FROM THE EDITOR

This has been a most difficult fall and winter season for us all...please know that we have kept all of you that have experienced great loss and grief after the atrocities of September 11, 2001 and in our thoughts and prayers and will continue to do so. I am sure it will take many ‘seasons’ for those grieving to move toward the light of ‘healing’...yet, as we slowly move forward, life for many is forever changed.

As mentioned in previous newsletters, mailing our printed newsletter depends on available funding, but once again, we made it to your mailboxes! Unfortunately, because of the extremely high postage rates for overseas, all of our Families and Professionals outside of the US may need to access our newsletter online after this issue. We will try, however, to continue to mail out the Family and Professional Updates so networking can continue. There is still a possibility that we will be going completely online and discontinue the printed issue due to increasing copying and mailing costs – but for now please enjoy all the important information and stories we have for you in this issue. PLEASE SAVE the Family and Professional Updates because we won’t have another complete List until possibly July 2002.

We were planning on having a Medical Update article, as well as a Nutritional article for this issue, but due to a late cancellation and a health concern, those articles will be delayed and hopefully printed in our next issue.

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

Professionals ~ PLEASE let me know if you’d like to share your knowledge and expertise. We can always use more information and research articles or ongoing FOD studies on our website as well. Included in this issue is a ‘Professional Questionnaire for FOD Referral Purposes.’ It’s for ALL professionals ~ researchers, dieticians, counselors etc. Please take the time to complete this one-page questionnaire so we can update our files, even if you are already listed on the printed Professional List.

Trish Mullaley of the National Coalition for PKU and Allied Disorders announces that the next Conference (FOD Families/Professionals are invited) will take place October 3-5, 2002 at the Marriott in Orlando so mark your calendars! This is a joint Conference with Exceptional Parent’s World Congress on Disabilities. The registration fee of $60 per person will allow you to attend both conferences. Stay updated on hotel costs, agenda etc. by going to www.pku-allieddisorders.org. We NEED FOD VOLUNTEERS so contact me if you are interested.

Our new Email List (signup on our website) is going strong. After a rough start with another server, we settled at topica.com and overall are very pleased with how it’s running. It definitely keeps us connected across the world!

We are still working on possibly offering iron-on transfers for our Logo and the NBS popup picture (as seen on our website) that you can iron onto your own t-shirt. However, because of unusual circumstances that project is on hold right now. We’ll keep you posted. There is another project in the works, however, so be sure to read about the FOD Recipe Book in this issue.

What isn’t on hold is spreading the word about FODs and expanding Newborn Screening for these disorders (and several others). PLEASE make an effort and help bring awareness of these disorders to your family, friends, neighbors, co-workers and ALL the Professionals you can think of because…

‘We Are All in This Together!’

Take care... DLG
Dear Deb and Dan: Thank you for sending us (in England) the Family Packet of past newsletters. My name is Lisa and my husband, Gavin, and I have 2 daughters, Abbie, 4, and Lucy, 3.

In September 2000, Lucy was feeling poorly with a normal sickness bug. Suddenly she began to fit and had trouble breathing. I managed to get her to our local Dr who immediately called for an ambulance. By the time we reached hospital, no pulse could be found and her blood sugar level was at 0.2.

After a long 20 minutes the team of Drs managed to resuscitate Lucy but the only way she could breathe was with a Dr hand pumping air into her lungs. Thankfully, during a brain scan, Lucy began to show slight signs of breathing for herself. After a week in hospital (intensive care), Lucy came home.

The consultant that was with us at the time was wonderful. When routine tests came back negative, he explained that he wanted to test for MCAD. In his career he had come across 2 similar cases and knew a bit regarding MCAD.

He was right…Lucy had the 2 genes for MCAD. He then decided that we should test Abbie and referred us to Professor Leonard at Great Ormond Street Hospital. He also said we were very lucky Lucy survived. If it weren’t for Dr O’Keefe, we would not know about MCAD and would be waiting for it to happen again.

In March 1998, when Abbie was 18-months-old (and on the day I gave birth to Lucy), she was rushed to hospital because she was very ill. She was then transferred by police escort to a hospital one hour away. Abbie had menningicoccal septacemia and we were told that she would not survive, but she did! At Christmas 2000, Abbie also tested positive for MCAD. Looking back, I believe Abbie not only battled the menningicoccal septacemia but also the first show of MCAD.

