



FOD

fatty oxidation disorder
communication network

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The FOD Communication Network Newsletter was created by and is currently edited by Deb and Dan Gould, 805 Montrose Drive, Greensboro, NC 27410 (phone/fax) 336-547-8682 <http://www.fodsupport.org> email: fodgroup@aol.com or deb@fodsupport.org
Medical Advisor: Dr. Charles Roe; cr.roe@baylordallas.edu



From the Editor

It has been a busy time for many in our Group. The summer heat is definitely here, but the HEAT IS ON for states to get onboard to test newborns for FODs. To see a complete listing of NBS media exposure go to www.tylerforlife.com web page. Their Main Mission is to advocate comprehensive newborn screening for 30 disorders using Tandem Mass Spectrometry, so they are very up-to-date on the latest developments. If you'd like to keep informed of all NBS efforts, also sign on for their NBS Advocacy Discussion Email List on their site. Please also notice the NBS Fundraiser Letter enclosed in this packet.

FOD Email List members have also been extremely busy. There has been an **exciting explosion of FOD Newborn Screening Awareness and Advocacy across the country** the last several months. If you are interested in **getting involved in Advocacy efforts, Gina R(VLCAD)** has compiled an "**Advocacy Packet**" of FOD information, articles, etc., that you can **take to hospitals, parent groups, or anywhere that you may want to spread the word about screening babies EARLY so lives can be saved!** Please contact her **if you would like a Packet ~ MJB3@frontiernet.net or call 914-928-9574.**

Networking has been very active on our **Email List**. Please be aware, however, that **when you Register for the Group on our web page** (for the printed newsletter) **that does NOT automatically sign you up for the Email List. The Email List has a separate registration.** Once registered/confirmed, you will be able to send messages to over 160+ individuals. Please take advantage of this supportive network!

Thank you **Dr. Georgirine Vladutiu** for generously sharing your knowledge and expertise on CPT 2. We are always looking to expand our knowledge on ALL the FODs and your article helps achieve that goal! **Professionals – PLEASE think about sharing your own expertise** so we can learn about the more rare FODs, in addition to what we know about MCAD and LCHAD.

Please notice all the extra INSERTS in this issue, such as a **Release of Information Form when you submit an article or story, an FOD Pen Pal Permission Form**, a listing of some of the **US Diagnostic Labs that test for FODs**, a **NBS Fundraising Letter, and for FAMILIES – a very important SIDS/Reye's Syndrome Survey.**

Also included is a Family/Professional List Update -- complete Lists will only be sent in the January issue.
Keep up the great work of spreading the word about FODs and Newborn Screening!

"We Are All in This Together!"

Take care ... DLG



IN THIS ISSUE

From the Editor...1	Letters to the Editor...2	Meaning of Our Logo...3	Family Stories...4	Medical Update...9
Love Messages...14	Kids Korner...17	FYI Insurance...18	Resources...18	Newborn Screening...19
Notes...19	Donations...20	Reminders...20		



Letters to the Editor

Dear Deb,

It has been one year since Megan's diagnosis of GA 2, and I wanted to share with you, and the other families, how things are going. As you may remember from our last story, Megan was left seriously delayed in all areas due to the effects of her undiagnosed GA 2. It took 17 grueling months to get a correct diagnosis. She is now two and a half years old. Megan has made some wonderful strides in many areas including: she now eats and drinks all of her nutrition by mouth (her feeding tube was removed one month ago), she can get up without assistance from the floor, she can walk across grass and small rocks without assistance, she can climb onto low surfaces, she has enough strength to hold and use crayons, she is speaking in sentences, and Megan is very expressive now. She really enjoys character acting to her favorite movies, including *The Wizard of Oz*. Some of her favorite things to do are play "house", read books, go to the playground, and anything outside. We are thrilled with her progress! Megan's suspected cognitive delay was not true, but was instead, a symptom of her being unable to move around and interact with her environment effectively. Now her limits remain primarily in gross motor strength, fine motor strength, and speech intelligibility. She has a sparkling personality and is a joy to be around.

A year ago, we were having trouble finding a pharmacy that would make the riboflavin suspension for us. The first pharmacist that agreed charged me \$40.00 for a small bottle only partly filled. I thought this was too expensive, so I searched further, and the next charged almost \$30.00 per bottle. Finally, with the help of one of the families that read our story, including my trouble getting riboflavin, I now get it through Sentry pharmacy in Milwaukee for just under \$5.00 a bottle! Thank you, graciously, for your suggestion.

Megan's diet is closely followed by her dietician from the Genetics Center. We still calculate amounts and calories for each meal, as we've done her whole life, but now, we also pay attention to fat and protein grams. We are becoming more lax in calculating these things as Megan becomes better at reading her own body cues of hunger, satiation, etc. She takes carnitine, in addition to the riboflavin, and a Flintstone vitamin. Along with typical baby and toddler foods, she also gets daily servings of Nutramigen (a baby formula), Rice Dream (a non-dairy drink with calcium), and corn starch (mixed in her formula to drink just before bedtime). Getting Megan to eat and drink orally has been one of our greatest challenges. We feel so confident now that we have overcome that obstacle. Despite her history of severe feeding difficulties and her metabolic disorder, Megan's nutrition is now superior to that of most children.

In the last year, Megan has experienced two serious episodes of vomiting due to a virus. One of the incidents required hospitalization to get her stabilized, and the other episode Megan overcame with only the use of her G-tube and some Coca-cola at home. I must admit, I fear for her getting sick. But, I hope that as she gets older, she will be less susceptible to illnesses and vomiting.

We have enjoyed being a part of this Network and reading all the family stories. Although we will never meet all of the families who share their stories, we feel a connection with every one of them.

Warm regards,
Stacy and Michael Ladwig
Captain@414.org

THANK YOU

I would like to personally thank Jeff Schmidt (Stacey, Undiagnosed) for developing and working on our 1st text website from 1996 - 2000. Thank you Jeff for generously volunteering your time and doing such a GREAT job over the years! -DLG

Meaning of our Logo...

Dan and I began the MCAD Family Support Group in 1991 to honor the memory of our 21-month-old daughter, Kristen, who had died suddenly on July 21, 1985 at our home in Champaign, Illinois, from what doctors initially called Reye's Syndrome. By 1991, we had already been working through our daughter's death from undiagnosed MCAD and our son's diagnosed MCAD for almost six years and five years, respectively. During those years, we felt as if we were the only ones in the world dealing with this disorder, but we just knew there had to be others out there going through similar experiences.

After discussing our concerns with Dr. Charles Roe, a metabolic specialist and researcher who diagnosed Kristen (1 year post-mortem), Kevin (at birth), and Brian (an MCAD carrier), we embarked on a journey to connect all the MCAD Families across the United States. Dr. Roe's encouragement helped us get started. To maintain confidentiality, Dr. Roe mailed our 'introductory' letter to all the families that had been diagnosed through Duke University Medical Center, which was where he was doing MCAD research at the time. He is now at Baylor University Medical Center as Medical Director of the Institute of Metabolic Disease in Dallas, Texas. He has been our Medical Advisor for the Group since 1991.

