few months, Andrew experienced below average and very little weight gain. We thought we were the luckiest parents in the world to have such a good baby. We literally had to wake him for most of his feeds. Andrew was such a sleepy baby. He had some developmental delay when we realized he could not hold his head up straight when he was three months old. At this point, he was vomiting at least five times daily, especially after his formula. This was not just a baby spitting up, the vomit covered him from head to toe. Due to Andrew being born prematurely, we were already seeing our family doctor every six weeks. My husband and I would continue to bring up these symptoms at every visit until he was four months old and voice them as concerns. These symptoms were routinely excused due to his premature birth and that a lot of babies before the age of one were "pukey" babies. We took the doctor's advice and changed Andrew's formula just in case he was intolerant to milk based formulas.

During Andrew's fourth month of life, we noticed he was slowly getting weaker. He was sleeping a lot and his vomiting was progressing. The only thing he could keep down was his morning and nightly feeding of Pablum mixed with water. Andrew eventually got to the point where he was vomiting after every feed. Then we ended up feeding him with a small syringe because he was too weak to suck on a bottle. We had a referral to see a pediatrician because his family doctor had noticed his low muscle tone. His pediatrician had told us that he believed that Andrew was categorized as failure to thrive. Andrew was to go to the Hospital for Sick Children for testing for cystic fibrosis. This appointment, which included an ultrasound, a sweat test and a blood test was a mere two weeks away. These would be the longest two weeks of our little family's lives. Andrew was slowly slipping away from us and nobody was doing anything to stop him.

Just days before the scheduled tests, my husband and I felt there was no choice but to bring Andrew to our local hospital because he wasn't eating anything. We knew he was becoming dehydrated and that he needed medical attention from a pediatrician who had probably seen children in the same state Andrew was in. We were confident they would be able to fix our son. Sadly, this was not the case. They simply admitted him, hooked him up with an IV (which in fact was the only thing that saved him) and left him in a private room for three days because they didn't have a clue what was wrong with our beautiful baby. They had noticed through X-rays that his liver was enlarged, but offered no explanation as to why. We were told that Andrew had a virus and that it would have to work its way through our baby's tiny, cold, limp body.

Finally, the day had come to go to Sick Kids for the testing previously scheduled. Andrew was not given any feeds through the night because he had to fast for his ultrasound the next morning. I thought we would be brought down by ambulance because Andrew was not getting better and he was still hooked up to an IV. Instead, I was told that my husband and I could take Andrew out on a day pass, travel in our own vehicle, and they would leave the catheter from the IV in his bruised little hand so he could be hooked back up when we returned to the hospital.

We arrived at the Hospital for Sick Children in Toronto and registered Andrew for his scheduled tests. He slept through most of the testing, which took up most of the morning and never complained except for when he had to be undressed for the ultrasound. When it came time to have his blood drawn, Andrew didn't even flinch when the sharp needle went into his tiny wrist. This immediately set off a red flag to the nurse who was drawing the blood so she told us that she was going to take us over to Emergency, just to make sure Andrew was okay. Within ten minutes, Andrew was intubated and barely clinging to life. Blood work confirmed hypoglycemia. The next hours were a total blur. We spent every moment with our son in the ICU. We took turns sleeping, sometimes for only minutes at a time.

Andrew needed a line put in to his main artery in his groin so that the doctors could analyze his blood work every few hours. Andrew still has scars to this day. This was the first of many painful procedures Andrew had to



endure. He then had a spinal tap to rule out meningitis. My husband and I started to fear the worst. At the same time, the anger and frustration was building up in both of us. Why didn't anybody know what was wrong with our little boy?

Finally, an answer came one week after our terrifying ordeal in the ER. Andrew had LCHAD deficiency. A genetic metabolic disorder that does not allow your body to break down long-chain fats to use as a main energy source. It is an extremely serious disease with very serious consequences, but is very much manageable by a proper diet. We must feed Andrew other sources of energy such as foods with high carbohydrate content and a high calorie content, however, very low in fat. We must also ensure that Andrew does not fast for any period of time greater than six hours. This means that he must always have usable sources of energy provided for him, even during the night.