We were devastated to know that Abbie and Lucy have MCAD after what we went through in 1998. Since last year, Lucy has been in hospital 4 times with sickness bugs etc.

Apart from our consultant (Dr O’Keefe) and Professor Leonard, we have yet to meet anybody who knows anything about MCAD. I find it difficult to explain to people what their condition is. Even our local genetics clinic had never heard of MCAD. So I gave them your website address.

I know we are very lucky to have both our girls with us! I was so relieved to actually receive your newsletter. There is nothing in England, no support groups etc., that I am aware of. We are due in Great Ormond Street London at the end of summer where I hope they will explain about carnitine.

We hope to be online soon so I look forward to reading more on your website. Thank you.

Lisa Sims
Wiltshire, England

Thank you for your note, Lisa. I hope you’ve been able to get online since this letter. I cannot encourage enough to sign on to our FOD Email List. Our List is so very important for emotional and practical support, especially when you are feeling alone in your challenges with MCAD...you are DEFINITELY not alone in this Group! Please also be aware that Lesley Greene at CLIMB (link on our Support Resources page) can probably connect you with other MCAD Families in England ~ we have several UK Families. Reach out and voice your concerns and I’m sure you’ll be heard and others will respond! DLG
The Survivor, The Hero, and the Angel
Trifunctional Protein Deficiency

The differences were evident beginning January 12, 1999 when James was born. He was my second child. Everything went well with the pregnancy, and I was healthy, so I never imagined the journey that lay before us. James was jaundiced. No big deal we thought, until we were sent straight to a special pediatric hospital. My husband, Jim, and I walked the halls of Children’s Hospital of Philadelphia (CHOP) in disbelief. We met super intelligent people, all who new about our 4-day-old son, and called him by name, “Sweet Baby James.” Test after test...hour after hour...“Rule outs” came from a long list of horrible could-be liver diseases.

James was diagnosed with biliary atresia, a condition that is a result of the malformation of the ducts that carry bile from the liver to the gall bladder. At the age of three weeks, James underwent a surgery to “correct” his condition by connecting a portion of his small intestine directly to his liver that, in theory, would alleviate his “plumbing” issues. (The operation is called a Kasai).

From February to June 1999, James received a lot of medical attention. James got an NG tube for nutrition due to failure to thrive, along with follow-up gastro visits for his liver. We watched his yellow color and monitored his sucking. We held his tiny arms for blood “sticks” and learned about hepatic function, liver panel, and bilirubin. Progress was only adequate.

At the end of July 1999, James underwent a hernia operation, and following, he lost the vast majority of his muscle control. He couldn’t lift his head. He couldn’t suck his bottle. He couldn’t grasp his hands and feet. The medical staff was confused because this hypotonia was not consistent with his “liver-related” issues.

As a result of not bouncing back like he should from the hernia repair, and with elevated liver numbers, the following week, James underwent liver exploratory surgery. The result, the doctors informed us, was that James’ liver was severely scarred and a transplant was eminent. Why hadn’t the Kasai worked? James had no reserve and after a second dose of morphine, he suffered respiratory failure. The feeling of powerlessness overwhelmed me as I watched the medical staff revive my son. There lay my infant boy, limp in the nurse’s arms and all I could do was pray. I hoped that God was with him to give him the strength to stay with us. I hoped that it WAS God’s will. I hoped that James would survive. James survived! Thank the Lord!

Our life of normalcy turned to that of hospital visits, tests, learning medical terminology, and worry beyond compare. We watched our son daily. We learned all his normal behaviors and could sense any dangers. We became pros at placing tubes, drawing meds, working machines. We became night owls...listening and distinguishing the nighttime bells. Our lives had changed.

In October 1999, James was having a bath with his sister, Gabrielle, then 3½ years old. I took James out, changed him, “hooked him up” to his feeding pump and placed him in his swing. (He loved his swing). About 20 minutes later, I heard a cry that seemed odd. I went to James, and grabbed him out of the swing. He looked like he was choking, so I pulled the NG tube from his nose. His breathing didn’t seem right, and as I yelled for my husband, Jim, James’ lips turned purple. James stopped breathing! I laid him on the bed and screamed out “Call 911”. Jim gave the info to the paramedics on the phone. I started giving mouth to mouth. I panicked, but could remember, “Tilt the head, back. Give quick short breaths.” But this was my son, my baby. “My God, please don’t let him die,” I thought.

