



fatty oxidation disorder
communication network

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From the Editor

We hope all of you had a very safe and HEALTHY holiday season. Time has gone by so fast ~ seems like yesterday that everyone was concerned about Y2K! Dan, the boys, and I were thinking about ALL of our Families, especially those experiencing their first winter and holiday season without their child ~ our prayers are always with you...

As with every issue, **getting the word out about FODs is VITAL and this May 4 and 5, 2001, families and professionals will have an opportunity** to hear more about FOD diagnosis, supplemental NBS, treatment, research, nutritional information and much more. The National Coalition for PKU and Allied Disorders has extended an invitation to our Group, as well as several other Groups, to **attend a National Metabolic Conference in Dublin, Ohio (see pg. 17 & Registration form)**. That Friday each Support Group will hold its own breakout meetings and then on Saturday everyone will come together and hear internationally known and respected speakers on various topics concerning all of our disorders.

Trish Mullaley, President of the National Coalition, has provided more information at www.pku-allieddisorders.org/upcomingevts.htm so please visit her site for updated speaker and registration information.

Another **IMPORTANT** notice that affects all of our members ~ **because of increasing copying and postage costs, we may be discontinuing the printed Newsletter and posting it ONLY on the website.** We realize, for some, it may be difficult to read our online issues but HOPEFULLY families, friends, and professionals will be able to access it directly though home, work, hospital, or library computers. **This issue or the July issue may be the final printed issue mailed to you.** However, we will **continue to send New Family Packets to all of our newly diagnosed Families.** Even though we now have **all the past newsletters in our Archive section of the website (link is on our homepage)**, it's always nice to have all the information right in front of you, especially when you just get a diagnosis and feel so overwhelmed. Discontinuing our printed newsletter may be disappointing for many, but we feel that the high costs and increasing 'membership' makes it difficult to continue. We HOPE you understand.

IF you feel that you will not be able to access a computer, contact me directly (mail or phone) and tell me that you want to continue to receive a printed newsletter. IF we have the funds to do so we will try our best to get you a printed issue.

This issue is filled with a variety of **Family Stories**. Thank you to all the parents that shared your sorrows, challenges and joys. **Keep your stories coming!** Also be sure to **send our webmaster and MCAD Parent, Mary Lingle (mcartwrite@aol.com) your poems, artwork, pictures etc., for our 'Love Messages' page on our site.**

Professionals ~ we NEED YOUR HELP! PLEASE let us know about your research, publications, and clinical work with FOD Families so we can get the much needed information on our website for all to read and learn from.

We learn from **EACH OTHER** so take your time and read all that we have in this issue and on our website and then **SHARE your knowledge.** And if you'd like to learn and talk with other FOD Families and Professionals, please be sure to **REGISTER for the National Metabolic Conference by April 15!** Even though all of us may not be able to attend, always remember that...

'We Are All in This Together!'

Take care... DLG 

IN THIS ISSUE

From the Editors...1 Letters to the Editor...2 Family Stories...4 Medical Update...8

Love Messages...15 Kids Korner...18 Notes/Resources...19 Reminders...20



Letters to the Editor

Dear Deb: Just a reminder that newborn screening is not alcoholism. In other words, newborn screening is not an issue where the more you're exposed to the bad stuff, the more of a "tolerance" you have for it. Families know what I'm talking about.

The American Academy of Pediatrics recently came forward and publicly declared that all the states should screen for the same things. Then the March of Dimes criticized this position, saying that's not specific enough. March of Dimes wants all the states to screen newborns ~ for only EIGHT disorders. Not the 30+ or 40+ disorders that are already being done in some parts of the world; rather, EIGHT disorders. **FODs are not included in March of Dimes' recommended eight disorders.**

When I jumped on them about this, March of Dimes told me their "eight" is only a start. Well that's nice but they are not utilizing urgency. Newborn screening is not alcoholism. **It is not OK to ignore known information and tolerate the bad stuff (death, brain damage, near death that we all KNOW will happen), which is exactly what so many people in power are doing.**

Wendy Nawn, MCAD Mom
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Vice-President, Tyler for Life Foundation



Dear Deb: I have felt for quite some time that I should share our story with other parents but after receiving the **Reye's Syndrome Survey** this week, I knew it was time for me to write.

On July 14, 1979 we had just moved into our new home. Our 3-year-old son, T.J. was playing and 'helping' us with our unpacking. His one-year-old brother, Timmy, slept unusually late that morning and didn't want much of his breakfast. I recalled that he had been quite irritable the evening before, and had refused most of his dinner. We spent most of the morning unpacking and had planned to go to my husband Terry's afternoon softball game. As game time approached I decided to stay home with the boys. Timmy seemed so sleepy and by noon, I gave in and put him down for an early nap. He slept all afternoon and by 4 o'clock, I was getting a little worried. I went in again to check on him and he was still sleeping peacefully. I touched his little face and his skin felt cool and clammy. I scooped my little boy up in my arms and walked into the living room to share my concern with my mother-in-law who had come from Michigan to help us with our move. She looked at Timmy and said, "If I didn't know better, I'd think he was dead!" I couldn't believe she'd say such a thing, but he DID appear unusually lethargic.

I called my sister who came right over and we took Timmy to the hospital. My sister-in-law rushed to the ballpark where the coach called Terry off the field. When I carried Timmy into the hospital's emergency room a nurse grabbed my baby out of my arms and asked, "Is he always THIS color?" I hadn't noticed how gray he'd gotten in the 15 minutes it took us to drive to the hospital. Seconds later a voice on the PA system yelled, "Respiratory arrest ER, respiratory arrest ER!" My legs felt like jello as I tried to move out of the way of the team of doctors and nurses as they worked on my little boy. I walked out into the corridor and into the arms of my parents. By now my husband had arrived at the hospital and we were all taken to the chaplain's room. I asked if my baby was dead and the chaplain said, "Not yet." My Dad and our Pastor knelt to pray with us and I begged God to help our son. Soon the doctor came in and told us that Timmy was breathing on his own, but that no one really understood what was happening. The only things being done were IV fluids and lots of lab work. Timmy's ammonia level was very high and his glucose was 15. He was almost comatose the entire night and part of the next day. Terry and I were being questioned regarding any poisons, etc. that Timmy could possibly have swallowed. I guess that's pretty much the routine when unexplained things happen to children.

The next day **we were told that Timmy had an "aborted Reye's Syndrome"** and that is was a miracle that he was alive. Thank God! Five days later we left the hospital with our very active one-year-old son. At age four, Timmy had a tonsillectomy. He spent only one night in the hospital, but was re-admitted for five days because I was having difficulty waking him to give him the popsicles, etc., that he was supposed to eat. He was re-hydrated and sent home.

Our third child, Jill, was born on October 24, 1980. She had febrile seizures at 10 months of age, but otherwise only the normal childhood viruses and infections. Her fifth year of life was very hard for all of us. Jill would get the 'flu' and end up dehydrated and in the hospital with IV fluids for 3 days. Twice she was semi-comatose and the nurses worked very hard trying to get her to respond. Finally during the fifth hospital stay in five months, my husband sat down with our pediatrician and expressed our concerns. **We felt like something was very wrong. Other children got sick and were well within a day or two. Jill got sick and was hospitalized each time.** She missed a lot of her kindergarten classes.

Continued on page 3

Letters to the Editor (cont'd)

Our pediatrician referred us to University of Wisconsin in Madison. We spoke with them on the phone and they asked us to come there on October 23, 1986. Jill was fed the normal breakfast, lunch, and dinner on the first day. She was not allowed to eat after 6 p. m. A needle was inserted into her arm just in case an IV would be necessary. By 9 a.m. the following morning Jill was complaining of a tummy ache and didn't seem to mind that she was not getting breakfast. By noon she began vomiting and appeared very sick after having fasted for only 15 hours. This was her 6th birthday and it was sad to see her so miserable, but the IV fluids were started and by the next day she was feeling much better and acted more like herself.

Terry and I were asked tons of questions about our other children. We told the doctors that both boys were very healthy and **the only real illness either of the boys had was Timmy's "Reye's Syndrome."** The doctors got very concerned looks on their faces and they nodded as they looked at each other. **They explained MCAD to us and asked if they could test Timmy, who was then 8-years-old. Sure enough, Timmy tested positive for MCAD.** T.J. does not have MCAD, but still may be a carrier.