When we first began with our 10-15 MCAD Families, we did not have an established logo to distinguish our Group. I (Deb) had first thought about using a simple rose design that my sister-in-law had drawn for me to symbolize my own journey through grief, but I felt that it did not fully reflect the larger Mission of the Group, which was to connect and emotionally support all of the MCAD Families in the U.S., and maybe someday across the world!

In 1996, after we had expanded our Group to include all of the Fatty Oxidation Disorders, we sent out word to our 250+ Families (now we have over 400) to **help us create a Logo that would embrace the essence of what we were trying to convey as a now international FOD Family Support Group ~ that "We Are All in This Together!"**

In our July 1998 Newsletter issue, we 'unveiled' a **design created by Melanie and Dell Ruff of Franklin, Tennessee, whose daughter, Anna, has MCAD.** In their design, the heart and chain symbolize all of our Families in the Group connected together in spirit no matter how far apart we live. The 'broken link' represents a missing enzyme(s) that each child/adult has that is dealing with an FOD. Their design symbolically demonstrates that despite that missing link in the metabolic process, we are all here to work together, as families and professionals, toward medically dealing with these disorders, as well as emotionally supporting each other.

THANK YOU Melanie and Dell for sharing your creativity and truly giving meaning to being "All in This Together!"

Deb Lee Gould, Director
FOD Family Support Group

Legal Concerns with your Insurance Carrier???

Mike McConnell (daughter Morgan, LCHAD) has offered his legal expertise to our Families in regard to coverage claims against insurance carriers; something they have experienced firsthand. They were denied reimbursement expenses for physical/occupational therapy, even after they were initially approved. The Insurance carrier said those services were educational and weren't covered! So if you have a coverage issue and think you might have reason for legal action, contact Mike at MMconn807@aol.com. Assuming his law firm has no conflict of interest with the insurance company, he will try his best to help you.





Family Stories - Morgan, GA 2

After two miscarriages and two failed in-vitro fertilizations, my husband and I were finally going to have our baby. I had begged my doctor to induce labor because I had retained a lot of water and I had developed a blood clot in my leg. It was hard to move at all. So on August 4, the day before my due date, I was admitted to hospital and induced. The next day, after many epidurals and only dilating to six centimeters, they decided to perform a C-section. Morgan Lynnae Jones was finally here. Five minutes after birth, she had apnea, and they whisked her off to the NICU. A few hours later, my husband tried to bottle feed her because I was still recovering. She could not keep any of her food down that first day, and still had trouble the next few days. The hospital gave her Isomil and said that it was just a lactose intolerance. They also said that she cried all of the time, but they said that it could be normal.

After being in the hospital five days, we finally got to bring her home. She always spit up and cried all the time (I had ear plugs around the house). Our pediatrician said that she looked good and that it was just irritable crying syndrome. We did not know any better because Morgan was our first child.

In November, our pediatrician noticed a few things. She noticed that her eyes were not focusing yet and that Morgan's head seemed to be larger than other children her age. First she sent us to see an ophthalmologist. He was perplexed at what he saw, so we were sent to see a pediatric ophthalmologist at the University of Kentucky. This doctor told us that he had never seen anything like this before. He thought it could be Morning Glory Disk Anomaly, but he was not sure. A retina specialist also looked at her. They both were stumped. The most they could tell us was that she possibly would be legally blind. Next, we went to Cincinnati Children's Hospital to see a neurosurgeon. He looked at her CAT scan and said that things looked normal. He determined that my husband's family had large heads, so she was just taking after his family. We thought everything would be OK other than her eyesight, but we were just happy she was here.

On January 9, I fed her cereal and a bottle. About five minutes later, it all came back up. When I went to change her I noticed something orange in her diaper. I got scared. I called the doctor's office, and they told me to go to the ER. We got to the ER at 11 am. They determined that she had the flu and that we were going to stay overnight. It was 5 pm when she finally got 'fluids, and she had not kept anything down that day. That night and the next day, she seemed to be okay. She could keep down Pedialyte, but no formula so we were going to stay another night. On that night the IV box beeped for 20 minutes. After not having fluids for that short of time, she started to have a seizure. Our pediatrician decided there had to be more wrong, so she sent us by helicopter to Cincinnati Children's Hospital.

The hospital did every test imaginable: X-rays, blood tests, EEG, MRI, eye exam, retina exam, and eye ultrasound. We had talked to so many doctors, we could not remember anyone's name. After two days of tests, they discovered that she had a small frontal lobe with fluid and blood around it, and that her retina was somewhat detached with blood on it. They immediately brought the Child Abuse Team. They asked us all kinds of questions and we were as open and honest as we could be since we had nothing to hide. The next day they told us that they were going to keep us there for four more days until they had some answers. During these days, Morgan had two more seizures because they had taken her off the IV fluids and she still was not keeping anything down. A geneticist came in and said that he was going to test for a metabolic disorder, but he did not think she would have it. All we could do was pray, and ask everyone we knew to pray for an answer. These were the worst days of our life. They wanted us to admit to something, but we didn't know what had happened either!

Finally on January 18, they let us go home, but without any answers. Two days later they called us and said they may have found something. It may be a fatty-acid oxidation defect and this is probably what caused my problems during pregnancy, her constant crying and her so called "injuries." We were glad that we finally might have an answer.

We went back up to Cincinnati that next week. They took a skin biopsy to send to Dr. Roe. At this time, they put her on Carnitor and a seizure medication. **After six weeks we got the results back. Morgan had GA2 or MADD.** At this time they put her on Riboflavin (she will only take it with caffeine free Dr. Pepper in a syringe), Polycose powder and a high carb. diet.

Continued on page 5

Family Stories - Morgan, GA 2 (cont'd)

Morgan is now seven and a half months old and all of her doctors are amazed at her progress. Her eye doctor says that some of the blood on her retina has disappeared and she is seeing a lot better. She can now follow objects, pick up objects and can respond to our facial expressions. The neurologist could not believe how much better her muscle development was. She can now roll over, hold her head up and sit up with some support. She can also stand with support, and they told us that she may never walk. They never expected her to do so well, but we did because we knew that we have a God who can do anything! We are thankful to God for giving us our precious little girl.

Chad and April Jones
Grayson, KY
606-474-2070



Family Stories - Kayla, VLCAD

When I was pregnant with Kayla my husband and I were so happy. Just as any other first-time expectant parents would be. The pregnancy was very normal until the end. There were some complications but nothing very serious. On July 7, 1992 only one day before my birthday our daughter was born. She was a healthy beautiful baby. She had normal APGAR scores and everything seemed perfect. I had a C-section so we were in the hospital for a few days but it was finally time to go home.

I woke up in the morning and they hadn't brought Kayla in to me to feed her yet. I thought this was strange but my mother told me to rest because when I get home it will be difficult to get much rest. I fell back to sleep and when I woke up there was still no sign of Kayla. I decided to get up and start packing up our stuff. My husband was on his way to take his new family home. I finished packing and thought I would go for a walk and see Kayla asleep in her little bassinet. When I got to the nursery she wasn't there so for some reason I walked to the NIC unit. Well I found her right there in front naked and tubes coming out from all over. I couldn't believe what I was seeing!

The Doctor came out and said Kayla's blood sugar dropped very low and they are testing to see what the problem might be. I went home that night and was so depressed. This isn't how it is supposed to be. We were all supposed to be home as a family. Little did I know at that point anything that was supposed to be normal would never be again.