All in all, Andrew spent six weeks at Sick Kids and slowly turned around for the better as the treatment progressed. He remained on a low flow of oxygen due to his low muscle tone for a period of two months after we were discharged. Also, due to the fact that he could not fast, Andrew required a G-tube to help sustain him with supplemental feedings. He was still too weak to suck sufficiently out of a bottle. He was in excruciating pain after this minor surgery. For the next two years, he wouldn't let anyone touch the left side of his body because he was that sensitive. To make matters worse, my husband and I have to change his tube every four months. It pained us to see our son in so much agony. However, we had to come to terms with this new lifestyle.

Andrew has suffered significant developmental delay due to his trauma and has had to overcome many obstacles most healthy children do not. He has had to endure painful therapy to become the active little boy he is today.

Today Andrew is doing very well, considering everything he has been through and we are extremely thankful for that. Our son is unlucky for what he has to live with. However, he is very lucky this disease did not kill him. Lucky we were at Sick Kids that day and lucky the metabolic team diagnosed him as promptly as they could.

This is why we are here today. A baby's life should not be left up to luck. Your baby's, your grandchild's or nephew's life does not need to be left up to luck. With expanded newborn screening in Ontario, our children can be tested for LCHAD and 28 other metabolic diseases. These silent killers can be found before another baby suffers severe trauma, mental retardation, or worse, death.

We learn from our past and our experiences make us wiser. I am asking all of the MPP's in the Legislature to learn from our past experiences. Expand Ontario's newborn screening program to include conditions such as LCHAD.

Allan and Carrie Ouellette
Aurora, Ontario
May 29, 2005
Email: acouellette@sympatico.ca

[Please note from DLG: Beginning in 2006, the Ontario government will test for 21 disorders. Several other provinces are still lagging behind. However, Saskatchewan is the only province that screens for 29 disorders.]

Family Story

Isabelle's Update ~ Unspecified Disorder

Many things have changed since I first told our story (see FOD website, Family Stories). Isabelle is now 7 and she has 2 siblings ~ Rebekah is 4 and Jakob is 2. Both Rebekah and Jakob have the same unspecified disorder that Isabelle has although we have been given the label Mitochondrial Myopathy to use since in many cases labels help.

We have moved to Fort Worth, Texas to be closer to better medical care than we were able to get in Nebraska. In January of 2003 we chose to have a g-button placed in both Rebekah and Isabelle and it changed their lives. We no longer struggled with the nighttime hypoglycemia as we had in the past. Since they were able to get proper nutrition they began to grow like any healthy child. But most importantly illness now doesn't always require the hospital and IV as it had in the past. However, in August of 2003 when our baby, Jakob, was 7-months we realized that he was heading down the same path as his oldest sister with delayed development and slow weight gain. So we decided to get him a g-button as it had done so much for Rebekah and Isabelle. Unfortunately the doctors and insurance companies did not agree. While we were fighting with them I was also charged with making my children ill to get money and attention. The charges were later labeled 'unable to determine.'

During this time we watched as our beautiful baby boy was wasting away. This was probably one of the lowest points of my life. Finally, with the help of some great doctors, Jakob received a g-button at the age of 14-months. Today Jakob looks and acts like any healthy 2-year-old. Of course all three children have to be on nighttime feedings through the g-button to control the hypoglycemia and we have had a few close calls when tubes came undone. But because we knew what to watch for we were able to catch the lows before any damage was done.

On August 10, 2005 Isabelle underwent a Nissen fundoplication. We had been putting it off since she was a baby but no longer could. During the surgery doctors discovered that her muscle was so weak that it had stretched out to be twice as large as it should be. She is doing well now and hasn't thrown-up once since the surgery. I can't say that our lives are perfect by any means. When the kids get sick, they get really sick. And, of course, we still have to use special pediatric strollers for going anywhere of distance. But thanks to amazing parents like those you read about on the FOD site (that blazed the way for all of us to follow), and some wonderful doctors who don't think that every word out of your mouth is just paranoia, my kids are living what I consider a blessed life.





Family Story

Rayce's Story ~ MCAD



I wanted to share with all of you the story of our precious bundle of joy. Our son, Rayce, was born on Tuesday, November 16, 2004. We were so happy that our beautiful baby boy was born. Rayce was perfect. Family and friends came and visited while we were in the hospital.