Presently, Jill is almost 20-years-old. A few years ago she lost 15 pounds in 10 days and we were told that her fats had become toxic. She watches her fat intake more carefully and maintains a weight of 115. Tim is 22-years-old and spent the night in the ER last month after a 3-hour episode of vomiting and diarrhea. He became very weak and his ammonia level was slightly elevated. He felt much better after a few hours of IV fluids. This was his first episode in several years.

Last week Tim became Daddy to a beautiful baby girl, Laci Renee. When her blood was drawn for the PKU testing an extra sample was sent to be tested for MCAD. This testing is not routine at our hospital and required a special order from her pediatrician. We are still awaiting those results. We hope our experience helps others who have children with MCAD. We are very grateful for the doctors who took care of our children. Now they are too old to see a pediatrician and it has been a little harder finding doctors who are willing to take the time to study up on MCAD.

Thank you for the wonderful support we receive through the FOD newsletter. **Our prayer is that someday EVERY newborn will have the opportunity to be tested for these rare diseases. We're all in this together!**

Jody and Terry Matthews
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SIDS/RS Survey (for 11 respondents)

(This is an ongoing Survey ~ see www.fodsupport.org 'Online Forms')

Question #1: How many of your children died?

6 Deaths (initially called ~ 2 SIDS, 3 RS, 1 pneumonia)

Question #2: How many had Reye/SIDS-like episode but no death?

5 had Reye-like/SIDS episode, no death (1 called near SIDS, 3 RS, 1 meningitis)

Question #3: How old was child at death or episode?

Deaths: 5mos, 11mos, 13mos, 19 mos, 21 mos, 2 years
Episode: 5mos, 8mos, 12 mos, 13mos, 4 years

Question #4: Were metabolic disorders discussed as a cause?

Yes: 2
No: 8

Question #5: Were you discouraged in pursuing metabolic testing?

Yes: 5
No: 6

Question #6: What was their reasoning?

- To avoid grief, further worry, just accept
- felt it wasn't possible because age of onset was 4 years
- "There is no answer - consider yourselves lucky she didn't die" (!!!)
- "There is nothing that can test to prevent SIDS"
- "It HAS to be RS, it's not this MCAD" (!!!)

Question #7: Did you insist on further testing?

Yes: 4
No: 2
No answer: 5

Question #8: How long between death/episode and CORRECT diagnosis?

4 days, 2 weeks, 3 weeks, 3½mos, 8mos, 9mos, 1 year, 1 year, 2 years, 4 years, 7 years

Question #9: Specific FOD/OA CORRECTLY diagnosed ~

6 MCAD
3 LCHAD
1 Unclassified FOD
1 Propionic Acidemia (Organic Acidemia)

Question #10: State of death/episode?

NH, AZ, GA, WA, 2 MI, MO, 2 IL, CT, and 1 unanswered

[PLEASE consider answering our Survey if you've had a misdiagnosis of SIDS or RS. It is IMPORTANT for Professionals to be aware of various Metabolic Disorders that may 'present' like SIDS/RS. Finding the CAUSE of a death or episode, and not just describing the 'symptoms,' is vital in helping families deal with their grief and/or their specific metabolic disorder.]



Family Stories - Andreas, Swiss MCAD

In March 1986, after 3 months of pregnancy, I lost our first baby. An old lady in the hospital said to me, "Same month next year you will be here with your newborn baby." And so it was! Andreas was born March 1987 in the same hospital.

My pregnancy passed without problems. After his birth, he was jaundiced and had to have a lamp-treatment, but for 11 months we had the healthiest baby the Doctors had ever seen. I breastfed him for 11 months (at the end only in the morning). He slept through the nights from the first day at home and caused not one problem. Then one morning he was vomiting and had diarrhea. The Dr told us to give him tea, orangejuice, salt and dry bread.

In the evening he was quite happy and stopped vomiting. The next morning he did not move anymore and from time to time he was very stiff and we could not wake him up. As we used to live in a small mountain village it was quite difficult to reach the Dr. He was at the local hospital at this moment. His wife tried to call him.

Lucky enough he asked for the helicopter to transport Andreas to Bern (Swiss capital city) at Children's University Hospital "Insel." They first made a puncture of the spinal marrow because they thought it was meningitis. He was very dehydrated and got an "infusion" (liquid IV). For me it was a terrible moment when the pilot told me that I could not fly with them, because there was no seat for me.

When my husband Daniel and I arrived in Bern a few hours later they did not give us much hope. Andreas' liver was at double size and excreted ammonia. They told us, that ammonia affects the brain and that he did not have much of a chance to survive, but if he did, he would be seriously brain damaged. They let us visit him at the intensive care station. He laid on a bed with lots of wires and tubes and was connected to machines. He looked as if he had a lot of pain.

The next day **they diagnosed Reye's Syndrome**. We were still without any hope, as Reye's Syndrome is deadly. My husband had to go back home because we had to run a small hotel and restaurant. I stayed with Andreas the same hours he used to be awake before his coma and left the hospital to get fresh air during the hours he slept. Fortunately, my sister lives next to the hospital. I passed a lot of time with her, talking and even laughing. She was a great support and helped me to turn away the terrible thoughts for a few moments to give me a rest. My parents and my parents-in-law were crying all the time and it was difficult for us to speak to them. All my thoughts were to keep Andreas alive. I was crying at his bed, was praying and talking to him, not to leave us.

Then at a certain moment I realised, that I was very egoistic wanting to keep my baby just for the good of me. I told him then, that if he wanted to go, he could. From that moment, Andreas did not show anymore the signs of pain in his face. He looked even happy.

Then a miracle happened. Andreas' Dr went out of his room and met by coincidence another Dr and remembered that the former, one year before, had a girl patient dying of similar symptoms. They checked Andreas again and decided that **it was MCAD**. He got 20% glucose and liquid L-Carnitine through an infusion. Andreas was 5 days in coma. When he woke up we did not see any brain damage at first.

The hardest moment for me was that he did not recognize me. He was very friendly with everybody at the hospital. After one day he knew who I was, and we were all very happy. His right arm was moving all the time and while walking we saw a little difference in moving his right leg. But with therapy everything is okay now (except that he does not feel heat on his right leg. He already burned himself at the heating). He still takes (for 12 years now) 1g L-Carnitine from Sigma-Tau and of course his low fat/high carbo diet.

One year later, on his 2nd birthday, after taking the drug penicillin, he suddenly was unconscious for about 2 hours. He woke up in Hospital like he was drunk, but after another hour everything was okay. When he was 6 he had his first "grand mal" seizure. With the drug Tegretol CR 200 (since february 99 CR 400) a carbamazepine, we luckily had only 2 "grand mal" seizures and a few smaller ones we did not detect until in the morning when his bed was wet.

The last few weeks he is not very well. He forgets everything and had a different kind of seizure. He was not unconscious, but he could not speak anymore. The Drs told us that he has to take more Tegretol. Andreas is of normal intelligence but with reading, writing and math he is like a boy of 9 years. He is often tired and not much persevering, but he is very happy with a big imagination.

Iris, his sister, was born in November 1989. We had to give her the low fat/high carbo diet for 8.5 years until the Drs were sure she had no MCAD. She had to stay a few hours in hospital and they gave her a kind of fatty cream and every 20 minutes they checked the blood. We still do not know if she is a carrier.

We all got used to the situation and are a happy family. We even went abroad with the children a few times. Our first trip was to England, to visit my mother's cousin. Our Dr gave us the address of Great Ormond Children's Hospital, if something ever happened. The next trip was to Paris, France. As I speak french fluently I was not afraid of special situations. The third trip led us to northern Italy in my mother-in-law's hometown. As she was with us, she could easily translate the orders of low fat meals.

When we travel, we always carry 20% glucose, liquid carnitine, an appliance for determination of blood glucose and stesolid microclisma diazepam (against seizures) with us. We all cope quite well with the situation of MCAD. Epileptic-type seizures cause more trouble.

We were very glad to have found this Group, as we do not know anybody in Switzerland, Germany or Austria having the same disorder. Our Dr would not give us the name of other families and he would not pass our name to others! Having surfed the Internet has given us a second Christmas and **it was a relief to know we are not alone.**

Love from Katrin and Daniel Halbenleib
Switzerland
Andreas (MCAD) and Iris
dhalbenleib@bluewin.ch



Family Stories - Ella, LCHAD in the UK

Our beautiful daughter Ella Jayne North died on the 8th April 2000 at age 11 months. We first noticed something could be wrong with Ella at 4 months when she had just started on solids. She became a bit more sleepy and floppy and started having very pale stools.