The doctors continued the tests and my husband and I visited daily until she was released. The tests all came back normal and it was explained to us that sometimes new babies' blood sugars may drop for unexplained reasons. When we got home and over the next three months things seemed fine. Kayla seemed to sleep a lot and not eat as much as I would have liked but I was told by the Doctor that I was just one of those first time moms.

About five days before Kayla was in the PIC unit, I took her to the Doctor again telling him that she is sleeping a lot, not eating a whole lot, and vomiting. That day I took her to the office she was screaming and I told the Doctor that this is the most she has cried in four days. He told me that she was colicky. At this point I was very upset because I knew she wasn't colicky. Well five days later all hell broke loose. Kayla spent the day not eating and vomiting. She seemed to be choking but I found out later she was having seizures because her blood sugar was so low. I immediately called the Doctor. He said she sounds like she is getting dehydrated and to give her some pedialyte and bring her to see him in the morning. Kayla woke up at 12:30 am and her body was jerking again as if she was choking. I called the Doctor and told him I was going to the hospital. When I got to the hospital, they did a bunch of tests and sent her to the pediatric floor for observation.

Continued on page 6

Family Stories - Kayla, VLCAD (cont'd)

The Doctor came in and said she was very sick and they don't know why. Kayla's liver was three times its size, she had about 2 oz. of fluid around her heart and her blood sugar was very low.

When I looked back on all this after the fact I realized Kayla had more strength as a newborn than as a three-month-old. After conferring with the Doctor they felt she needed to be sent to a hospital that had a PIC unit. Kayla was sent by helicopter to Rockford Memorial where she spent the next month. After we got there her lung collapsed and things were not looking good.

Two weeks after getting to the hospital we finally had our diagnosis -- she had VLCAD. The Doctors at that point gave us the list of things we could expect as Kayla lived her life. They began treating her aggressively and in two weeks we were able to go home. Everything seemed fine until she was 15-months-old. From that point we were in the hospital once or twice a month. She developed asthma at the age of two. At the age of six my husband and I decided Kayla had to have a G-tube placed. We had it placed in January of '99 and we were out of the hospital until July 4 and we haven't been back.

Kayla has been sick in that time-span but we have been able to monitor it at home and I have gotten very good at figuring out the signs of when she needs to be supplemented to avoid an episode.

Kayla is now seven years old and full of life. The list of complications that the Doctors said we should expect with VLCAD has not happened. She is doing very well in school and has participated in numerous physical activities without complications. She did need speech therapy at the age of three and four but not since.

Kayla is very aware of her body and she knows when she has to eat and when she needs to take it easy. When Kayla is healthy you would have no idea that there was anything wrong but when she gets sick she is very sick. Her legs are the first to bother her. She can't really walk, then her appetite decreases, she will begin vomiting, and then her blood sugar will begin to drop. We never know whether her VLCAD is acting up because of illness, her asthma or any other number of reasons.

I have stopped trying to figure it out and just treat what I need to so she can get back to her regular life. Kayla eats a normal diet and every night before bed she has a shake consisting of skim milk, carnation instant breakfast, 2 TBSP polycose, 1 TBSP dextrose, and 1 TBSP MCT oil. She also takes Carnitor, and allergy/asthma medication daily.

Kayla is a very bright child and knows what is going on. She was hoping that **if there are any other children that would like to be pen pals with Kayla, please contact me.** She really wants to know if there is anyone else that has to go through what she does and sees it from a child's point of view. Thank you all so much for your stories. They always seem to inspire me when I need them the most.

Kelly & Michael Madej
Aurora, Illinois
yadmad@aol.com

New Baby! Claudia, Rick, and Nicholas Evans (undiagnosed FOD) proudly announce the birth of their son and brother, Aaron James (unaffected) on March 24, 2000. Congratulations to the Evans Family!!

Online Love Message Memorials

Please share your poems, stories, artwork, pictures etc that honor the memory of your child, grandchild, sister, brother, niece or nephew that died before or shortly after receiving an FOD diagnosis. Because these may be displayed on both the graphic (NEW!) and text version of our web page, please **send your contribution along with a signed Release of Information Form included in this packet.** Please indicate if you want your name, address, phone and/or email listed on the site.





Family Stories - Natasha, undiagnosed FOD

Dear new and old friends,

Things are looking brighter with regards to Natasha. Just an update along with the readers digest version of her story. For some it will be redundant... I am sorry, but seemed too hard to try to separate it all out. Thank you for your patience and friendship over this long road with us.

I have waited to get this update to you because it is still sinking in. Some of you know, but for others... We have a lovely 4 year old daughter Natasha. She was born 4 pounds, 17 inches almost full term after the most horrifying terrible long awful pregnancy (I had: Pre-eclampsia, Preterm Labor, Hyperemesis for 4 months, lost 46 pounds, Subclavian line surgically implanted, spent 2 months in the hospital, bedridden 6 months, liver problems and a C section)

At birth she was adorable, healthy and full of life right from the start. Came home 2 days after birth. She has remained remarkably healthy except for 3 unexplained severe dehydration problems that resulted in hospitalizations for a few days. The first was the worse, the doctor refused to admit Natasha and she threw up for 24 hours. She became almost non-responsive.

Also, she has remained below the 3% in growth and weight. Pretty healthy though. No infections, few fevers, few runny noses, no reflux, and a good appetite for her size.

In December 1998, at 3 years old Natasha was 32.5 inches and weighed 24 pounds. We insisted that Natasha have testing to see if she was a dwarf and if so prepare ourselves to help her be all that she can be.

It has been anything but easy. In a nutshell without too much drama this is what happened:

Approximately March 1999 she had some baseline tests. Bone age, Baseline growth blood tests etc. WE insisted for a referral to a specialist. She was seen by an Endocrinologist at a San Francisco Teaching hospital. Meanwhile we decided to get our daughter a Pediatrician. The town we live in is too small and we have to travel a bit to get to one. We got a referral from a social worker and changed primary care. The new doctor started a nightmare that wouldn't end till we got a new one, but at the time we didn't know.

I will not go into all the terrible stuff she put us through, but she attempted to diagnose Our princess with over 8 different some horrible, illnesses. None of which Natasha has (heart failure, Fanconies anemia, hypochondroplasia, turners, smith limi opitz syndrome, Renal Tubular Acidosis, LCHAD, MCAD and the list is much longer). It has been horrendous. Her physician was not available during any of the emergencies with dehydration and they were disasters. It has been a nightmare. On top of that, we had taken her to several specialists at a San Francisco Teaching hospital and they wanted to do a battery of invasive and provocative testing. We felt it was overboard. We did allow blood work ups and said no to the invasive testing till we could get second opinions. On one of the last visits with her pediatrician she tried to tell me some other awful possibility and questioned us for wanting a second opinion. She was not supportive on us getting a second opinion and I wouldn't even listen to her tell me about this other terrible disease she suspected.