After half a minute, James started crying, I knew this was a good thing. The rescue squad arrived and took over assessing James, and giving him oxygen. James was transported to CHOP.

This was the first of many hospital transports over the next two years. We became quite attuned to James’ signals. We knew when he was happy. We knew when he was hot (with fever) and not so hot. And we knew when he had to go (to CHOP). And go quickly! After many more tests, James was diagnosed with a fairly rare metabolic condition. Initially it was thought to be LCHAD, but now we know it is specifically, Tri-functional Protein Deficiency (TFP). It is a Fatty Oxidation Disorder, and gone undetected can cause liver, heart, retinal and muscular damage...even death. Treated with glucose and special diets, the child can live a healthy life. Many states are trying to expand Newborn Screening tests. And many families are trying to spread the word of awareness, since some pediatric doctors are not even knowledgeable about these disorders. And some, although aware, are not familiar with treatments.

James was placed on the liver transplant list as a Status One. We stayed in the PICU (pediatric ICU) of CHOP for eight weeks. We waited for James to come off the ventilator. We waited for him to be fever-free. We waited and listened to all the helicopters coming in the night. We waited for a liver! We prayed it would come. We prayed that someone’s misfortune would be our son’s blessing! December 29, 1999. The day had come! Jim and I left James’ sleepy head to grab a bite in the hospital cafeteria. It was about 12 noon when our nurse came to find us. “James will be getting a liver. They’ve got one. It will be today. We’ve got a lot to do. Yeah!!”

Oh how the chills ran up our spines...and smiles to our faces. Our miracle will happen. James will be fixed! He will be well again. We raced to the room and everyone prepared James. More blood tests, paperwork, and preparing meds. The nurses worked like a well-tuned machine. Everything was in order.

It was about 11:00 pm when everything was getting underway. The red cooler arrived. The delivery man said “God bless you.” The doctor said “…All looks good. Get rest. It will be a long night.” How could we sleep?! I hugged my baby and gave him kisses. “Mommy loves you. Mommy loves you,” I whispered. “You fight in there. You be strong.” The operating room door shut. And I latched onto Jim and cried in his arms. Will I ever see my baby again? Will he make it? What if there’s a complication? What if...?

The waiting room was cold. We sat in the hard light blue cushioned chairs for hours. Soda and snacks kept us awake. The silence, kept us asleep...in our fears, in our hopes. We waited for the updates. All was going well. And finally about 5:30 am, the doctor came before us. James made it! The new liver was in! “This is it. It’s all over,” I thought. He made it. He’s a survivor!

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Family Stories - The Survivor...(cont’d)

I surely was wrong that it was over. That day we held our baby’s hand and must have sung, “You are my sunshine” about 300,000 times. He looked miserable like he was beaten. He was puffy. He was weak. He had tubes and more tubes. His bandages covered his torso. But he had his new liver! Right? Early evening arrived, and with the doctor’s rounds came the blow. “The graft is not functioning”, he said. He repeated it. What the hell does that mean? I couldn’t believe what I was hearing. I didn’t understand. What it meant was that the liver was not working. The liver somehow didn’t survive! Maybe this and maybe that...but what it meant was that James was dying. After all the waiting...after the fight...after all those hours...“The graft is not working,” haunted my thoughts. I’ve got to be dreaming.

We weren’t dreaming! Reality was all but too true. James got placed back on the transplant list again ~ Priority in the country. And we waited, but this time only for hours. By the grace of God, a liver was born. James underwent his second transplant less than 36 hrs from his first. This time it was not an exact blood type match. Will it work? Will he live? Will he survive another surgery?

James survived. And as we slept most of New Year’s Day, we silently thought Happy New Millennium!

One week later, James was in severe rejection, and again underwent surgery. The surgeon needed to be sure the issue wasn’t clogged connections. They were fine. James’ body just was rejecting this foreign object. This foreign object that he needed to live!