One night Ella had her solids and bottle and vomited everything, but only seemed to want to sleep rather than eat again. Mike and I were going bowling with friends and thought how good Ella was being asleep all night. Eight o'clock the next morning we went in to her and found her hard to wake, so we took her to the hospital. Luckily even though they had no idea at this stage what she had, they put her on a 10% dextrose drip probably saving her life.

After days of tests and finding she had an enlarged liver we were transferred to Kings College Hospital for further tests. Luckily there we met a metabolic specialist, Dr Mike Champion from Guys. **He took a look at Ella**, her notes and along with her blood and urine tests and **told us she had LCHAD**.

At the time we felt relief because we thought she was going to have some unknown metabolic problem and die. A few days later, however, she developed bronchiolitis and ended up in ICU for 6 days. We then realized the seriousness of her condition.

Ella came home in time for Christmas, which was wonderful. With a good dietary plan and good management, Ella became strong, healthy and more beautiful and cheeky than ever. **We really thought we were out of the woods.**

On 5th April, Ella caught gastro-enteritis (nothing to a normal child). For 2 days she was on a drip because she was very worn out with sickness and diarrhea. But the saddest point came when Ella bent the needle from her drip so the nurses took it out. The doctor tried to put another in but couldn't find a vein ~ he said he would try again later.

In the meantime Ella had her nasogastric tube put down and was fed her maxijul, but **her blood sugars were not checked regularly as this would have found this option was not "strong" enough for Ella**, already being too weak. **Our beautiful daughter passed away early the next morning ~ they were unable to start her heart.**

The reason for writing our story to the Family Support Newsletter is to say to the other **parents of LCHAD children please never forget how special these children are.** When ill, start the emergency regimen as quickly as possible, get their blood sugars checked regularly when in hospital, and keep them on a drip until completely well. **Mostly don't be scared to speak up ~ we know our LCHAD children better than anyone.**

As you know we have a 1 in 4 chance of having another LCHAD baby. We'll keep you posted when we have some good news, but until then **our hearts, memories and love are with Ella.**

Julie and Mike North
United Kingdom
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Online 'Hearts of Light' Memorials

We would like to **add a new graphic page to our 'Love Message Memorials' ~ titled 'Hearts of Light' ~** where you can post your child's (living or deceased) picture, as well as special poems, artwork, stories etc., written or drawn by your children, yourselves, or other family members. It will be like having your own special web page in honor of or memory of your child or children. If you are interested in sharing your pictures or other contributions **please email them to Mary Lingle**, our webmaster and MCAD Parent, at mcartwrite@aol.com. You can also **snail mail them to me (Deb Lee Gould) at 805 Montrose Drive, Greensboro, NC 27410.** Please remember to **include a Release of Information Form** (see our website under 'Online Forms') with your contribution so we have your permission to post it on our website.





Family Stories - Kristen, MCAD Diagnosis in time!

Kristen's first episode happened the day after Christmas in 1998 just before her second birthday. She didn't eat very well on Christmas and was very busy opening presents. We all went to bed late and were very tired. We woke up very late, but that was usual when we went to bed late. When I checked on Kristen, she was unresponsive and she had mucous all around her mouth. When I picked her up, I could tell something was wrong. I took her down stairs and on the way down she had a seizure. I didn't know it at the time because I had never seen one.

We took her to the hospital where we waited and waited. I tried to get Kristen to drink some ginger ale when she was semi-conscious. Thinking back that was probably what saved her, because the doctor did absolutely nothing for her. We followed up with testing for a seizure disorder but it came back okay. The following February (1999) Kristen was hospitalized for pneumonia and dehydration. She was in good health until August of 1999.

Kristen had a major episode on the same day her little sister Jamie was born. She stayed over night with my parents and woke up very sick. She threw up all day and I tried to talk to her on the phone but all she would do was cry. I felt so helpless because I was an hour away in labor and I knew she needed me. As soon as Jamie was born I sent my husband home to be with Kristen. That was around nine o'clock ~ he rushed her to the hospital later that night.

She was unresponsive and throwing up brown bile. The doctors at the hospital thought she had taken some Insulin medicine, but nobody in our family had any. The IV was put into the bone of her leg because her sugar level was only 9 and they didn't have time to try a regular IV. I wasn't told of how serious her condition was because I was an hour away, and I had lost a great deal of blood during labor. Kristen was in the hospital ICU for three days.

We took her to a specialist and he **diagnosed her as hypoglycemic and that she would "grow out of it" and not to worry. He made that diagnosis without even seeing her medical records, which were lost.** I had to track down her medical records and I found out they were lost in the hospital's computer.

Kristen had one more episode after that. It was February of 2000. I dropped them off at the sitters and not even fifteen minutes later she called me and said Kristen was throwing up. I took her to the hospital and her blood sugar was a 55, but it was several hours before they could get the IV into her. They tried about thirty times before they called the surgeon down to do a cut down. As soon as the IV was in she perked up. It was then that the specialist mentioned the glucose gel and tablets that diabetics use. I couldn't believe it ~ something so simple could have possibly saved her this hospital trip. That is when I decided to get a second opinion and I am glad we did.

We scheduled a controlled fast for the end of June 2000. It went okay but she got really sick and had a lot of 'pinches' as she calls them. **On July 1, 2000 I got the call from the doctor telling me about MCAD.** I have never been so scared. Not only was it a life threatening disorder, but that both of them could have it! Jamie was hooked up to a monitor and I had to wake her up to feed her during the night. Not easy to do since she has slept through the night since she was two-months-old.

We are very lucky to have Kristen with us today. Kristen has special shakes (cornstarch) before bed and is taking Carnitor®. She is responding well to the Carnitor®. She has a lot more energy. In August she will have another EEG to make sure she doesn't have a seizure disorder. My advice to parents is to **always go with your gut instinct and don't let others tell you that everything is fine. If you think there is something wrong find out for sure.** If I didn't pursue this further we might have lost Kristen.

Sheri and Eben Merrill
Kristen, 3 (MCAD) and Jamie, 1 (MCAD carrier)
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Kristen, 3 (MCAD) & Jamie,
7 months (Carrier) Merrill





Family Stories - Sarah, MCAD

We are the parents of Sarah Ann Hall. She is a beautiful, active two year old. She is learning to go potty and do puzzles. She loves to watch Blues Clues and she can talk like the little puppy. She carries a doll all over the house and worries when she loses their bottles or shoes. She loves to swing in the baby swing in the tree in the backyard. She likes to jump in at the pool and play in the water. She is the most precious thing, next to our other children, that we have ever known.

Sarah died on July 30,2000. She died of a condition called **Medium Chain Acyl-Co-enzyme A Dehydrogenase Deficiency (MCAD)**, one of the many **Fatty Oxidation Disorders** that makes it impossible for them to use the fatty stores they have for energy. How can we not have known that Sarah had a fatal condition from birth? **Sarah had NO symptoms until the morning of her death.** She appeared to be a totally normal, healthy child. She had an acute episode brought on by the common cold or flu. We rushed her to the hospital in the ambulance. We, and the doctors, were completely shocked as we watched her slip away from us. Even when she was gone, the autopsy could not give a cause of death until 3 weeks later, when genetics tests were returned, and we finally knew Sarah died from MCAD.

MCAD usually affects children from the age of birth to 2 years. If you have a child of this age, get them tested now. **TEST YOUR CHILD!!!** I only wish someone had told us to have Sarah tested. **Ask your pediatrician and don't take no for an answer.** If you need help, call our pediatrician, Dr. Holly Bondurant, 573-874-3300. She is at the Boone Clinic, 401 Keene Street, Columbia, MO. There are two great websites about this at www.fodsupport.org and www.tylerforlife.com. There are two large labs that complete the testing for newborns for about \$25 per test. The **Comprehensive Newborn Screening (CNBS) test for 30+ metabolic disorders may be completed by Baylor University Medical Center in Dallas, TX at (800) 422-9567 or by Neo-Gen Screening in Bridgeville, Pennsylvania at (412) 220-2300 (*see more info on FOD website under Medical Info/Diagnostic Labs).**

We need your help to educate parents about this disease. We would like for you to make parents aware that screening for metabolic disorders is possible. It is being screened for in many other states. Someone must tell parents to screen for these disorders. We would have if we had known it was important. **IT IS IMPORTANT!** Parents should have it done in the hospital as a newborn. **If you know someone who is having a baby, tell them. IT IS IMPORTANT!!!**

We need your help to get the laws in the State of Missouri changed to include all metabolic disorders to be included in the ROUTINE COMPREHENSIVE NEWBORN SCREENING. If Sarah would have been screened as a newborn, her MCAD would have been detected and we could have treated her with medication and diet. **She could have lived a normal, healthy life IF she would have been tested.** The State of Ohio is beginning this screening now and hopes to help almost 1 in 1,200 newborns by detecting these disorders.