Luckily we have had tremendous support from Natasha's Nurse Case Manager and she was extremely instrumental and helpful for us to obtain second opinions and we made a 3rd change of primary treating physicians. She has 3 children herself and said she changed 5 times till she met a doctor she trusted for her kids and that we should not feel bad, just look out for the best interests for Natasha. Through a friend that I actually met through the FOD network, I got a recommendation to a geneticist at another teaching hospital in San Mateo County. We met him last month. He wanted Natasha to be followed up by the same specialties at his hospital to see where they would recommend going or doing next. From the Geneticist point, Natasha may or may not have a genetic/metabolic problem and would consider for her to have a skin biopsy for further ruling out, but after seeing the other specialists -- make it a interdisciplinary approach.

So here goes:

Monday Dec. 29th, met with Chief of Pediatric Gastroenterology at the hospital to get second opinion on whether Natasha needs- NG feeding tube, and a Bowel Biopsy. He came in and was very informative, explaining that he only had summary reports and not actual tests.

Continued on page 8

Family Stories - Natasha, ...(cont'd)

He examined Natasha and said she looked very healthy, well nourished and not sickly nor failure to thrive. He said she does not need bowel biopsy, nor any tube feeding. Told us that the blood tests that have been done rule out most all diseases of bowels and absorption problems were within normal range (other doctors had not explained that to us).

He would recommend an Upper GI series done to rule out possible malrotation of the intestines, that is the only concern he had in relation to previous bouts of unexplained dehydration, vomiting and diarrhea. He said it is a simple test and relatively non-invasive. If Natasha had a disease of the bowel, she would probably be in poor health and skinny. She doesn't present that way and her blood tests reveal that she is in fine health.

Tuesday we met with an experienced Pediatric Endocrinologist at the hospital. She came in and explained to us that Natasha has had an extremely thorough battery of growth testing via blood tests (we were never informed of this). She also told us Natasha produces totally normal amounts of hormones for good health and growth. Natasha's stature is not related to hormones in her opinion. We asked if they wanted to do stim tests that were recommended by the other facility. The doctor stated "We here do not believe in Stimulation or provocative testing."

"We feel it is dangerous and poses unnecessary risks to our patients." I gather from her comments and that of the nurse that it has been their policy for many years. They have tried to lecture and pass their information on how to obtain enough information through blood tests. Blew us away. But we were so relieved. We didn't feel it was a safe thing for our daughter and that is why we had not jumped at every word or recommendation of the other hospital or specialists.

There are still many questions and the doctor stated that Natasha is a mystery child. But, the main follow up and point of concern is Genetic. Natasha has had a pretty thorough bunch of tests from a genetic point of view, but there are still a few more. The Endocrinologist did say Natasha is still extremely short-statured and feels that she would like to see if a trial of growth hormones may be able to get her a bit taller when she is older, say around 6 or 7 years old, and that she would be happy to assist at that time.

The philosophy at this hospital is to let the Child be involved in making decisions when it comes to issues that are not life threatening. That she is her own person. So far, Natasha really likes herself, understands that she is a little person and is fine with it, and we are so happy for her. So where does this leave things for us? We are not going to pursue the short stature, growth hormone thing unless she asks us to. We have Joined Little People of America, they are a wonderful support network and very strong in advocating for all persons with short stature.

Oh, both the GI and Endo doctor did state that the hospital's approach is interdisciplinary, so if Natasha should become ill again, she would be admitted to the San Mateo County Teaching Hospital and all Depts. would get together and come up with a game plan on her treatment and try to decipher everything together, with family involved every step of the way. They would like to follow up in a year to see where things are at. Of course if Natasha does get ill again and begin to fall apart like she did before, they want us to bring her to the hospital. Or at a minimum have her checked out by a local doctor.

We also got her a new pediatrician. We interviewed a few on the phone and set up an appointment to meet her new doctor. He is a very experienced doctor and his practice is limited to 5 doctors and they are only on call for each other... He seems very caring and concerned. Held a meeting with his partners to have them all up to speed on Natasha's possible condition. Also, he assured us that if she is extremely ill, even if he is not on call, his partners always get a hold of him in difficult cases. So far so good though. Natasha has been very healthy and has not had to go to a doctor for 2 months!

She may or may not have a metabolic condition. If she does, the doctors think it is mild and probably not life threatening. One thing that still concerns me is that all three of the episodes happened during hot weather. Since it has cooled down she has been fine. I worry a bit about summer, but will not dwell. Take each day as it comes.

The differences in her care and the treatment of the whole family differs from doctor to doctor and hospital to hospital. We were the ones who requested a second opinion on everything. Her ex-doctor asked us why we were bothering. Please always know that as parents we have every right to ask for second, third and fourth opinions if we feel they are necessary. Fight for your rights. Our children depend on us.

Thank you all for all your information and support through these last many months. Feel free to ask away if you have any questions.

Ann Leach, Sebastopol, California -- Mom to Natasha. Anacabana@aol.com
Short Stature, undiagnosed and possible metabolic syndrome, undiagnosed



Medical Update

Matter Over Mind: The Realities of a Common Muscle Disease

Georgirene D. Vladutiu, Ph.D.

The adult form of carnitine palmitoyltransferase (CPT) II deficiency has been labeled as the most common lipid myopathy in humans even though its prevalence has not as yet been determined in the general population. This autosomal recessively inherited disease has earned this distinction in part, because the other lipid disorders with muscle symptoms, such as very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency or long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency are very rare. CPT II deficiency may be even more prevalent than generally believed due to under-recognition of the disorder. Before discussing the characteristics of the disease and which individuals may be at risk for having it, it is important to understand what role CPT II and closely related enzymes normally play in lipid metabolism.

CPT exists as two genetically and functionally distinct mitochondrial enzymes (CPT I and CPT II). CPT I is embedded in the outer mitochondrial membrane and CPT II is associated with the inner mitochondrial membrane. They work together with carnitine-acylcarnitine translocase, an inner mitochondrial membrane enzyme, to facilitate the transport of lipids (fatty acids) across these membranes and into the mitochondrial matrix where they ultimately are converted to energy in the form of ATP. There is a liver form and a muscle form of CPT I that are encoded by genes on different chromosomes. There is only one ubiquitous form of CPT II in the body.

Several disorders of varying severity and age of onset have been reported due to defects in either the CPT I or CPT II enzymes. Liver CPT I deficiency is a rare infantile disorder with the hallmark of potentially fatal hypoglycemia and the inability to produce ketone bodies. To date, no cases of muscle CPT I deficiency have been reported and a deficiency of this enzyme may be lethal given the importance of the enzyme for heart function. CPT II deficiency has three distinct clinical forms: the common adult myopathic form, the lethal neonatal form affecting many body systems, and the severe infantile form which has liver, heart and skeletal muscle involvement.

The adult-onset disease is the common lipid myopathy referred to above and is characterized by muscle pain and stiffness and, in severe cases, myoglobinuria (the presence of dark-colored urine caused by the release of myoglobin from damaged muscle tissue) occurring primarily in young adults who are exposed to certain triggers. Individuals with this disorder may be symptom-free until they are exposed to prolonged exercise, fasting, extremes in temperature, viral infection, sleep deprivation or general anesthesia during surgery. Even though the disorder is called an "adult-onset" disease, it has been reported in people of all ages including young children.