On Jan 12, James turned a year old in the hospital bed of the PICU. We celebrated with balloons...with a cake...with a sedated child...and with an unlit candle that brought many tears. James was in pain. Our sweet Baby James lay silent because that was all we could do for him. Make him sedate. And pray. We weren’t sure if James would make it. And if he did, did his brain suffer damage? Would he be our James? The doctors and nurses were our therapy. God was our pillar of strength...and definitely watching us all.

James pulled through again. He was our survivor. Yet, at the end of February, James got a virus, “It’s EBV (Epstein-Barr Virus)...like mono. With tiders in the “millions,” they said. The quantitative number didn’t mean much except that it was really bad. “It will make him really weak. It’s like a lymphoma sort of thing.” We were beyond crazy. The tears and horror filled my head for days. I was frozen in time. I was still waiting for Thanksgiving. How could this be? He was supposed to just need a new liver! Right? What could be done? Something has got to be done! I wasn’t ready to lose my son. And James...he wasn’t done fighting.

“We could always get another liver, but we can’t get another body,” the surgeon said. And so we agreed to give up the liver. All anti-rejection drugs were stopped. We prayed for healing. We prayed his body would fight the virus and not the new liver. We waited. We watched. We prayed some more.

During James’ fight with “OKT3” a horrible drug that compromised James’ body, and breathing and life...my body ached with pain. Mentally and physically I was exhausted...I was having contractions...I was having another baby!

That’s right, Samuel, our third child was born, February 26, 2000. He lived in his stroller his first couple of months of life. He reveled at the beeps and lights and hissing of oxygen. He was a baby. He was also the recipient of a defective gene. Samuel also had TFP! I would take turns holding each of my boys.

Singing lullabies. Changing diapers. Placing NG tubes. Wondering what was next! Wondering what was God’s plan...wondering if we would all come home...wondering and standing still in time. Was it Spring? I had hardly noticed.

From February to June (of 2000), James (and Sam and I) remained at CHOP with a few interim “welcome home” days. We celebrated with balloons...with a cake...with a sedated child...and with an unlit candle that brought many tears. James was in pain. Our sweet Baby James lay silent...and  definitely watching us all.

Finally the EBV titers went down. We were on our way home. James was a survivor. James was a hero. “The first year is the hardest,” I remember everyone saying. They did warn us, and they were right. In a year the most consecutive time spent home was about 12 days! Imagine that. Two weeks then a hospital stay. We learned the ins and outs. We were regulars. We were a family with a chronically ill child. And in spite of all the chaos, our family made it. We stayed strong. We stayed secure and we stayed sane. (Well, almost.)

Then came February 2001. More hospital stays. Samuel had a scheduled admission for his g-tube placement and James followed him due to a routine cold. “He needs some IV glucose,” we thought, “48 hour rule out!” God had another plan. James had cardiac arrest. For 2 hours, we watched the team “treat” the code blue. Hours went by as doctors pumped drugs into James. The doctors prepared us that evening, “...We don’t think Jimmy will make it this admission.” The words struck like daggers to my ears.

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Family Stories - The Survivor,... (cont’d)

The shock and the panic consumed our hearts. “He just had a cold,” I remember shouting in tears. The familiar faces were all a blur. Some strange metabolic phenomenon…unknown causes. Unanswered questions. More worry and more prayers. And unbelievably, more miracles! More chances from God! James survived again.

It is now August 2001. And life is changing again. Our family has endured. And James continues to be a rock. The medical regimen has become our life. Hospital visits have become the norm. Tubes and pumps and therapy and oxygen…all just normal routine. James works hard at getting strong. He has good head control now and tries to bear weight on his arms. He can roll and he can babble. He speaks with his eyes and his smile will brighten any room. He has therapy daily and even takes swimming lessons. He is very proud of his accomplishments. His road ahead is long, but thanks to the miracle of organ donation he has a road to travel. Thanks to genetic research he can be safe for now. He has a chance for a good life.

And Sam has been saved by James. We are grateful for that. Sam is developmentally fine, but also has a nutrition (g-tube) regimen due to the lack of appetite, the inability to fast, and the lack of experience in chewing. He’s our crazy boy, and will surely be James’ best friend.