We are asking that Doctors recommend Comprehensive Newborn Screening for the 30+ metabolic disorders. It is the RIGHT THING TO DO to advocate to parents to have their child tested. Please do the RIGHT THING beginning today!!!

We are praying for you,

John and Carol Hall

Paris, MO

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(*Sarah's story also on our website, NBS page)

New Baby ~ # 6!!!!!! On November 22, 2000 the world welcomed Madison Daniele, daughter of Kristi and Gary Jones (Ashley and Mackenzie, MCAD). Madison weighed 7lbs 14 oz and was 20½". We have recently heard that Madison does not have MCAD. Congratulations to the Jones' on their new addition!



'Hope' for the Meadow's Family! Congratulations also to Linelle and Matt Meadows (Cole, undiagnosed MCAD, deceased) on the arrival of their daughter, Hope, on November 26, 2000. The family is excited about her arrival and even more excited that she is MCAD-free!



Medical Update

Distinction between: Supplemental Newborn Screening for amino acids and acylcarnitines by tandem MS and Quantitative Acylcarnitine Profile by tandem MS for Patients with Clinical Symptoms

By Dr Larry Sweetman and staff
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Tandem MS (MS/MS or tandem mass spectrometry) is a technology that is used in various ways to measure many different compounds. Specific tandem MS methods can detect elevated levels of biochemical compounds in blood that are useful for diagnosing many inherited disorders of metabolism. **It is important to realize that the different types of tandem MS tests are designed to detect specific disorders and therefore may not diagnose some diseases.**

Supplemental Newborn Screening is typically done in conjunction with state newborn screening and may include other tests in addition to tandem MS. To determine what your child may be tested for, one should request an information sheet from your testing laboratory. This should list what disorders will be tested for, such as amino acid disorders, organic acid disorders, fatty acid oxidation disorders or other types of disorders.

For patients with clinical symptoms or family histories of inherited metabolic disorders, diagnostic tests for specific disorders are much more appropriate than a general screen. The specific tests, such as urine organic acids, plasma amino acids, plasma or dried blood spot acylcarnitines should be determined and ordered by clinicians based on the clinical symptoms.

Supplemental Newborn Screening by tandem MS: In our laboratory, the Supplemental Newborn Screening is done by tandem MS for amino acids and acylcarnitines. The test can **detect more than 30 inherited disorders of amino acid, organic acid and fatty acid metabolism.** It is an inexpensive screening test with dried blood spots designed for screening healthy newborns at 2-3 days of age. **It is not recommended for the diagnosis of children with clinical symptoms.** Cut offs for normal levels of amino acids and acylcarnitines in newborns are chosen to minimize false positives. Approximately 1% shows some slight elevation of an amino acid or acylcarnitine, which causes us to request a repeat dried blood spot card to see if the slight elevation is significant. Usually the repeat is normal. The false negative rate is not known but is believed to be very low. In order for this test to be inexpensive (\$25), it is highly automated. The quantitative results are analyzed automatically by tandem MS computer programs for "normality" using cut offs established for different groups of subjects. Although the screening is most accurate for newborns it is still worthwhile to perform for older infants if they were not screened in the newborn period.

Quantitative Acylcarnitine profile by tandem MS is a more expensive test designed for the differential diagnosis of fatty acid oxidation and organic acid disorders in children with clinical symptoms or family histories suggestive of these disorders. This can be performed on dried blood spots, plasma, postmortem blood (or amniotic fluid with prior consultation). Because it is **intended for diagnostic evaluation**, the cut offs used for acylcarnitines are lower and every profile is examined by a technician for any abnormalities in the mass spectrum, including peaks that are not part of the quantitative analysis. **Every profile is reviewed by an experienced professional and reported with a professional interpretation.** The acylcarnitine profile does not include amino acid disorders.

Update on NBS Pin Fundraiser: Thank you to ALL that participated in the NBS Pin Fundraiser that helped promote awareness of Supplemental Newborn Screening for 30+ metabolic disorders in September 2000. **We participated along with the Tyler for Life Foundation (www.tylerforlife.com), whose main mission is to advocate that cause. A total of 26,000 pins was purchased (many other organizations participated) and TFL distributed 17,000 of them ~ 1000 of which were from our FOD Families!**

Eileen McMullin, who coordinated the project for the FOD Families, has asked me to post a **RE-MINDER** to those families that still need to turn in their money for the purchased pins. **Please send your money in the next few weeks directly to Tyler for Life Foundation, 6340 Holborne Lane, Douglasville, GA 30134 (home office # 1-888-454-3383).** Tera is looking forward to hearing from you! Families and Professionals can still **purchase pins for @\$1.00/each from the TFL website** or by emailing Tiffany Young at tiffany@tylerforlife.com. THANK YOU!

Thank You



Family Stories - GA2 x 3 in the UK

'Working with the schools to get what your child NEEDS!'

Hi Everyone in the FOD Family Support Group. **My grandchildren, Jamie (9), Lewis (2½), and Emma (1½), all have GA 2 or MADD.** To say the least, we have been overwhelmed at times with all of their needs (i.e., medical needs due to residual effects of late diagnoses), but **this time we have GOOD NEWS to report!**

The good news is, that **after five years of talks and meetings** with the Local Education Authority, they have decided that **Jamie, because of her medical needs, should have a nurse with her so that she may attend her new mainstream school with safety.** But it really has not been a smooth ride. It is the result of attending meeting after meeting for 5 years! **Persistence is important!**

In order to come out on top at these meetings it is vital to arrive at them prepared and ready to argue your point calmly. If a suggestion for a possible plan comes from one of the earlier meetings and you know it is unworkable for whatever reasons, the temptation will be to dismiss it ~ DON'T! Instead point out your doubts but agree to try it. Next, if something gets fouled up, document it ~ write it down including time, date, and staff involved. **The school staff CAN BE your strongest support for your child, but they must be aware of your child's particular problems and concerns.** Discuss your doubts with them and don't worry too much at this stage about the response you get. You'll find out soon enough whether they have listened to you or not! We have learned that the hard way over the years!

Go ahead and let them try THEIR new plan (but you know it's unrealistic) and watch it fall flat. The staff you have talked with will THEN remember your doubts and your suggestions. **Remember that these are the people that have to make whatever plan is chosen work.** You will need their input. Also never forget that they work for the people you are trying to influence (administrators). They know the way the system works and this is where they can help you.

Once you have a member of the staff that is able to understand your doubts and concerns ask them to make a report for you to take to the next progress meeting, because these issues are likely to be complex and having them written down will help others better understand your point of view. **That staff member can be an advocate for your child at that next meeting and any future meetings.** You now have someone that understands your child's particular needs and problems (i.e. medical, physical, cognitive, emotional etc) and because they know how the system works, you can use them as a sounding board and support person just before meetings with other teachers and administrators.

You now have two people in that meeting (yourself and the staff member) that are aware of the facts at grass root level and believe me it really makes things a great deal easier. A word to the wise ~ **these meetings will most likely make you very angry on occasion so learning to redirect that anger and get a fair deal for your child is VITAL!** It worked and is still working for us. Warmest wishes,

Woody and Company (BIG HUGS FROM THE KIDS)
chazle@ukonline.co.uk

*****Note from Deb:** Thanks for all of your calls, Woody and Clare (GA2 mom) ~ I also REALLY enjoy talking with Jamie. Even though we can't see each other and it's sometimes difficult to hear her, **I KNOW we are 'connecting' across the ocean in a very different and important way. Hugs your way too!**

IRS tax deduction for Medical Expenses

The IRS has issued a ruling (Section 213 – Medical, Dental, etc., Expenses) that will **allow parents to deduct some of the costs associated with attending medical meetings related to their child's health condition.** The ruling reads: **Medical expenses.** *Amounts paid by an individual for expenses of admission and transportation to a medical conference relating to the chronic disease of the individual's dependent are deductible as medical expenses under section 213 of the Code (subject to limitations of that section), if the costs are primarily for and essential to the medical care of the dependent. The cost of meals and lodging while attending the conference are not deductible as medical expenses under section 213.*



Family Stories - Josey, misdiagnosed as SIDS

I want to thank you for sending me the Newsletters. Before I got those in the mail my husband, Steve, and I thought we were the only ones going through this. We couldn't believe how many other families have gone through exactly what we have. The day we got the newsletters we both sat down and started reading the stories of other families. We were told this disorder was rare, so we were in shock to hear just how many families have lost children or have children living with a metabolic disorder. **All the stories really got to our hearts. We know the pain you all felt. There is no greater pain a parent can feel than the pain that comes from losing a child.** I am so sorry it took me so long to send my story to you. This was a very difficult thing for me to do. Well here it goes.