I decided that I would try and breast-feed our son. At the beginning, it seemed like everything was going fine. The next day I told the nurses that I wasn't sure if he was getting what he needed from me and maybe we should supplement with formula. They said that they thought everything was okay. On Thursday he was just lying there and hardly moving much. He was looking a little on the pale side and hadn't eaten much. The nurse came in and I told her what I was seeing and she took him down to the nursery and took his temp. Rayce's pediatrician came in and said that his temp was low and so was his blood sugar. They put him under a warming lamp and gave him some sugar water. About 45 minutes later the nurse came back in with Rayce and told me to keep him wrapped in the blankets to keep his body temp regulated; and that the sugar water helped with his blood sugar. She said it was probably low because he hadn't eaten for awhile. I really didn't give it much more thought. It was after this incident that I felt he wasn't getting enough to eat so I decided to switch to straight formula. He was taking about 2 oz every 2 hours.

Thursday night we were finally discharged around 7:00 pm. Our first night home went fine. Friday morning we got up and started the day. Around 10:00am the telephone rang ~ it was Rayce's pediatrician. She informed me that Rayce had a blood test come back abnormal and that we would need to be seen at the St. Louis Children's Hospital and that the metabolic dietician would be contacting us to set up an appointment. About 45 minutes after I hung up the phone with the Dr, she called. Of course I was all upset because I didn't know much about this. She informed me that everything would be fine because we caught it early and that we would be able to treat it. I asked her what it was he had and she told me that Rayce has a rare metabolic disorder called

MCAD. She said that this was something we could treat with medicine and diet. She wanted me to make sure he ate every 2-4 hours and no longer than 4 hours. We didn't have a problem with this because he was already eating every 2 hours.

On November 24th we drove to St. Louis Children's Hospital. We first spoke with the metabolic dietician and she explained about the feedings, how to take blood sugar when Rayce was sick, etc. We then saw Dr Dorothy Grange. She checked Rayce over and said everything looked great and to make sure he eats every 2-4 hours. She prescribed L-Carnitine for Rayce, 1.5 ml. twice a day. We made another appointment to see her again in March.

We have returned to St. Louis Children's Hospital several times, but only for our check-ups. They have increased his carnitine since he is a growing and healthy baby. He now takes 3ml twice a day. Rayce usually goes to bed around 9pm after eating a snack and usually wakes up around 2/2:30am and takes a bottle. Rayce has had 3 ear infections, but luckily we have not had any problems with him not wanting to eat. He loves his food too much. We are very thankful that Rayce was diagnosed through the expanded Newborn Screening. We found out only 3 days after birth. I believe this saved us from having another serious crisis.

We have already celebrated Rayce's 1st birthday party ~ WOW how time flies. Rayce now eats anything we give him. He is only taking a bottle in the middle of the night usually around 2/2:30am. He drinks from his sippy throughout the day. And we just make sure that he has something every 2 hours to eat.

St. Louis Children's Hospital gave me this web site. I got on and just couldn't believe some of the stories. I sat and cried and cried. Deb then made a personal phone call to me and sent me a packet of the past information on MCAD to help me understand better. I just want to say, Deb you are a very dedicated person and I want to Thank You for that. This web site has helped me so much. Any time I have a question everyone is right there to help me with it (via our FOD Email List). Like the bracelet says 'All In This Together.' And that's what we all need.

My heart goes out to those who have lost loved ones from an FOD.

Julie Giddens
Illinois

Family Story

Anna's Story ~ Unclassified FOD

Anna was born not breathing and had swallowed lots of meconium. She ended up being suctioned and then on oxygen. Her first apgar score was 3. After 10 minutes of workup she was better, with an apgar of 8. She was monitored in the nursery for an hour, with my husband alongside her. I finally got to hold her and the nurses told me they were stunned by how long and thick her eyelashes were and how alert she was. She was holding her head up on her own by the time we left the hospital. She was so alert and strong.