Not only does the age of onset vary in CPT II deficiency but also there has been a wide range of symptoms reported among affected individuals. Because some individuals remain asymptomatic until triggered, there are likely to be many people with the disorder who are completely unaware of its existence. The disorder has even been called "benign" in some scientific reports which could not be farther from the truth among individuals who have experienced life-threatening kidney failure following a severe episode of rhabdomyolysis (trigger-induced muscle breakdown). At the other end of the spectrum there are people with mild episodic symptoms who have never had a major attack and wonder if they have a disease at all or if their symptoms are "all in their mind". They may never have gone to a physician for evaluation or if they did, their doctor may not have known enough about the disease to recognize it or how to treat it. Many people have been initially misdiagnosed as having other disorders, such as fibromyalgia or chronic fatigue syndrome, before arriving at the correct diagnosis of CPT II deficiency.

For additional reading about the features of this disease, patient surveys on the struggles of obtaining a diagnosis, common triggers, and effective treatment are regularly presented in a newsletter about CPT II deficiency, entitled *The Spiral Notebook* (Managing Editor, Barbara Seaman). This semi-annual publication also contains feature articles about the experiences of individual patients, scientific updates, nutritional tips, and important website resources. The newsletter may be found on the web at www.spiralnotebook.org.

Continued on page 10

Medical Update (cont'd)

More than 20 different mutations and 3 polymorphisms (so-called harmless changes in the gene's DNA) have been identified in the *CPT2* gene among patients with CPT II deficiency and many other mutations are yet to be identified. One mutation, known as Ser113Leu, accounts for 60% of the mutant alleles (different forms of the gene) responsible for the common adult form of the disease. Different mutations in the *CPT2* gene are believed to account for the varying severities of CPT II deficiency. However, other genetic or environmental factors may also be involved in modifying the expression of the disease because unusual patient situations have been observed. For example, individuals in the same family with the same mutations causing their disease have been known to have varying symptoms (see the Spring 2000 issue of *The Spiral Notebook* for a personal account). Also, individuals known as "manifesting carriers" have been reported to have muscle symptoms although they have only 1 mutation in the *CPT2* gene together with residual CPT II enzyme activity of about 50% of normal in their muscle biopsy. As carriers of a recessively inherited disorder, they should not be expected to have symptoms. In most cases, manifesting carriers either have a second metabolic muscle disease that compounds the *CPT2* gene defect to produce symptoms or they are exposed to severe triggering such as combinations of triggers that may co-exist during rigorous military training or during training for competitive sports.

In the recent past, CPT II deficiency could only be diagnosed by biochemical analysis of a muscle biopsy. While this remains the best specimen for a definitive diagnosis, the enzyme's activity can also be measured in white blood cells and mutation screening can be performed for the common mutations in white blood cells or buccal (cheek) cells, both relatively non-invasive specimens. The problem with mutation screening for virtually any genetic disorder is that one cannot be certain that all mutations have been identified. However, a screen for a panel of 6 known mutations in the *CPT2* gene rules out over 90% of causative mutations.

The treatment of CPT II deficiency is variable and often patient-specific considering the wide variation in symptoms and lifestyles. There are certain things affected individuals can do to prevent symptoms. Most try to avoid the triggers such as prolonged vigorous exercise, fasting, and extremes in temperature. If they need to have surgery with general anesthesia, they alert their physician to the need for alternative anesthetics that do not trigger symptoms, plus most people with CPT II deficiency wear Medic Alert tags describing their condition and the associated risks. Individuals with CPT II deficiency try to keep their water intake high, especially if they are athletes, and keep sources of simple carbohydrates handy such as Gatorade and Powerade. However, there are specific treatment regimens for CPT II-deficient patients, including the administration of MCT Oil (medium-chain triglycerides) or carnitine that should be established on a case by case basis under the supervision of a metabolic physician and nutritionist. Patients should not try to treat themselves without the involvement of these professionals because they may have adverse or suboptimal effects from certain treatments.

In spite of the fact that there is no cure for CPT II deficiency, it is a manageable disease in most cases. It is important that primary care physicians are informed about this disorder and its manifestations so that they can recognize the symptoms, provide genetic testing for a diagnosis and ameliorative treatment. Frequently patients who remain undiagnosed wonder if their symptoms are imagined or exaggerated because of the chronic nature of the disease. We have documented numerous cases of individuals who finally received a confirmative diagnosis after years of searching, and sometimes after many fruitless muscle biopsies.

CPT II deficiency is not a matter of mind, it is a real disease with real symptoms. We received very positive feedback recently from the wife of a 42-year old patient who was diagnosed by enzyme and mutation analysis in blood. She claimed that her husband was a "whole new man" after finally having a name for his condition. For 25 years he had thought his pain, cramps, and stiffness with exercise, as well as chronic bouts of myoglobinuria, represented an undefined disorder that he longed to understand. At the age of 20, he underwent a muscle biopsy that was considered to be normal but it was never tested for CPT II deficiency. After many years, he noticed on his own that he could decrease the risk of cramping by eating prior to activity or shorten the time to recovery by drinking carbohydrate-containing beverages after physical activity. Once he had his diagnosis, he knew that his symptoms were characteristic of a specific disorder and a new life began where his condition would no longer be trivialized as a case of mind over matter. Now he and his physicians were able to take control over his symptoms, improve his quality of life and that of his family and, perhaps most importantly for his peace of mind, he had a diagno *Continued on page 11*

Medical Update (cont'd)

References

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Thuillier L. Carnitine palmitoyltransferase deficiencies. *Molec Genet Metab* **68**:424-440, 1999
Smail D, Gambino L, Boles C, Vladutiu GD: Rapid, cost-effective gene mutation screening for carnitine palmitoyltransferase II deficiency using whole blood on filter paper, *Clin Chem* 1999; 45:2035-2038

Dr. Vladutiu is an Associate Professor of Pediatrics, Neurology, and Pathology at the State University at Buffalo School of Medicine and Biomedical Sciences and is also the Director of The Robert Guthrie Biochemical Genetics Laboratory at Children's Hospital of Buffalo. The laboratory performs over 3,000 esoteric diagnostic tests annually for inborn errors of metabolism with a particular emphasis on the metabolic myopathies and mitochondrial disease. Dr. Vladutiu is the principal investigator of a research grant from the Muscular Dystrophy Association awarded to improve the diagnosis of carnitine palmitoyltransferase II deficiency disorders. While seeking to understand the genotype (gene mutations) and phenotype (clinical symptoms) correlations in this disease, her laboratory performs both biochemical and molecular diagnostic testing for this and other relatively common disorders of exercise intolerance. (Contact Dr. Vladutiu at mitomaven@aol.com or 716-878-7513)



Family Stories - Melissa, MCAD

I've been thinking of contributing to the newsletter for years, so I guess maybe its time I shared our experiences with everyone. We've been getting the newsletter since its beginning. I think a story like ours is worth telling, because it is mostly a Happy Story. Thought you might like to hear from someone in Canada.

A HAPPY STORY

When our daughter Melissa was about 4 years old, she became very ill suddenly. We had no idea why she was so ill, but she was not responding to us very well, and just laid in bed, lifeless. Brenda took her to our family physician, and he became concerned immediately, and sent us up to see a pediatrician in Edmonton. By the time Brenda came to get me from work (on the way to the doctors), Melissa was having seizures. We raced into the doctor's office and he dropped everything to look at Melissa. He took one look at her, and told us to take her to emergency. Luckily the hospital was next door. The doctor dropped his patients, and left to come to the hospital and met us there within a few minutes. By this time Melissa was having more severe seizures. There were nurses and doctors running everywhere, and we just stood there helplessly, watching. At one point Melissa stopped breathing and they had to resuscitate her. I don't think it hit us until the hospital priest came and asked us if we would like to go to a private room. I don't think Brenda and I have ever been so scared in all of our lives.