Josey Deanna Rowland was born on September 30, 1996. She weighed 8lbs. 4 1/2 oz. and was announced to be a healthy baby girl. Steve and I were so happy. We already had a two-year-old son, Devin, and now a beautiful baby girl. We felt like the two luckiest people on earth. Our dream was to have a boy and a girl. Now our family was complete. Josey was an angel sent from above. She was always smiling and making everyone around her smile also. Even when Josey was sick she still would be happy. The first year of Josey's life she got sick pretty often. She was hospitalized four times before her first birthday. And each time she always had bronchial pneumonia and would be in the hospital for at least five days each time. **We were concerned how often she got sick but the doctors told us she just got sick easily.**

Well, when Josey turned ten-months old she was put in the hospital for the fourth time, and still the doctors did not seem to be concerned on how often she got sick with pneumonia. We were at the point of wanting to get a second opinion, but when Josey got out of the hospital that time she never got pneumonia again. She never was hospitalized again after that. We were no longer concerned anything was wrong with her. Josey was happy all the time. She loved playing with her big brother, what time she was not under me. She definitely was a mama's girl. And because of that she never stayed away from home.

Except for on October 27, 1998. That day Steve and I were having some personal problems and I decided to stay with my parents for a couple of days. Later that night, I decided to go home and work things out with my husband. My sister told me to go on home and she would keep the kids so we could talk. Before I left I gave Devin and Josey a bath, and fed them supper. Josey was in such a good mood and did not let on that anything was wrong with her. She had a little cough and that was it. I remember before I left that she had one diarrhea diaper, but I thought something she ate, may have torn her stomach up. When I left Josey that night she was fine. She was happy and playing around like she always does. **When I left that night I had no clue that would be the last time I would see my little girl alive.** I did not even tell her bye or I love her that night before I left, because I knew she would cry to come with me. I never could leave her crying. **I regret so much not telling her I loved her before I left.** I thought I could make up for it the next time I saw her, but how was I to know something so tragic was going to happen to me ~ something that would change my life forever.

Well it is hard for me to go into exact details on what happened the night Josey died, because I was not with her. All I know is what my parents and sister told me. That morning when my husband left for work his mother was supposed to go and pick up the kids and bring them to me. I remember getting up that morning in such a good mood. Steve and I worked out our problems, and I couldn't wait for the kids to get home. I will never forget that morning. I heard a car coming up the drive and I assumed it was my mother-in-law bringing the kids home, but when I looked out the window and saw that it was the secretary that worked with my husband my heart dropped. I knew right then something was wrong. She got out of her truck and she was crying, and all she would tell me was to get in the truck. The first thing that came to my mind was my husband got into a car accident. **Not once did it run through my mind that it was one of my children.** I was crying so bad that I could hardly talk. I tried to get her to tell me if Steve was all right. I just knew he was killed in an accident. She looked at me with so much pain in her eyes and said, "Janice, I was told to get you and Steve to the hospital as fast as I could." When I heard her say you and Steve, I really broke down, I knew then that it was not Steve I was going to the hospital to see. I felt sick to my stomach by this point. **I knew then it was one of my children. I just didn't know which one.** That five-minute drive to the hospital seemed like an hour. All I could say the rest of the way there was "God, please let my baby be all right. Which ever one it is, please let them be all right."

There was so much going through my mind at that time. Not once did I imagine pulling up at that hospital and seeing what I saw. When we pulled in the driveway of the hospital I saw my dad, and when I saw his face I knew then it was bad. I started screaming as soon as I got out of that truck, and all my daddy could say to me was "I am sorry, Jancie. I'm sorry." **I ran in the hospital and saw it was Josey lying in that bed.** I practically threw myself on Josey, and I told her that she had to be all right. Still not knowing that she was already gone I asked the doctors why were they just standing around and doing nothing, and I told them to help her. **The doctor then told me they were sorry but they did all that they could do for her.** I assumed that they meant that it was up to God on whether or not she got better. Not one time did her being dead cross my mind. **I just hugged her and kept telling her that she had to be okay, because I couldn't go on without her.**

At this point Steve realized I was thinking she was still alive. I will never forget when he turned me around and said to me, "**Janice, she is gone.**" I went hysterical and it took almost everyone there to hold me down. I just couldn't believe what I was hearing. I didn't want to believe what I was hearing. I picked her up and she was so fragile. I called her name and she didn't respond. At that moment I knew she was gone. I can't begin to tell you what I was thinking and how I felt at that very moment. I honestly felt like my life was over. I felt like my life couldn't go on without her.

I didn't understand what could have possibly happened to her. The night before she was fine and now my precious baby girl was gone. **I questioned the doctors and all they could tell me that she probably died from SIDS** and we wouldn't know for sure until the autopsy report came back. I remember when they took her away from me that day. A part of me died at that moment. I couldn't believe this was happening to us. We left the hospital in total shock. I then questioned my parents on what happened. They began telling me everything she did from the time she went to bed and the time she woke up the next morning. My mom said that Josey woke up around 4am coughing. And then she threw up. She thought she threw up because she was coughing. She said when she cleaned her up Josey went right back to sleep. Around five that morning my mom left for work.

Continued on page 11

Family Stories - Josey, misdiagnosed... (cont'd)

Daddy said she woke up around six moaning and shaking a little. He thought she was cold and covered her up. He said that when she moaned he would rub her face and she would go right back to sleep. He said around six-thirty or so she woke up and was crying a little, so he got up with her and sat at the kitchen table and gave her cup to her and he said that she acted like she couldn't hold her eyes open so he took her back to bed. Thinking that she was only sleepy. He said at seven thirty a policeman came to the house to get him because my brother got into a bicycle accident on his way to school, so he left Josey with my sister until she had to go to work and my grandmother was to look after her until my dad got back.

Well my sister said she left for work around eight-thirty and she looked in on her and she was still asleep. She said she didn't go up to her because she was scared she would wake her and she would cry to go with her to work. She said before she left she told my grandmother that she was still asleep. Well a few minutes after my sister left my mother-in-law came to pick them up to bring them to me. She told me that she went to get Josey out of the bed, and when she walked in she called her name and she did not respond so she went to pick her up and she knew something was wrong. She said she then took her to the emergency room.

That was everything I was told. My daddy blames himself for not knowing that something was wrong with her. It was hard hearing all that. I, too, felt that if she would have been with me I would have known that something wasn't right when she was moaning and groaning in her sleep. I feel like I could have saved her if she was at home with me. I don't blame my dad, he had no idea what she was doing was not normal. It was hard on all of us. With the help of family and friends we made it through the funeral.

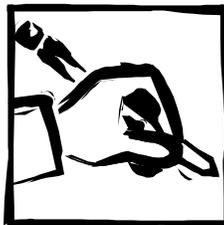
It was three weeks later when we got the call and was told that **Josey died from viral pneumonia in the background of MCAD.** They couldn't tell us much, because no one knew anything about it. All they told us was it made the blood sugar drop rapidly. They set us up an appointment with a counselor to explain to us exactly what MCAD was. **When we learned that this type of disease was treatable if known about, we were so angry.** We were upset that these tests were not performed on babies at birth. We also learned that our older son could also be affected. We had him tested and found out that he was a carrier and not affected. We thank God for that.

Well two and a half months after Josey died, I found out that I was pregnant. I was in shock, and we were so scared that this child would also be born with MCAD. On October 5, 1999, I had a 7lb lloz baby boy. **Drake seemed to be healthy, but the test was done at birth and showed that he, too, was affected with MCAD.** We were grateful to know about it this time, but we were scared to death. We didn't know what to expect. We had to take him to Atlanta at Emory University, and there we learned how simple the treatment is. He was on simlac with iron, and we had to add polycose, provimin, and Carnitor® to his formula. We learned that he had to be on a certain diet and as long as we did his diet right and not let him fast for a long period of time he would be fine. Well Drake is now one-year-old and I am happy to say that so far he has been fine. He has been hospitalized twice, but nothing serious. He had a cold and they put him in the hospital to watch over him. He is energetic and happy all the time ~ just like his sister.