Two weeks later, she started losing weight and throwing up everything she took in. The pediatrician wanted us to be in his office daily to monitor her. At 4 weeks, we saw a GI specialist and Anna started Tagamet medication and Nutramigen formula. She still threw up a lot of what she took in but slowly gained weight so, while it wasn't ideal, we were all relieved for the short term. Since there were problems with aspirating in her sleep, Anna slept in our room for the first 6 months. I kept her in a bassinet beside me, propped up on her side and swaddled (it was the only way she would sleep). Every morning I changed her bedding, it was always soaked with formula. Up until then, I had always had a sense to check and knew if something was wrong. I know we were lucky that she didn't choke to death in her sleep. We had a good sleeper ~ Anna slept through the night starting at 4 weeks. This meant the only way I knew she was okay for sure was to hear her breathing. I didn't trust the monitor. Anna didn't gain weight as fast as we hoped, nor did she gain height and head circumference as well. We saw a neurologist at 8 months and again at 18 months for her physical development delay. Anna wasn't walking yet either.

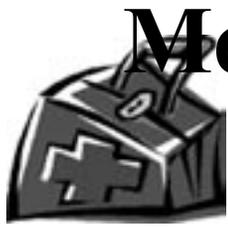


She didn't sit up until 8 months. She crawled, pulled to a stand and cruised all at 13 months. She did everything in spurts and a little late. We got a diagnosis of hypotonia at the neurologist appointment at 18 months and started physical therapy in October. She walks very well now, but is still behind. Anna's weight lagged so I started giving her more concentrated Nutramigen in her bottles until I was pouring pure concentrate into them to pack calories. She would only take 1-2 ounces at a feeding and wanted to feed every 30-40 minutes. This was in between throwing up so feeding took up my days in her first year. She never liked eating much but I could manage to get her to eat regular cereal throughout the day. I had to keep offering to keep her eating. She still eats like a bird but has her foods that I know she will eat well. Now that she is walking her appetite is better, but nowhere near where I'd like it to be. She takes PediaSure in her sippy cups (they go to nap and bedtime full with her), along with Carnation Instant Breakfast powder to make a whopping 45 calories per ounce. This has helped maintain her weight growth. She now throws up less, about once a day, but it's messy and projectile. I miss wearing a towel that could contain the mess when she was smaller!

The neurologist ran other tests and one of them pointed to a fatty oxidation disorder. We had a consultation with a Metabolic specialist but she has decided to hold off on a muscle biopsy unless another surgery is scheduled. Anna has never presented with a crisis. She has constant snacks and always has a sippy to bed with her. We always feed her whatever we had for dinner as she started solids so she always has a low fat, high carb diet. I also have a low fat high carb diet because I run and do aqua aerobics 3x a week. Anna isn't carnitine deficient based on the blood tests (not fasting ones) so we are in a holding pattern.

The above is about Anna's possible disorder. Let me tell you about Anna. She is blond-haired, blue-eyed, and very beautiful. She has long, slender piano hands. I think she will make a great pianist or surgeon with such skillfully shaped hands. I have lots of hopes for her. She loves reading books and will pretend to read the words to herself to her favorite stories now. She is learning 2-3 new words a day by pronouncing words she hears around her. Starting to walk later didn't slow her down one bit, it made her language get better. She sings along, claps her hands, plays her xylophone and dances with The Wiggles. Her evening show is Wheel of Fortune for learning her letters and she loves to encourage the contestants. It's as good as Sesame Street is in the morning. Anna is very shy around other children and prefers to be around other adults. I am proud to say that her biggest asset has been, and probably always will be, her mind. This 22-month old girl is the most stubborn yet sweetest miracle in our lives. She is passionate about cats and cuddles with every cat doll we have. She gets to go to the park (and the mailbox!) every day. She loves the big red heart on my keychain and chews on it all the time. Her weekly trip to the Children's Museum toddler time is a source of delight. She loves to go to story time. Playgroups are finally interesting to her. She is my daredevil, trying to rock herself standing on her rocking chair (falling doesn't dissuade her!). She will pretend to cook in her kitchen area when I am cooking in mine. We make funny faces in the mirror together and laugh. She will ask you to tickle her. The joy she brings is indescribable and I couldn't imagine what life was like before she illuminated it.