And Gabrielle. She is the angel in our story. Our outstanding daughter, Gabrielle, now 6, who is really all…survivor, hero and angel. She has fought through her brothers’ hospitalizations. She has fought through mom and dad whisking away almost weekly. She has fought through holidays, birthdays, vacations and celebrations in the hospital. She has learned about “Level 6” and how sick a child can be. She has been versed in medical apparatus that even some adults haven’t understood. Yet she is a happy wonderful child. She is secure and she loves both her brothers, although silently wishes for more attention. She plays “Barbies,” recites “Mary Kate and Ashley” and eats…not through a tube. She is mentally and physically healthy. Sometimes she even teaches us, and calms our fears. She is very deserving. She is an angel!

There is a story to be told of organ donation, genetic tests and special needs families with FOD (Fatty Oxidation Disorders.) Not to mention family, trust, parenthood and relationship! If I had the eloquent and thorough words of John Irving, I would write a book, but for now, I seek your writers, your editors. Help raise awareness. Help tell the story of Sweet Baby James…and the survivor, the hero and the angel!

“There is memory almost foreign.
There is fear, now routine.
There is hope.
And there is peace in being home.”

-MaryAnn Raccosta

Sincerely,
MaryAnn Raccosta@8/8/2001
jraccosta@hotmail.com

Family Stories - Michael and Crystal, CPT1

November 17, 1995 was a day we will always remember. Our son, Michael Wurz, who was 2 years old on November 13, became ill during the night with diarrhea. We let him sleep in the morning until 9 o’clock and when we tried to wake him, he was unresponsive and rigid. We rushed him to the hospital 40 miles away [we live on a farm 40 miles from town] where they did a little blood work and right away called an air ambulance to transport him to a bigger hospital 100 miles away. The doctors did a lot of blood work and different tests to determine what was wrong ~ the symptoms had pointed to Reye’s Syndrome, but with some inconsistencies, which led to some doubts about what was really wrong.

Our doctor, who was very kind and helpful and did his very best to treat Mike, did talk to us about metabolic disorders, but was unable to come up with anything other then Reye’s Syndrome. Mike was in a coma for 10 days and was not expected to live, but after 2 weeks he started to show some improvements. However, he was very weak, his muscles had deteriorated, and he was not able to hold his head up, much less walk like he was able to before. Mike, who was in the hospital for 4 weeks, started showing seizures before he was discharged, which he still has occasionally even though he gets medication.

Since we were told Mike had Reye’s Syndrome and had partially recovered, we felt all he needed was to recover his strength and he would be the same happy child he had been before…but we were wrong.

Three months later on March 13, after a single episode of diarrhea, he was back in the hospital with severely low blood sugars. After they were brought back to normal, he quickly recovered, but left the doctors scratching their heads. Even after still more blood tests, nothing new was discovered. After this we started monitoring Mike’s blood sugar levels that were normal except when he was not feeling well which caused us a lot of anxiety.

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Family Stories - Michael and Crystal, CPT1 (cont’d)

The doctors changed their minds about Mike's illness, when a year later on February 15, 1997, our daughter Crystal became very ill with some of the same symptoms that Mike had his first episode such as low blood sugars and weakness in her muscles. Tests were still inconclusive as to what was wrong.

After doing a skin biopsy on Mike in April 1997, which was sent to the Institute of Metabolic Disease in Dallas, Texas, it was discovered that Mike had a metabolic disorder called CPT 1 and because Crystal had had much of the same symptoms, it was concluded that she also had the disorder, which was later confirmed through DNA testing. There were only a couple of known cases of CPT 1 in the USA at the time, which explains why the doctors couldn't make a correct diagnosis. After her illness Crystal had a slight limp for about a year and her muscles seemed to be weaker in her right leg up to 2 years later, but now is healthy and strong and seems to be keeping up with her classmates. Mike's episode left him with seizures, poor muscle tone and developmentally delayed. He is going to school and seems to be learning, but very slowly.

In October 2000 both kids participated in a dietary therapy study at Baylor University Medical Center, done by Dr. Charles Roe and Dr. Jay Cook, which involved a low fat diet supplemented with a special oil called Triheptanoin. Testing done on both proved it to be beneficial. In school we are seeing very positive results with Mike. He is more attentive and is able to concentrate better but still has a lot of problems.