Since Josey's death I have wanted to do something to make others aware of these disorders and to get the states to allow these screenings on babies at birth. I want you all to know that I will do my part to help Tyler For Life Foundation to raise the funds to help make this happen. I am sorry to hear about all of you that have experienced what I have, and I want you to know that you all are in my prayers. I believe that all of our children are in heaven watching over us and helping us get through everyday. **I do believe that if we all work together we can make this happen. Our children will live forever in our hearts.** God bless all of you.

Janice Rowland
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229-686-3929
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Pen Pal Program ~ Would your child be interested in making a new friend and getting to know another child like him/herself? **Why not sign them up for the FOD Pen Pal Program!** What a great way for them to share how they feel about their FOD. Contact **Kelly Madej** at yadmad@aol.com, 630-375-9543 or (fax) 630-375-9546 for more information. Also see www.fodsupport.org (Online Forms) for the Permission Form to sign up your child.



Comprehensive Newborn Screening Kits to detect 30+ disorders in newborns, children and adults (when calling, specify if for an adult, procedures may vary) can be ordered for @ \$25 through:

Baylor University Medical Center ~ Institute of Metabolic Disease
1-800-4BAYLOR
www.baylorhealth.com/newbornscreening

Neo-Gen Screening
412-220-2300
800-892-1288

www.neogenscreening.com

[Be aware that if the individual is experiencing symptoms, a different test or tests (i.e. acylcarnitine profile) may need to be performed ~ refer to the article in our Medical Update section]**



Family Stories - Joshua, MCAD in the UK

Our story begins on the 20th May 1997. My son, Joshua, became ill with sickness and diarrhea, which we were told not to worry about ~ "It's just a tummy bug and if he is no better by the morning, bring him back." The morning came and we could not wake Josh. He was rushed to the North Devon (United Kingdom) district hospital where he needed to be brought back to life. He died on several occasions before slipping into a coma and being put on a life support machine. He was nearly 9-months-old.

All the tests taken came back negative except he had no blood sugar. He was then airlifted to Great Ormond Street Hospital London where he stayed in a coma for 5 days. **He was later diagnosed as having MCAD** after a week of having extensive tests done on him by Professor Leonard. When he awoke from his coma, he then fitted (seizure) for 53 minutes. We later found out that it had caused him to twist at the hip and that he was severely brain damaged from the episode.

When we brought him back to Devon, my wife and I realized the damage was far greater than first thought. He had also lost his sight and his ability to swallow and needed a gastrostomy tube fitted. He also was going to suffer from fits. We spent many weeks that year at the hospital with Josh.

In December of 1997, Josh was again critically ill with RSV bronchial pneumonia and we were told he would not make it through the night. The doctor put Josh on oxygen and by the morning he somehow had made an amazing recovery. He spent two weeks in hospital. We then went back to Great Ormond Street for a check with Professor Leonard, who said he would discharge Josh and would not see him again because he didn't think he would live because of his illness and having RSV made it very difficult for Josh (Josh went back to Great Ormond Street in May 1999 and Professor Leonard was shocked to see him!).

We spent most of 1998 at the hospital with various chest infections, plus he also caught sudamonus membrenas colitis and again we were told that Josh would die because the antibiotics he was on he had become immune to and there was nothing that could be done. **Again, he made an amazing recovery.**

The amount of drugs he has been on has been reduced dramatically. **We were informed that Josh would never walk or talk because of the damage to his brain, but we were also told he would not reach his first birthday ~ he will be four on August 29 this year!**

He is getting stronger. I have read that no child has died after being diagnosed with MCAD. Josh has a very weak chest because of the amount of chest infections he has had and it will be THAT that kills him, not the MCAD.

I need your experience on this matter and what to expect in the future ~ such as what other things are we likely to endure? We are alert to Josh and his chest 24 hours a day. Our other two children are young and have accepted Josh as a 'disabled' brother. My ten-year-old son is very protective and my eight-year-old daughter has just taken it for granted as every-day life.

What (without me building my hopes up too much) is the chance of Josh making a full recovery or at least being able to walk or even talk or seeing properly? **I wonder if you have any other families in the same situation who have older children who have suffered the same or just as bad as Josh and how are they coping?** Are the children able to lead nearly normal lives? Where do we go from here? Are you the angel I have been looking for in the last three years to help us with our son's condition? **Can you (or the other MCAD/FOD Families) spread some light on this very rare disease so I can keep ahead of it and help Josh to lead as normal of a life as possible? We are all in this together!** Many thanks for your time and effort,

Mr. and Mrs. Gwyn Morgan
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GOOD HOUSEKEEPING MAGAZINE

Be sure to look for the **FEBRUARY 2001** issue of *Good Housekeeping* Magazine. They are devoting a section to **Supplemental Newborn Screening** and a few familiar families were interviewed for the article.

To all of you that are **GIVING A VOICE** to
ADVOCATING this VITAL SCREENING ~
Keep up the GREAT WORK!



Family Stories - Joseph, Unclassified FOD

Our family's story began two weeks after our son's first birthday on April 20, 1998. He had not been feeling well ~ severe stomach pains, high fever, throwing up constantly. This was the first time he was ever sick at all. By the second trip in three days to the doctor's office, the doctor said it was not just a stomach bug as he initially thought, but something more serious and we needed to bring Joey to the hospital immediately. Thankfully the hospital was right down the street. The doctor there said that if it were something very serious he would send us directly to Children's Hospital in Boston. They did some initial tests and an x-ray and thought that it might be an appendix, but that's not common at such a young age. So our son was transported to Children's by an ambulance. My husband and I had to leave Joey in the hands of strangers, doctors and nurses, yet strangers just the same. We were to meet them at the hospital.

We got to the ER at Children's where it was eventually **determined that Joey had an intussusception. This is when the intestine telescopes up inside itself and causes severe blockage, and it is fairly common among children his age.** First they try an air enema, but after about three attempts it doesn't work, and they don't want to blowout his intestine, so he would have to have surgery. During the operation they may or may not have to remove some intestine, and he may or may not have to have a colostomy bag. They wouldn't know until they opened him up. We waited, and waited, and waited for what seemed like forever, especially since it's after 3 am. Nothing good happens at three am. By about 4 am or so, about three or four hours after the surgery started, the doctors told us that the surgery went well. The intestine was slightly red and raw, but they didn't have to remove any of it and he didn't have a colostomy. Thank God, this was the best outcome we could have asked for, or so we thought.

A few hours after the operation Joey woke up for a little while and was being goofy with me, being silly with his binky and blanket ~ that was the last I would see of my son for a very long time ~ the silly little boy that we knew, anyway. **Over the next few days we watched our son slip further and further away from us, although he was supposed to be recovering.** We watched his little 20-21lb body swell up like a balloon. His skin got so tight that it looked as if you could pop it with a pin and he would go flying around the room. The doctors believed he was feeling worse because the meds were wearing off and the body does tend to swell after a trauma, such as an invasive surgery like his. They said he should be fine by the weekend and we could go home then. "Get him up and moving around," they said. We tried, but he couldn't move. He was too swollen. On one of these "walks" around the floor, we carried him just to get him out of his room. **I had an overwhelming feeling that there was something wrong with his liver.** I knew a friend of a friend when I was younger and she would swell up or get puffy after a treatment, but I didn't know if it was from her meds or the problem with her liver.

I told my husband this story as we "walked" with our son, and now believe it was God's way of telling me or preparing me for what was happening to my son. **It was an intuition like nothing I had ever experienced before.** We continued to question Joseph's condition, but we were always given an answer that seemed logical. After all, they are the doctors and nurses and we are just the parents. I would later come to realize that **yes doctors and nurses are very good at what they do, but there is nothing like a parent's instincts.**

Through help of other parents **we learned never to doubt those feelings again.** Doctors know symptoms, and medications, and a lot more, but **only you know your child and better than anyone.** Never dismiss that. Joey's condition worsened to the point that we could not get him to focus on my husband's finger, his eyes were all over the place. He was hallucinating, trying to drink from an imaginary bottle. He eventually started turning red, getting stiff, shaking and screaming. He was convulsing in a seizure. The doctors tried to get blood samples to tell what was going on inside him, but he was so dehydrated that they couldn't get any. They asked permission to try a vein in Joey's head. We said do whatever you have to, just save our baby. No luck there. They finally went to the groin area where you can usually always get blood. After pulling back and empty syringe for the third time the frantic look on everyone's faces was obvious. By this time Joey had stopped crying and fussing. He was just lying there. This was more disturbing than the constant crying and screaming. I do not know if he was clinically in a coma, but that's how I would describe him. He was breathing, but he was asleep with no response to verbal or other stimuli.