Andrea Todsén
bjorka7769@yahoo.com
Albuquerque, New Mexico



Medical Update

Volunteers Taking Cholesterol-Lowering Drugs Needed for Research Study

The Robert Guthrie Biochemical Genetics Laboratory at the Women's and Children's Hospital of Buffalo, Kaleida Health System and University at Buffalo is currently seeking participants for a research study. Participants should be adults who are:

(1) Parents of affected individuals (obligate carriers) or affected individuals with a lipid storage disease that is characterized by skeletal muscle symptoms, for example, carnitine palmitoyltransferase (CPT) II deficiency. (VLCAD, LCHAD, MCAD, and carnitine deficiencies will also be included). Note: Participants may or may not have muscle symptoms at the time of entry in the study.

(2) Taking or have taken cholesterol-lowering drugs in the past with or without the presence of muscle symptoms. Common cholesterol-lowering drugs include Lipitor, Zocor, Pravachol, Crestor, Mevacor, and Leschol. Other lipid-lowering drugs of interest to our study include fibrates, such as Gemfibrozil (Lopid).

The purpose of the study is to determine whether carriers or affected individuals with lipid storage diseases are at increased risk for developing new or more severe muscle symptoms while taking cholesterol-lowering drugs. In preliminary studies, we have observed an increased prevalence of both carriers and affected individuals with certain lipid storage diseases among those who develop muscle symptoms. Approximately 2 teaspoons of blood is needed for the study and a questionnaire will be given to participants for a health history before, during and after drug therapy, as applicable. The principal investigator will reimburse participants for blood drawing fees and shipment costs. The benefits to participants include the provision of risk factor information that may help with future medical care.

For additional information, please call Jeanne Catalano at 716-829-3141 ext. 148
Tuesday/Thursday or by email at jcatalan@buffalo.edu.

Note: Grandparents of affected individuals are also encouraged to participate.

The study protocol has been approved by the Health Sciences Institutional Review Board of The University at Buffalo. The Principal Investigator is Dr. Georgirene D. Vladutiu, Professor of Pediatrics, Neurology and Pathology at the School of Medicine and Biomedical Sciences, The University at Buffalo.

Health Sciences IRB Approval

FROM: 8/26/05 TO: 3/22/06

For IRB Use

Research Opportunity for FOD Families

The Human Genetic Cell Repository (HGCR) at the Coriell Institute for Medical Research is currently seeking volunteers to donate a blood sample for research. Any individual with an inborn error of metabolism, such as a fatty acid oxidation disorder or an organic acidemia, is eligible to participate. The also accept first-degree relatives of affected individuals. This would include children, parents and siblings. The HGCR, which has been sponsored by the National Institute of General Medical Sciences at the NIH since 1972, serves the research community by collecting, storing, and distributing cells from individuals with genetic diseases. Qualified researchers can obtain these cells to study how cells function, to identify new mutations, and to develop ways to diagnose, treat, and possibly prevent genetic disease.

Due to the expansion of newborn screening in the United States, the HGCR has undertaken a project to obtain samples from all 29 disorders on the newborn screening panel. The increase in the number of disorders tested for via newborn screening will generate new research initiatives to better understand these disorders. With your help, the HGCR can help provide the appropriate materials needed to accomplish this research.

A blood sample and clinical information about the donor is all that is required. All samples and information will be kept anonymous, bar-coded, and no identifiable information will be released to researchers. For more information or to sign up to donate, please contact Tina Sellers, MS, Genetic Counselor for the HGCR at (856) 966-5062 or tsellers@coriell.org.

Research opportunity: Children with long-chain fatty acid oxidation disorders

A new study looking at the effects of 4 months of a high protein diet on body weight and metabolic control of fatty acid oxidation disorders is being conducted at OHSU. If you have LCHAD or TFP deficiency or if you have VLCAD deficiency and are 8 years old or older, you may be eligible to participate. Participants must come to OHSU and stay at the Clinical Research Center for 4 days on two different occasions. Participants may be asked to follow a standard high carbohydrate diet or a diet high in protein. The chance of being asked to follow the standard high carbohydrate diet is 50%.

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

OHSU

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OHSU is an equal opportunity, affirmative action institution.