Luckily for us, and Melissa, the doctor was smart enough to check her sugar levels. They found out very quickly that her sugar level had bottomed out! They then gave her glucose IV, and she responded well to that. They put a tube down her throat and told us they would have to send her to the University of Alberta Hospital, as they were not equipped to handle this. They sent her by ambulance to the other hospital, and we could not even go with her as they had no room, with all the doctors and nurses. We went over to the other hospital, and when we got there, she was already in pediatrics ICU. She was alive, and resting better.

Continued on page 12

Family Stories - Melissa, MCAD (cont'd)

That's when we met Dr. Ferreira from the genetics clinic at the U of A. He told us that Melissa's ammonia level was high, and they suspected Reye's syndrome, which he told us was serious. Eventually, they ruled that out. Then they suspected fructose intolerance, because she had had some candy floss they day before. Eventually, they ruled that out. Melissa spent a week in ICU and a week on the pediatric ward. She was doing much better now, but we still had no answer about what was wrong.

A while later, Dr. Ferreira called us to say he found out the problem, and he wanted us to come in so he could explain it to us. He said she had carnitine deficiency, and needed to take L-carnitine orally for the rest of her life. Compared to some of the other things she could have had, we were relieved somewhat. A couple of weeks later, the Dr. called us back to say that he sent a blood sample to Dr. Charles Roe, at the Duke University in North Carolina [now at Baylor in Dallas] and he diagnosed her with MCAD. The treatment would be the same, L-carnitine orally, with a low fat, high carbohydrate diet. We were lucky here in Canada, as we have the orphan drug program, and Melissa's medicine was paid for by the government. Melissa seemed to be doing well, neither of our older two kids had any signs of it. I talked on the phone one day to Dr. Roe, and he asked me how many kids we had lost over the years. I told him none, and he said we were very, very lucky. As Melissa got older, she seemed to tire out very easily, especially if she missed a dose of carnitine. Dr. Ferreira increased her doses a couple of times as she got older, and that seemed to help. Melissa is now 15 years old, and as healthy as any teenager I know.

She is responsible for taking her own carnitine, but seems to forget it more often than not. She has gone for long periods of time without taking carnitine, and seems to be fine. In summary, we feel very blessed to have survived a near tragedy, and it breaks my heart every time I read about people not as lucky as us in the newsletter. Most of the time now, we don't even think about Melissa having MCAD, as it doesn't even seem like she has anything wrong with her. She is just a normal teenager growing up. I wanted to write and share this experience with other people out there, as it is truly a story with a happy ending.

Regards,

Perry and Brenda Sampson
Alberta, Canada
p.sampson@home.com

Riboflavin Tip from Gay Grossman (sjg.egg@mindspring.com) – Having difficulty with your child taking riboflavin? Try using 1 tsp of karosyrup, open the riboflavin powder capsules, mix together, then add 1 tsp chocolate syrup. The karosyrup is no fat or protein, as well as the chocolate (Hershey's lite). Can take it like that or also add a scoop of rice cream, an ice cream but from rice, so it's non-dairy and tastes pretty good!



*"All life is change,
Growth is optional"*
~ Unknown



Family Stories - Kristopher, undiagnosed FOD

It was Mother's Day -- what a day to find out that you are pregnant with your first child. I was excited and so was my boyfriend. We both seemed a little apprehensive since we both had college to deal with. I went to the doctor to confirm that I was pregnant and I sure was!!!! Since I am diabetic we were under the best care we could be. For 9 months we went through ultrasound after ultrasound and test after test. By our 7th month the Doctors assured us that everything was going well.

Then on December 21, 1997 after practicing our lamaze we settled down to get a good night sleep since we were going to the doctors the next day to see if it was time to be induced. We did not get much sleep because no sooner did I lie down that my water broke. We got to the hospital around 11:30pm. By 7:00am I still was not having contractions strong enough to move the baby. Everyone decided that it was time for PIT and after a long labor, at 7:01pm on the 22nd of December our beautiful baby boy made his entrance into the world.

He had two nines on his apgar test and he seemed to be fine. Our son roomed in with us. I was breast feeding and wanted to spend every minute with our blond haired angel. He had a little trouble at first with the sucking reflex. He seemed to have no interest in it at all. I questioned the nurse and she said that some babies just had trouble and to give him a couple of days and he would come around. We had a breastfeeding counselor come talk with us and she told us not to give up.

The three of us were discharged on Christmas Eve day. On the way home while Kristopher Howard was sleeping peacefully in the back seat the song "How could I live without you," played on the radio. Little did I know that song would have so much meaning. We had one of the best Christmas' ever.

Christmas morning we noticed that our son was very yellow so we called his doctor and took him in the next day. The doctor looked at him and told us he was fine and to put him in a onesie in front of a window in the sun. The next 2 days went as normal. He still would not eat and almost seemed colicky at times.

On the night of the 28th of December we all went to bed as normal, except we put Kristopher between us. We had never done that before. We had just gotten a puppy that morning and he was jumping at my side of the bed and crying. I rolled over to have Howie take care of the puppy. For some reason I looked at the clock and it said 4:30 am. It seemed strange to me that Kristopher had not yet awoken to eat. I turned on the light and my son was lying on his side. I put my hand on him and said "Kristopher, Kristopher." He did not respond to me so I put my finger in his and he didn't squeeze. I remember saying "Oh my god something is wrong."

I picked up my son and his head dropped back, his pacifier fell out of his mouth and he wasn't breathing. I gave him two breaths and I choked. I just started yelling "Mom, Mom Kristopher isn't breathing, Help me." She took Kris and I called 911. I can still remember the conversation. I looked up and my boyfriend was standing in the door with a look of desperation and disbelief on his face. Before I could hang up, the police were at our home. They began to breathe for my son. I stood by the door yelling "Kristopher, Kristopher, come on baby breathe."

We got to the hospital shortly after the ambulance. They put us in a small waiting room. A nurse came in and gave me Kristopher's onesie he was wearing. I knew then it was bad. Then not more than 5 minutes passed when a doctor came in. I knew by the look on his face it was bad. He said "I am so sorry. We did all we could but your son has passed on." Then someone asked me if we wanted our son baptized and we immediately said yes. Then a man came in and asked about an autopsy. We wanted to know WHY so we said yes. Then I had to do the hardest thing in the world and call my mom. I remember saying "Mom, Kristopher is dead."

Howie went out and called his mom. I heard him tell her and I broke down a little. The nurses asked us if we wanted to go to Kristopher or if we wanted them to bring him to us. We asked for them to bring him to us. I was in such shock that I was afraid that I couldn't walk. When they brought him in, I could not hold him. Howie held him and I kissed his little lifeless head and said "I love you, I am so sorry."

Within minutes Howie's entire family and my mom and dad were in the room with us and we were all holding each other up. Two days later I found myself in the funeral home picking out caskets, flowers, and writing obituaries for a person who never had a chance to live. At the funeral and the wake we had the support of Howie's family and it felt good since my family other than my mom, dad, brother and grandmother, didn't even call.