The head surgeon did finally manage to get the blood. They rushed Joey to the MICU (multidisciplinary intensive care unit) and were trying to get him ready for another surgery. The doctors came and talked to us. He had gone into liver failure, kidney failure, and pancreas failure, too, and they had no clue why. They needed to go back in to see if he had intussuscepted again and possibly do a liver biopsy, but they needed to stabilize him first. They told us he may not make it, they were going to do everything they could, and you know they meant it, but we needed to be prepared. The hardest part was putting him under. He was so unstable that putting him under the anesthesia alone could kill him. What choice did we have?

Continued on page 14

Family Stories - Joseph, unclassified FOD (cont'd)

They came back to us just after the surgery started. He had gone under and was doing all right ~ so far so good. A few hours later, our families by our sides again at 3 am, we saw them wheel his bed back into the MICU. The child I saw was now bloated to 30lbs. That's 10lbs of fluid under the skin on the body of a 13-month-old baby. He had tubes in his nose and down his throat, he was on a ventilator, breathing for him, and he had monitors hooked up all over his body. My son had become this unrecognizable, swollen, creature on a breathing machine with blood on his face and tubes coming out from all over his body, and beeps going off, and I couldn't handle it. For the first time since this whole ordeal started I had this anxiety attack and I ran from the room. That couldn't be my little boy lying there, it just couldn't be. I had cried before, I had felt hopeless, I had prayed constantly, in fact I never felt closer to God, but in that moment I felt complete despair and disbelief. **Nothing could have prepared me for what I saw ~ nothing could ever prepare a parent to see their child like that.**

But do you know what the real kicker is? **We now know all of this could possibly have been prevented. If there was infant screening we may have known Joey had an FOD** and his care for the intussusception could have been modified to compensate for his condition. Joey hadn't intussuscepted again, but they did manage to get a liver biopsy. Joey did eventually get better. He began breathing on his own and got moved out of isolation. We started to see glimpses of the little boy we'd known, again. But he also went through bumps along the way ~ fevers that they could only get down with cooling blankets. Because of the damage done to his liver, it was only 30 percent functioning at one point. They could not give him Tylenol. They put him on an experimental drug that they sometimes give to those who have overdosed on Tylenol, but it gave him a rash all over his body and it had to be stopped. He got a few doses in before the side affects showed, so it could have helped. His heart failed at one point. He had a rapid heart rate and there was fluid around it. It was as if the toxins were making their way through his whole entire body, and they were. We just prayed it wouldn't go to his brain next.

The first time he opened his beautiful blue eyes again I was asking him where Brach was. Brach is our beloved chocolate lab. From that point on I knew if he remembered Brach he would remember us. I just wouldn't allow myself to think otherwise. The doctors were not so sure, he had toxins in his brain, that is why he had those seizures, and no one was certain what damage had been done. The only way to assess the damage was to wait and see how he reacted to stimuli, mental and physical. **My Mother's Day gift that year was that we got to leave the MICU, and go back to the 8th floor. It was the best present I will ever get (besides going home).**

A month after we had come to Children's we were finally going home. And two months later the metabolic doctor called to say that the **skin biopsy they did on Joseph when he was in the hospital came back positive for an FOD.** It was a sad moment, but it was also a relief because it explained a lot about what had happened to Joey. It gave us much anticipated answers. The reason Joey had gone into multiple organ failure was that when his body ran out of energy it went to his food stores, but because he lacks the proper enzyme to break fat down and turn it into energy it caused a toxic effect and those toxins then went through out his body. It only takes a child 4 to 6 hours without an outside energy supply to go to his food stores, in Joey's case he hadn't been able to keep anything down for about 48 hours before he was brought to the hospital. When he was in the hospital **he was given IV fluids, but not at the proper dextrose level for a child with an FOD. That IV, however, is what kept him alive, but at the same time it wasn't enough dextrose and that is why he kept getting sicker but stayed alive.** He was allowed only a minimal amount of oral liquids because of the type of surgery he had gone through. It was almost a week after he first got sick that they took him off the IV and that is when he spiraled down and started the seizures.

Although we still don't have a complete diagnosis, **we know he has an FOD and are still working with doctors to label his exact disorder.** The anesthesiologist, that kept Joey alive during the second operation, told us that he had never seen a child as sick as Joseph who survived, and on the rare occasion a child had survived, he hadn't seen one survive without any residual side affects. We credit God, all the many doctors and nurses that worked and continue to work with us, and the many people who prayed for Joseph, many of whom we don't even know, for his survival. We also thank our families and friends for always being there ~ you all made a very difficult time a little more manageable.

Today Joseph is a healthy, happy, beautiful, and silly 3-yr-old boy, whom you'd never know anything had ever happened, except for the large scar across his abdomen just below his belly button. **He is living proof that there is power in prayer and a living reminder, for which there are many not living, that infant screening is a necessity. No parent should ever have to go through what we went through or have to lose a child to these treatable disorders. No child should have to die or almost die to be diagnosed. Our sincere hope is that no other child ever will.** God Bless,

Stacey and Paul Webber (Joseph, too!)
Marshfield, MA
staceylwebber@cs.com



Love Messages

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

Joan & Tim Aalberts
Luke - Birth June 7, 1994 Death May 1, 1995

Sandy and Howie Aitken
Kristopher - Birth Dec 22, 1997 Death Dec 29, 1997

Jeanne and Mark Barilla
Michael - Birth Feb 2, 1990 Death Nov 25, 1990

Jodi and Wayne Barnes
Amy - Birth Feb 20, 1995 Death Sept 27, 1995
Baby Barnes - Death in-utero Oct 7, 1999

Delane and Althea Becker
Warren - Birth June 9, 1987 Death Feb 4, 1990

Sue and Jim Berneski
Michael - Birth Dec 28, 1992 Death Jan 3, 1993

Jacque and Mike Bradford
Eric - Birth Aug 22, 1990 Death Aug 26, 1990

Joseph and Barbara Brown
Amber - Birth June 18, 1989 Death May 17, 1990

Barry and Julie Bryson
Skyler - Birth Nov 25, 1989 Death August 25, 1990

Carolien Grootaert - Callens
Liese - Birth March 2, 1988 Death Aug 25, 1990

Tom & Lynn Camino
Stephanie - Birth June 28, 1995 Death Feb 6, 1996

Mark and Karen Carpenter
James - Birth May 7, 1985 Death Dec 6, 1986

Jenny and John Carroll
Sarah - Birth March 4, 1992 Death Sept 1, 1992

Mark and Diane Casey
Matthew - Birth Apr 15, 1974 Death Jan 13, 1975
Jennifer - Birth Oct 19, 1975 Death Nov 13, 1979
Lori - Birth Aug 31, 1980 Death July 1, 1984

Valerie & Chris Ciachette
Benjamin - Birth Jan 12, 1987 Death April 18, 1987

Toni and Mark Cline
Kasie - Birth June 6, 1990 Death March 10, 1991

Sandy and Jon Cooper
Noah - Birth Oct 5, 1997 Death June 30, 1998

Martin and Kathy Davis
Mary Katherine - Birth June 27, 1996 Death Nov 7, 1996

David and Amy Deshais
Megan - Birth Feb 11, 1991 Death July 7, 1991

Doug and June Evenhouse
Marie - Birth Dec 15, 1985 Death Nov 19, 1986

Andrea and Phillip Franklin
Brandi - Birth Dec 2, 1986 Death Jan 1988

Lance and Dawn Goldsmith
Joel - Birth Feb 15, 1990 Death Nov 16, 1990

Deb and Dan Gould
Kristen - Birth Oct 6, 1983 Death July 21, 1985

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

Michael and Nicole Gumiel
Michael - Birth March 28, 1998 Death April 4, 1999

Carol and John Hall
Sarah - Birth June 8, 1998 Death July 30, 2000

Robin and Vince Haywood
Ben - Birth Feb 19, 1998 Death Aug 8, 2000

Ralph and Angie Hedrick
Chelsea - Birth Jan 11, 1995 Death Apr 3, 1986

Nikki and Toby Hiatt
Reece - Birth Aug 1998 Death April 18, 1999

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

Continued on page 16

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

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

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

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

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

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

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

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

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

Lisa and Pete Leonardi
Devin - Birth July 18, 1997 - Death July 19, 1997

Mary Lingle
Candice - Birth Feb 2, 1991 Death Nov 8, 1993

Darlene and Larry Lopez
Marissa Death Feb, 1999

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

Ron and Paula Matthews
Daniel - Birth May 19, 1981 Death Jan 12, 1982

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

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

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

Elvira Melendes
Katherine - Birth March 6, 2000 Death May 3, 2000

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

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

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

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

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

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

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

John and Sally Reichelderfer
Zachary - Birth March 24, 1997 Death March 27, 1997

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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