We have an older son, Patrick, who does not have the disorder and our youngest, Benson, does not have the disorder but is a carrier. In both Mike and Crystal the disorder didn’t seem to affect them until after their second birthday - between the ages 2 and 4 Mike was in the hospital 10 times, once with pneumonia, then he had to have his appendix removed. Not enough food intake usually led to hospitalizations, but since 4½ years of age, thankfully, we haven't had any illnesses where he had to be hospitalized. The disorder didn't seem to affect Crystal as severely as Mike, as she never got as sick as he, nor as often.

After a few episodes of 'hypoglycemia' we learned to watch them carefully whenever they aren't feeling well, and to try and keep their intake level up, which means getting up during the night and giving them juices or Gatorade.

The monetary costs have been very high for both Mike and Crystal, but the real heartache was seeing them sick and not knowing what was wrong or what to do about it, but as doctors learn more about these disorders, they should become easier to manage. The expanded screening that is now available at birth, for 30+ metabolic disorders, is something that every parent should be made aware of and requested for their child, for their own peace of mind and for their child's well being.

*(Please note ~ Mike and Crystal had diagnostic FOD testing done and it is a different test than the supplemental NBS test). Hopefully in the near future hospitals will be required to get this screening done on all newborns.

Ben and Debbie Wurz
Sunburst, Montana
406-937-3040 (Fax)

Notes

FOD Family Questionnaire
If you do NOT see your name on the Family List or on this issue’s Update, it is because I (Deb) never received the FOD Family Questionnaire that I sent you in the Family Packet when you first registered with us. If you would like to be listed for networking purposes, please go to ‘Online Forms’ on our website (www.fodsupport.org) and print out the Questionnaire. Then SIGN it and DATE it so I have your permission to list you. Please mail it to me via the regular mail (see page 1 of this issue for address) so we can list you in the next List Update.

Professional Questionnaire for Referral Purposes

All Medical/Health Professionals: Please complete the enclosed Questionnaire (even if you are already listed on our printed Professional List – this is an Update) if you would like Families to know that you diagnose, clinically treat and/or do research with Children and/or Adults with Fatty Oxidation Disorders. Please return to Deb Lee Gould via email (form on our website under ‘Online Forms’) or regular mail. deb@fodsupport.org or 805 Montrose Drive Greensboro, NC 27410
March 29, 2001 was the happiest day of my life. Not only did we have a wonderful little boy, Tommy, but early that Thursday morning I gave birth to a beautiful little girl, Nora. She was born two hours after her due date! All of the women in my family were born at the end of March, so Nora's birthday being on the 29th made it extra special. My husband, Jay, and I were ecstatic. My pregnancy had been easy. We did have an ultra sound and when we found out that we were going to have a girl, we both were so happy that we cried! Nora kicked so hard during the ultra sound, that the lady doing the procedure told me "Be ready and put your running shoes on for this one!!" I also had to go to have a non-stress test done a few times, because my doctor thought that Nora might be a little bit too small for her gestational age. When Nora was born, she was 6lb and 5oz, so she wasn't exactly a big baby, but then again both of our families are on the small side.

All that mattered was that Nora was as healthy as could be! She right away got an Apgar score of 9!

We could not have been any happier. Everyone kept telling us "Now you've got a perfect family, a boy and a girl." We felt so blessed! Tommy adored his little sister and Nora was crazy about Tommy. Soon Tommy learned that when he'd do something goofy in front of Nora, she'd laugh out loud. It was so much fun watching the two of them "play" together. I stayed home with our children and life could not have been better. Nora was an extremely easy baby. Immediately she started nursing really well. She hardly ever cried and was very happy and content. She started smiling early on and she shared her special smile with everyone who looked at her. Nora slept well and ate well. She would never cry when she woke up from her naps, she'd just make cooing sounds and when I'd go to pick her up, she'd smile to me. She loved sitting in her little swing and watching everything around her. When you held her, she liked sitting with her back against you, so that she could see everything that was going on. She was very alert. She loved being sung to...I had a special song that I used to sing to her every day and whenever I'd just sing the first few notes, she'd give me a big smile. When Jay was not working, he would help me with everything. He'd bathe Nora (she loved baths), change her diapers, bring her to me when I'd nurse her, etc. He loved his little girl.