Recipes

POTATO SOUP

1 ounce Canadian Bacon, chopped
1 can (14 1/2 ounces) fat free chicken broth
1 celery stalk, chopped
1 small white potato, sliced
1/4 cup chopped onion
1/4 cup minced parsley
1 cup chopped cabbage
1 cup chopped carrot
2 1/2 tablespoons flour
1/4 cup water
1/2 cup evaporated skim milk

1. Cook the first eight ingredients in a sauce pan until vegetables are tender
2. Mix flour in water until it dissolves. Add it to the sauce pan and cook for approximately 2 minutes
3. Add evaporated skim milk and heat through *Salt and pepper to taste. Extra salt will increase the sodium content.
Makes 3 servings
Serving Size: 1 cup. Calories 142, Protein 9g, Carbohydrate 26g, Total Fat 1g, Sodium 252mg.

Betsy Furler, Houston TX
Henry, unspecified FOD

Texas STROGANOFF

2 tablespoons dried onions
1 pound lean ground beef, chicken, or turkey, browned and drained (I add the onions to the meat)
1/4 teaspoon powdered garlic
2 tablespoons flour
1 teaspoon salt
1/4 teaspoon seasoned salt
1/4 teaspoon pepper
1/4 teaspoon paprika
2 4-oz cans of mushrooms
1 10-oz can low fat cream of mushroom soup
1 cup low fat or fat free sour cream

Combine meat, onions, garlic, salt, pepper, paprika, and mushrooms in a large saute pan. Saute for 5 minutes. Add soup and simmer for 10 minutes. Stir in sour cream. Serve over rice or noodles. Serves 4-6 (we do not have all the fat etc amounts for this recipe)

Betsy Furler, Houston TX
Henry, unspecified FOD



Question: How are carnitine levels monitored?

Answer: Monitoring Free Carnitine levels is most important after a diagnosis is made. "Free" is the carnitine available for the body to use as needed. The AC/FC ratios can be helpful in helping us identify kids with metabolic disorders but often becomes normal after treatment is started and can be abnormal in metabolic crises. It is most important to follow the "free" carnitine levels. It is the free carnitine that is available to bind with organic acids and get them excreted in the kidneys. The "total" carnitine can be helpful when measured in blood only if it is low. Remember that carnitine is mostly found in muscle where the amount can be 10-100x what we measure in blood. So, if total carnitine is low in blood, it is VERY low in muscle and we see more hypotonia etc.. Most of us no longer follow AC:FC ratios after diagnosis is made because the ratio is altered by the disease specific acylcarnitines - the free is most useful for chronic management.

Lynne

Metabolic NP

Questions: How painful is a biopsy? Where, normally, is the biopsy taken from? I've heard some Clinics will use fresh biopsies? Any comments? Has anyone regretted having a biopsy done? Has anyone had a biopsy done with negative results only to still get an FOD diagnosis? Are skin biopsies usually reliable and should I consider a skin biopsy before a muscle biopsy if the suggestion is made?

Answers: Great questions...

1) Skin biopsies are usually done before muscle biopsies because they are less invasive & less painful, and because many times enzyme testing for multiple disorders can be done on the skin culture (aka fibroblast culture) because some of the cells can be frozen and re-grown as needed. But not all enzyme function that is abnormal will be found in skin biopsies. The general rule is we are best able to detect an abnormality in the tissue most affected. Skin biopsies can be done on the buttocks, the forearm etc...

2) Muscle biopsies are usually done on the thigh in children. Motrin or Tylenol is all that is needed for pain control. It is a very small incision, but some kids will refuse to walk for a few days. Muscle does not re-grow like skin, so IF this is what you pursue be sure the Doctors get all the muscle they will need for multiple tests...needing more than one muscle biopsy is not your first choice. Muscle tissue provides us with a really good look for a number of structural abnormalities in muscle which can lead to a diagnosis plus enzyme testing for many Mitochondrial disorders, Carnitine disorders and some of the FODs especially SCAD.

No test is ever 100% guaranteed to find an answer, so it may be possible that no diagnosis will be found no matter what the Drs look for right now - just a limitation of our science today. It is possible that a diagnosis could be made with just blood & urine tests without any biopsies etc.