Continued on page 14

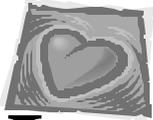
Family Stories - Kristopher, ...(cont'd)

A few days after the funeral I got a call from my Medical Examiner and he asked about medications, since he had fat deposits in his liver. About 4 weeks passed when our pediatrician called and said that it was an undetermined FOD. He told me he didn't know much about it but he thought that I would not be able to have any more kids. That is when we got in touch with Dr. Rinaldo. He assured us that we could have more children, but the guilt set in that we caused a disease that killed our son. After talking with Dr. Rinaldo we felt much better.

We have a 1 year old son now who is not directly affected by an FOD, but since we are not entirely sure which disease it was he still could be a carrier. For both our sons' sake we will never give up looking for the exact cause.

I thought my life was over at 19 when I lost my first son. Boy was I wrong! I have a chance with my soon-to-be husband and our baby. When I get down about Kristopher, I just think, **as long as you remember and hold them in your heart they are never really gone. Every day I think of Kristopher and thank him for all he continues to give us in our lives.**

Sandra Laviana and Howard Aitken
Canton, CT
Triggerhappy1222@aol.com



Love Messages

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

Joan and Tim

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

Sandy and Howie

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

Jeanne and Mark

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

Jodi and Wayne

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

Baby Barnes - Death in-utero Oct 7, 1999

Delane and Althea

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

Sue and Jim

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

Jacque and Mike

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

Joseph and Barbara

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

Barry and Julie

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

Steve and Liz

Caitlin - Birth July 10, 1989 Death May 10, 1996

Carolien

Liese - March 2, 1988 Death Aug 25, 1990

Tom and Lynn

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

Continued on page 15

Mark and Karen

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

Jenny and John

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

Mark and Diane

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

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

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

Valerie & Chris

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

Toni and Mark

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

Martin and Kathy

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

Sandy and Jon

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

David and Amy

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

Doug and June

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

Andrea and Phillip

Brandi - Birth Dec 2, 1986 Death Jan 1988

Lance and Dawn

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

Deb and Dan

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

Jeannette

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

Michael and Nicole

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

Ralph and Angie

Chelsea - Birth Jan 11, 1995 Death Apr 3, 1986

Nikki and Toby

Reece - Birth Aug 1998 Death April 18, 1999

Brad and Kim

Brittany - Birth March 20, 1991 Death Feb 21, 1997

Debbie and Dave

Lauren - Birth May 4, 1988 Death Dec 15, 1989

Robert and Dixie

Cody - Birth July 30, 1987 Death Dec 26, 1992

Meredith and Neil

Claire - Birth Sept 1, 1986 Death June 23, 1997

Brian and Kim

Patrick - Birth July 15, 1996 Death July 28, 1997

Vickie and Burnell

Paul - Birth Mar 31, 1993 Death Sept 20, 1993

Annie - Birth Nov 26, 1998 Death April 22, 1999

Diane and Mickey

Marie - Birth Dec 1, 1989 Death Oct 5, 1991

Andy and Temple

Nancey - Birth Feb 8, 1989 Death July 20, 1990

Robert

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

Jamie and Tom

James - Birth Dec 8, 1996 Death Aug 13, 1997

Lisa and Pete

Devin - Birth July 18, 1997 - Death July 19, 1997

Mary

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

Darlene and Larry

Marissa - Death Feb, 1999

Heather and Phillip

Toni Marie - Birth Oct 8, 1990 Death March 22, 1991

Ron and Paula

Daniel - Birth May 19, 1981 Death Jan 12, 1982

Randy and Misty

Jeremiah - Birth April 3, 1991 Death Dec 15, 1991

Christine and Mark

Erin - Birth Aug 26, 1988 Death Jan 21, 1989

Linelle and Matt

Cole - Birth Mar 21, 1999 Death Oct 18, 1999

Lori and Jeff

Jordan - Birth Feb 19, 1997 Death March 21, 1998

Simone and Michael

Michael Dylan - Birth Aug 24, 1991 Death Aug 24, 1991

Continued on page 16

Robert and Dixie

Cody - Birth July 30, 1987 Death Dec 26, 1992

Mike and Sheryl

Justin - Birth April 22, 1990 Death April 22, 1990
Nathaniel - Birth Aug 15, 1991 Death Aug 18, 1991

Verna

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

Albert and Arleen

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

Jennifer and Jason

Alexander - Birth June 1, 1995 Death June 3, 1995

John and Sally

Zachary - Birth March 24, 1997 Death March 27, 1997

Tanya and Pat

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

Brian and Cherryll

Kylie Ann - Birth Feb 7, 1990 Death Feb 11, 1990

Litzy and Jesus

Jesus - Birth Sept, 14, 1996 Death March 16, 1998

Jackie

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

Janna

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

Anne and Gary

Sydney - Birth Oct 13, 1995 Death May 20, 1996

Rick and Stephanie

Trina - Birth July 1977 Death Jan 14, 1978

S. Elizabeth & G. Douglas

Philip - Birth April 6, 1994 Death April 8, 1994

Darren and Karen

James - Birth Jan 15, 1996 Death Aug 31, 1996

Richard and Amy

Andrew - Birth May 1978 Death Nov 18, 1979
Scott - Birth May 1983 Death April 25, 1985

Denise and James

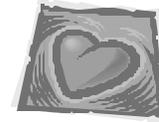
Benjamin - Birth March 11, 1987 Death Dec 20, 1988
Beau - Birth July 10, 1991 Death Feb 19, 1995

Mike and Darci

Brett - Born June 14, 1993 Death June 17, 1993

Lori and Dean

Brennan - Birth June 1, 1999 Death June 6, 1999



*"All that we love deeply becomes
a part of us"*

~ Helen Keller

**Visit our NEW Graphic
Website...**

*The last several months, **Mary Lingle** (MCAD parent) has worked very hard to not only update our site with new information, but with her design expertise she has added a dimension of warmth and comfort. So please visit **www.fodsupport.org** to see OUR new look!*

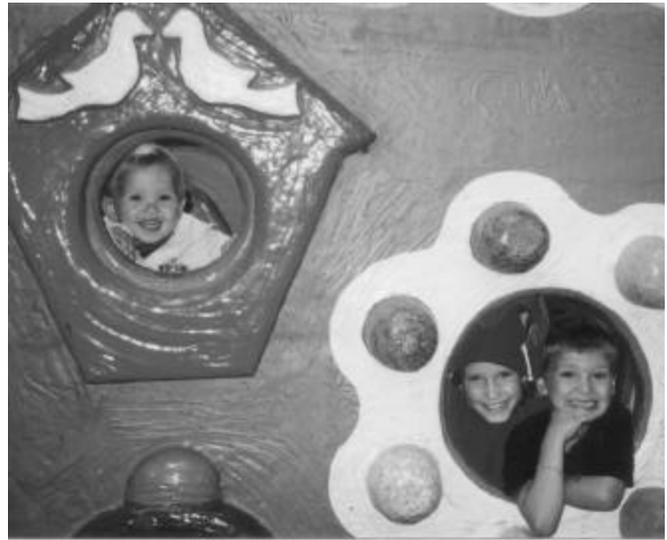




Kids Korner



Jane (LCHAD) & Megan Carroll



Brett (VLCAD), Morgan & Jake



Rosemary & Jennifer (MCAD)