*'You can't heal a wound by
 saying it's not there.'*

~Jeremiah 6:14



Pharmaceutical Update

Sigma-Tau Pharmaceuticals, Inc., makers of **Carnitor®** can be reached at **800-447-0169** or on their web page www.sigmtau.com. They have launched 2 new websites to **Educate and Empower Providers and Consumers** ~ www.Carnitor.com and www.ReimbursementCommunity.com. The sites are designed to provide healthcare professionals and patients important information about **carnitine deficiency and Carnitor®** (levocarnitine).

• • •
Metronidazole used for the treatment of body odor associated with L-carnitine supplementation ~ see article by Dr. Susan Winter in the October 2000 issue of Organic Acidemia Association newsletter at www.oaanews.org



National Metabolic Conference May 4 and 5, 2001

(see www.pku-allieddisorders.org for Registration/Speaker/Travel Updates)

The National Coalition for PKU and Allied Disorders is holding a **two-day metabolic conference** at the **Embassy Suites Hotel, Dublin, OH** (near Columbus, OH) on **Friday, May 4, and Saturday, May 5, 2001**. The **FOD Family Support Group and 6 other Groups** have been invited to attend.

Conference Registration (due **by April 15**) is **\$50.00 per person** for the two days. **Room rate** is **\$99.00 per night**. Each room, may sleep 6, has a refrigerator, microwave, coffeemaker, blow dryer, and pull out couch. **Price includes** continental breakfast and an early evening snack/soft drink. **Making Reservations:** You may call **(800) EMBASSY** anytime. **To receive the special room rate:** must tell them that you are attending the meeting for National Coalition for PKU & Allied Disorders. There are 13 rooms with accessibility for wheelchairs, there is also an indoor pool and a hot tub. **Travel Reservations:** contact **MSUD Mom and travel agent Cathy Codner** at **800-825-8125** or do on own. **Ground Transportation:** Taxis \$30-\$47 one-way. No hotel shuttle service.

Speakers for the **Friday, May 4th FOD breakout meeting** include **Dr Charles Roe and Dr Jay Cook**, Baylor's Institute of Metabolic Disease and **Dr Rani Singh**, Emory University. The Agenda is still being worked on so refer to the National Coalition's website for further speaker/agenda/topic updates.

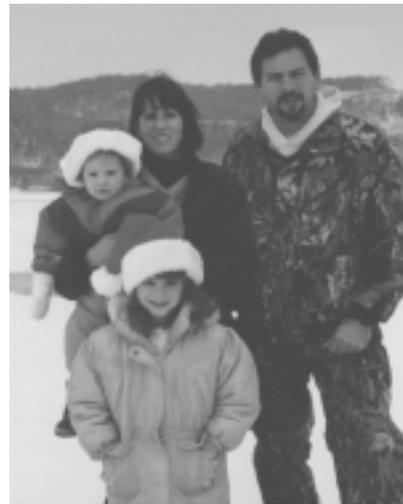
Day Care provided for FODs, Organic Acidemias, and GA1 ONLY: **Children over 1 year of age can sign up for childcare, but must pay the \$50 registration fee (will cover meals, childcare)**. We hope to have several activities as well as the Columbus Zoo's small animals. Please **Register as soon as possible** so we can accommodate appropriately. **Contact Teresa Cornette, FOD Rep (T3Cornette@aol.com) with questions or if you'd like to HELP with the planning!** **Kathy Stagni, OAA Director, is also helping our Group with daycare plans (oanews@aol.com or www.oaanews.org).**



Kids Korner



Emily (*MCAD*) and
Parker White



Megan, Jenny, John, &
Jane (*LCHAD*)
Carroll



Dr. Roe & Dr. Cook with...
Kelly & Kayla (*VLCAD*) Madej
Lisa & Rachel (*VLCAD*) Gibson
Gina & Brett (*VLCAD*) Revinsky



Alex Nawn (*MCAD*)



Candace Boyd (*LCHAD*)



Notes

New Jersey Autopsy Law

“In order to win the battle against SIDS and SUDC every state and country must enact comprehensive models of investigation!

On May 11, 2000 in the state of New Jersey, Governor Christie Whitman signed into law- **Senate Bill 661**. This landmark legislation **creates a system in New Jersey that will ensure comprehensive investigations of children up to three years of age whose deaths are sudden and unexpected. It is a model for the nation. New Jersey will be able to provide the most comprehensive investigations into these unexpected deaths.** It will benefit not only the families, but also the investigators and researchers who seek to better understand SIDS. From a parent’s point of view, this system compliments the care we give our children when they are living. It is thorough, open and clear to all those involved.

The **NJ Sudden Child Death Autopsy Protocol Committee** will include 13 individuals. They will represent all related fields of medicine; include law enforcement, as well as, parents who have lost children to SIDS and Sudden Unexplained Death in Childhood. The committee will **not only establish a comprehensive autopsy protocol, but also standardize the death scene investigation.**

This **legislation establishes the ability for a pediatric pathologist to work in conjunction with the Medical Examiner on cases of sudden and unexplained death in children up to three years of age.** This is the right thing to do. The specific expertise of the Medical Examiner and the Pediatric Pathologist, together, create the most comprehensive and ideal investigation for all.

Lastly, this law allows for parents to give consent for their child to participate in medical research. Very few states allow for tissue samples to be collected in cases of sudden unexpected death in infants. Thankfully, the rewards of studying even these small groups have already proven immensely beneficial. Significant research findings have located differences in the brainstem of some SIDS babies. This was discovered from the California SIDS victims. These babies were and are studied under strict protocols and under laws permitting tissue sampling for approved research purposes.”

[PLEASE refer to <http://crand.tripod.com/Legislation> for more information or contact Laura Crandall at Laura.Crandall@bigfoot.com for more detailed information.]

Metabolic Conference ~ Register by April 15th!
See www.pk-allieddisorders.org



Resources

Wisconsin First Step Hotline ~ provides statewide information and referrals to assist professionals and families who have a child age birth to 21 with special needs. Services available 24 hours a day.

1-800-642-STEP

Disability Resources, Inc. (www.disabilityresources.org) ~ Expanded ‘Disability Information at Your Fingertips’ Guide to toll-free and online disability resources. Covers a variety of topics, such as accessibility, arts, assistive technology, children, legal rights, etc. Cost is \$10.00. Contact Avery or Julie Klauber at info@disabilityresources.org or phone/fax 631-585-0290.

ACCESS ~ to help with disability questions and solve insurance problems. Chronic disease communities can call 1-800-700-7010. (I do not know if there is a fee for this service ~ DLG)

Hamilton Books ~ Discount book service at www.hamiltonbook.com



Family & Professional Donations

Family Donations: Joanne and Dan Bregman in honor of Maya (ETF), Patti and Chris Starkes in memory of Henry Rogan, Jr. and in honor of Rachel (SCHAD), Pam and Mark Held in honor of Claire (LCHAD), Barb and Robert Steiner in honor of Sadie (undiagnosed), Wendy and Chris Nawn in honor of Alex (MCAD), and Laura and Joshua Crandall in memory of Maria (undiagnosed).

Professional Donation: PerkinElmer Wallac's generous donation will help us with expenses for the May Metabolic Conference in Ohio (see enclosed Registration info). We **greatly appreciate donations** to help with copying, postage, and website fees. **Checks can be made payable to DEB LEE GOULD.** Please note in the check's memo section that it is for the FOD Family Support Group. Also, please be aware that we are not a non-profit organization, so 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

'It is not because things are difficult that we do not dare ~ it is because we do not dare that they are difficult.'

~A nonymous~



Reminders

Families: Please send **TYPED** stories by **May 1, 2001**. 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 **May 1, 2001**.