Hope that helps,

Lynne

Metabolic NP

Welcome New Babies...

Sara and Steve Cywinski welcomed Ashlyn into the world in March 2005.
Brother Nicholas (MCAD) is thrilled to have a sister!

Rodney (adult MCAD) and Deb Porter are excited to have a new brother for their daughter, Anya ~ Alex was born May 4, 2005.

Jennifer and Darrin Arveson also welcomed a new baby brother, Dane (unaffected) to their home on May 16, 2005. He was 7 lbs 11 oz and 19 inches long. Big brother, Gabe (MCAD) loves him A LOT!

Melissa (GA2) and Doug Cummings introduced their 4th child to his siblings on September 22, 2005. Zachary Richard (unaffected) 8lbs 5 oz and 21" long, has big sisters, Seirra and Shawna (MCAD) and big brother, Vincent, to play with now! Their picture is on the KidsKorner page.



Love Messages

Archived 'Love Messages' are available in previous issues on our website, <http://www.fodsupport.org>

New Love Messages

Mary Thorson
Wendy - Birth Sept. 20, 1987
Death Sept. 10, 2005

*'In the dark night of the soul,
bright flows the river of God.'*

~St. John of the Cross~



Pharmaceutical Update

If your Physician needs more information about L-carnitine (Carnitor®), dosages, or has other questions, please have him/her contact Sigma-Tau Pharmaceuticals, Inc., and ask for the Medical Information Department or state that he/she has a question about carnitine. This service is available around the clock 7 days a week. The phone number is 1-800-447-0169.

A new L-carnitine (generic form) supplement was approved for distribution by the FDA a year or two ago. This generic form (as well as the brand name Carnitor®) needs a Prescription from the Dr. Please also note that the generic 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 <http://www.fodsupport.org/pharmaceuticals.htm>). The term 'generic form of a drug' should NOT be used interchangeably with the term 'over-the-counter supplement.'

Condolences

We all had a very heavy and saddened heart when we learned of Wendy Thorson's (TFP) death on September 10, 2005. Wendy had medical challenges her entire life mainly due to not being diagnosed until she was a teenager (initially as LCHAD). She was just shy of her 27th birthday when she died after almost a week of trying to fight off an infection. Wendy was a gift to not only her mom, Mary, and her siblings, but to the entire FOD Family. Please keep the Thorson Family in your prayers.

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

NBS Update

FOD website: Be sure to visit our website (In the News page) for the current articles on NBS efforts across the US and Canada. 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!

Kids Korner



Meaghan Blankenship
(Dietrich) Unsure
diagnosis



Jessica LaVallin, UK MCAD



Jordan Dougherty, VLCAD



Fiona Johnson (MCAD) and dad



Cummings Family ~ Shawna
(MCAD), Zachary (baby),
Seirra, and Vincent



Carson and Caden Richards (GA2/MADD)

Resources

The KID Foundation (<http://www.kidfoundation.org/what/index.html>) focuses on research, education, and advocacy related to Sensory Processing Disorder (SPD). SPD is a complex disorder of the brain that affects developing children (and some adults). People with SPD misinterpret everyday sensory information, such as touch, sound, and movement, giving them a different experience of the world from everyone else. For example, some feel bombarded and overwhelmed by ordinary sensations. Some can perceive sensory information but can't interpret it correctly. Others seek out intense sensory experiences. This can lead to behavioral problems, difficulties with coordination, and many other issues. Children with SPD are often misunderstood and labeled as aggressive or clumsy. They frequently are socially isolated and have trouble in school. Effective treatment is available, but far too many children with SPD are misdiagnosed and not properly treated. The KID Foundation (the Foundation for Knowledge In Development) was founded by Lucy Jane Miller, Ph.D., OTR, in 1978 to help these children and their families. The foundation's goals include:

- Increasing public and professional awareness of the scope and severity of Sensory Processing Disorder
- Providing information and resources to families, educators, physicians, and other health care professionals
- Supporting advocacy for and research on Sensory Processing Disorder

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Help for the Child with Asperger's Syndrome: A Parent's Guide to Negotiating the Social Service Maze

By Gretchen Mertz, Foreword by Tony Attwood

Jessica Kingsley, 240 pages

(This review came from another List I (DLG) am on. This book may also be helpful when dealing with other medical/behavioral/educational issues that require parents to seek out services for their child/children.)