MCAD Teens – We are trying to **determine if there is an interest** among any of the Teen MCADers for being involved **in a research follow-up study** by an FOD researcher. Please email Deb (deb@fodsupport.org) if you are interested

New FOD Pen Pal Program - See enclosed Insert



FYI: Insurance Tips

My name is **Marcia Dunahoo** and I am the grandmother of Josias Wilson, who was diagnosed with GA1 in June 1999. Deb saw my email responses on the Organic Acidemia Email List and asked me if I would allow her to use some insurance information that I had conveyed over the List. I told her definitely yes, and that **if anyone had questions concerning their health coverage that I'd be glad to help**. I have been in the Insurance business for over 20 years; working with various groups such as BC/BS of TX, Drs' offices, and presently with a large insurance carrier in the US and Canada in Group Benefits. Please **email me at M2WD@aol.com** if you have any questions or comments about the following "Tips." Love and light to all!

When charges are denied as 'unreasonable' it is usually the charge they are denying, not the treatment. To determine reasonable and customary charges they consider the charges of the doctors (by specialty) in your area and average the charges. That is then considered the "reasonable and customary charge." It is called R&C or URC charge. This method of payment was developed many years ago as part of the first 'managed care' implementation. If the charges are over R&C for your area I would suggest talking to the billing department for your doctor. They may not be billing your insurance company correctly. If they will not help, talk with the doctor. He/She is usually the one that sets charges for his/her services.

Find out if your insurance plan is ASO (Administrative Service Only) or Fully Insured. If it is an ASO, it is self-funded, which means the company you work for has determined the benefits for their employees. Talk with your Human Resources Director. Since the things your children need are not common, they may not have been aware these services were needed by anyone anywhere and may be able to revise their plan to cover these items. Also find out if the plan complies with State mandates (laws) and extraterritorial mandates. ASO plans do not have to comply, though some do. The reasons for the 'extraterritorial' statutes is that if you reside in a state that has mandated certain benefits the 'extraterritorial' laws apply even if the policy was written in a different state.

If it is a Fully Insured plan (which is very rare these days), they DO have to comply to State mandates. These plans follow the guidelines of the insurance company that insures them.

Appeal the denial of any claim that has been denied as soon as you receive the denial. Do this in writing and on the phone (call your insurance carrier customer service line). We are all human and sometimes the claim processor makes errors.

When open enrollment times comes for your company, get out of the HMO. If your company offers a PPO (Preferred Provider Plan), Open Access (expanded PPO plan) go for it. It may cost you a little more out of your paycheck every month, but is well worth the difference. With these plans you can choose your doctor (as long as he/she participates in the plan) and they are contracted with an outside vendor to provide discounts to your insurance carrier and ultimately to you.



Resources

Recipe Booklet for MCT Oil/Portagen users Mead Johnson 1-800-247-7893

LCHAD/VLCAD Support in UK www.lchad.freeserve.co.uk

SNAP Information for Families with Special Needs Children www.snapinfo.org

National Patient Travel Center www.patienttravel.org 1-800-296-1217

Books to "check out":

Living with a brother or sister with special needs by Don Meyer & Patricia Vadasy

Introduction to nutrition & metabolism by David A. Bender



September is Newborn Screening Awareness Month
'Saving Babies One Foot at a Time'
 (See below and enclosed insert in this issue)



Newborn Screening Advocacy Update

I'd like to thank everybody who responded and acted since last issue's article, "Things you CAN do to advocate newborn screening." Most unborn children are still at risk, so please keep working on it to the extent you're able!! We've done well to raise awareness. Since the last issue:

- * about 30 articles and TV broadcasts have been done across the country on newborn screening,
- * pamphlets have been passed out
- * Someone from OAA began a newborn screening pin campaign, similar to breast cancer awareness pins.

* **Tyler For Life Foundation, a new Newborn Screening Awareness organization** has been active nationally as far as promoting NBS. I am on their Board, as Director of the Tandem Mass Spectrometry Division. My job, with the help of MANY others, is to fix the problem of 3.7 million babies each year in the US alone NOT getting newborn screening with this technology. We plan to go international in a few years. Tragically, 2700 kids each year in the US alone are dead or brain damaged because no one knew they should have a \$20 newborn screening test. Please contact Tyler For Life online or offline.

We can't do it alone! The world has GOT to know NBS for many of these disorders EXISTS!

As always, call me any time to discuss newborn screening. I am desperate to make a difference and no one can do it alone.

Wendy Nawn, MCAD Mom
 Malvern, PA
 (610) 251-9876
 wendy@savebabies.org

Tera Mize, Tyler For Life Foundation, President
 6340 Holborne Ln
 Douglasville, GA 30134-4023
 770-947-3638
 www.savebabies.org

NEWBORN SCREENING PIN FUNDRAISER

We, as individual members of the FOD Family Support Group, have the opportunity to assist the Tyler For Life Foundation (www.tylerforlife.com) with the promotion of supplemental Newborn Screening which is their MAIN Mission. A number of nonprofit groups (of which TFL is one) representing many of the detectable metabolic disorders have **joined forces to heighten public awareness of and to raise money** for obtaining Newborn Screening for every baby born in the US.

The **Newborn Screening Pin is the first collaborative project.** These circular lapel pins come attached to a card that dramatically describes the cause and lists all the groups involved in the project. The groups are declaring September to be Newborn Screening Month and asking everyone to wear these pins for the entire month. **PLEASE read the enclosed insert and HELP US SAVE LIVES!!!**



Family & Professional Donations

The FOD Family Support Group would like to thank recent contributors: Renee & Randy Palmer in honor of Justin; Wendy & Chris Nawn in honor of Alex (MCAD); Katrin & Daniel Halbenleib in honor of Andreas (MCAD); Shannon & Barrett Breuckman in honor of Reece (MCAD); Suzanne & Darren Lowe in honor of Elizabeth (GA2); Angela & Jim May in honor of Joshua (MCAD); Linda & Danny Farmer in honor of Anna (CPT2); Glenda Romany in honor of grandson Devan (MCAD); Kerry & Brian Tobin in honor of Hailey (Undiag); and Marianne Genetti.

We greatly appreciate donations to help with postage and copying fees. **Checks can be made payable to DEB LEE GOULD.** Please note on the check that it is for the FOD Family Support Group. Because we are not officially a non-profit organization, donations are not tax deductible at this time.



Thank You

Thank you to Erika Wallace (Mailing Lists), Mary Lingle - Mcartwrite@aol.com (Web Page) and Brian Gould - BulaBri2000@hotmail.com (newsletter) for all your hard work. Special thanks to Sigma-Tau Pharmaceuticals, Inc. for their continued financial support.

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



Communicate with us

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

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

Please include ideas for future issues or your questions

A Big Thank You

Because of her HUGE heart,
Shelli Craig
volunteered to format our
Newsletter for several issues.
THANK YOU Shelli for your
expertise and commitment to
helping our Family Support
Group!!!



Reminders

FAMILIES - Please send **TYPED** stories by **December 1, 2000**. 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 **December 1, 2000**.