From the publisher: Parents of children diagnosed with Asperger's Syndrome frequently find themselves embarking on an exhausting and disheartening hunt for the services and interventions that their child needs – and is entitled to. Gretchen Mertz presents a readable and comprehensive parent's guide to developing an intervention program for a child with AS.

Beginning with the search for a diagnosis, she outlines an accessible overview of the helpful and appropriate services that are available, how to find them and get funding for them, what to expect of them, and how to evaluate them. The issues raised and the information given will be pertinent to any parent of a child that has (or may have) AS. It will also be an invaluable resource for social workers, psychologists and psychiatrists who wish to find or offer appropriate support for children with Asperger's Syndrome.

From Robert Naseef: How do you avoid being chronically angry while getting services and still be an effective advocate for your child? Gretchen Mertz threads the needle on this vital issue guiding the reader with the lessons she learned parenting her own child who has Asperger's syndrome. This is a thoughtful and intelligent guide to effectively negotiating rather than fighting the fragmented system of services.

2006 FOD and OAA National Metabolic Conference Information

Location: Adam's Mark Hotel 400 North Olive Street, Dallas, TX 75201
<http://www.adamsmark.com/dallas/index.asp>

****Special room rate: \$ 95.00 (single/double) ~ You must state that you are attending the FOD/OAA National Metabolic Conference hosted by The Institute of Metabolic Disease**

Call for Hotel Reservations: (214) 922-8000 (Each Family/Professional makes their own reservation)

Transportation to/from the airport (DFW and Love Field): (Each Family/Professional must make their own reservation on the Yellow Checker Shuttle website (www.yellowcheckershuttle.com/jeff) in order to get the discounted price of \$13.00 one way – or book your own company choice)

- June 22 Welcome Reception/Cocktails 6:30 - 11:00 pm at Hotel
- June 23 Metabolic Conferences (Each Group in own room) 8:00 am - 5:00 pm
 - June 23 Dinner on own ~ Good time for Family Networking 6:00pm on
 - June 24 Combined FOD/OAA Session 8:00am – 12:00 noon (wear T-shirts!)
 - June 24 Tour of Institute of Metabolic Disease 2 – 4 pm (van provided)

Family & Professional

Family Donations: Darlene and Ted Carson, Judy Jilderda, Janice and Richard Cox, Ettie and Arthur Alligood, Bobbie and Warren Fields, and Gina and Chet Revinski in memory of Wendy Thorson (adult TFP). Alecia Park and Lee Bernstein, and Lauren and Jonathon LaConti in honor of Alexis Hammer's (MCAD) 3rd Birthday. Judy Solores and Anne and Lawrence Lodding in memory of Nolan Mitchell (undiagnosed LCHAD). Kay Simon in memory of her grandson, Nolan Mitchell (undiagnosed LCHAD). Marcella Lineberry and Family in memory of Matthew Christerson (adult VLCAD). Dorene and Ron Bellaire and Family in memory of their granddaughter, Alyssa Boucher (Undiagnosed MCAD). Stephanie and Andrew Plaisted in loving memory of their son, Drew (undiagnosed MCAD), on the '5th year of his passing.' Leslie and Robert Zemeckis in honor of Zane (MCAD) and Rhys Charles. Thank you for the hand-knitted scarf too, Dorene! 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 - erikawallacepa@yahoo.com (Mailing Lists), Mary Lingle - Mcartwrite@aol.com (Web Page) and Brian Gould - gouldbr1@msu.edu (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, 2006**. 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, 2006**. Also, please return to Deb the **Professional Questionnaire** even if you are already listed on the printed Professional List.

Your vision will become clear only when you can look into your own heart. Who looks outside, dreams; who looks inside, awakes.

~ Carl Gustav Jung ~

COMMUNICATE WITH US

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

Pease REMOVE me from your mailing list
 Name/Adress:

Please include ideas for future issues or your questions.