Nora's checkups went well. Even though she was small when she was born, she quickly became 50th percentile in height and weight. The only thing that bothered me was that sometimes when I'd feed her, she'd have projectile vomiting. I often wondered about this. I read about it in a couple of books, and I even asked our pediatrician about it. But the fact is, that MANY babies do throw up. And it seemed that when I'd burp her more, she wouldn't throw up so much. And since she was gaining weight so well, nobody was concerned about it.

I never left Nora and Tommy for longer than an hour or two. My sister came to see them almost every day. Tommy and Nora love Tuula. I knew that the day would come that I'd have to go back to work, but I tried not to think about that. I worked part time, only six days a month, but my work would take me out of state twice a month, three days at a time.

On August 7th I was supposed to go to work for just a day of training. I did NOT want to leave Nora and Tommy. But I kept telling myself "You won't even be gone overnight. Before you know it, you'll be back home again." My big fear was that Nora would not take a bottle because I had been nursing her exclusively. We went through the same with Tommy. On that early morning I nursed Nora, kissed her and Tommy good-bye, told them both that I loved them, and headed to the airport. When I got there I found out that my flight to my destination was canceled. I turned around and came back home. I was only gone for about four hours, but that morning Nora had cried hard and refused to take a bottle from Jay.

When I got to the terminal to take my flight home, I called my sister again. This time she told me that Nora didn't seem to be her happy self. Tuula said that she just wasn't smiling like usual. Maybe it's mother's instinct, but I got a terrible feeling that something awful was going to happen. We kept thinking that maybe Nora just missed me, but we then decided that Tuula would call our pediatrician.
Family Stories - Nora, LCHAD (cont’d)

I got on my flight, but then found out that the flight was canceled because of a mechanical problem and it would take them an hour and a half to get another airplane! I was so upset! I called my sister again, and she told me that she was waiting for the doctor on call to call her back. By the time I boarded the new plane again, she still hadn't heard from the doctor...We found out later that the "operator" had given the doctor on call a wrong number!

As soon as I got off the airplane, I called home again. There was no answer. I was very upset and knew that something had happened. When I reached my car, Jay called me from the hospital, and told me that they had taken Nora to emergency. Upon arriving at the hospital, the emergency room doctor had asked if my sister was sure that something was wrong with Nora, because she looked so well. Suddenly Nora then became unresponsive. They rushed her to a treatment room and began taking her vitals. They could not figure out what was wrong. They thought that a CT might reveal what Nora's problem was. While all of this was going on, I was driving as fast as I could, trying to get to the hospital. It seemed like the longest drive of my life. All I could do is cry and pray...

Prior to the CT, Nora went into cardiac arrest. They attempted to resuscitate her for almost an hour. They were trying everything they could, but nothing worked.

Leaving home that morning, I never would have thought that Nora wouldn't be there when I returned and that I'd never get to hold my baby alive again.

We couldn't understand what had happened. We agreed to an autopsy. Our Pathologist was very thorough and with our connection to the Mayo Clinic, we quickly found out that Nora had died from a Fatty Acid Oxidation Deficiency ~ LCHAD specifically. We had never even heard of such a thing and had no idea what was ahead of us...

Our world was crushed and to add to the devastation we learned that Nora could have been screened for her disorder, and her prognosis would have been excellent!

Now every day we go to the cemetery to visit Nora's grave. We miss her more than we can say. It is so sad to see Tommy without his little sister. He talks about Nora a lot and we know that he misses her too. Every night I say a prayer in front of Nora's picture and tell her that I love her. In bed every night we cry ourselves to sleep.

Now we are very busy advocating Newborn Screening to other parents and not a day goes by that we don't ask ourselves "Why were we not informed about the screening?" If we had been, we knew that our little angel would still be with us.

Sirpa and Jay Waananen
JMWaananen@aol.com

Family Stories - Kyle, SCAD

My husband and I have been blessed with three wonderful boys. My oldest child, Matthew, was very healthy and met all developmental milestones early. My middle child, Kyle has been diagnosed with SCAD. Kyle was born November 22, 1993 by cesarean section at 38 weeks gestation due to my gestational diabetes. Kyle was 7lb 6 oz and appeared to be a healthy newborn. He was a poor eater in the hospital but the nursing staff and doctors just felt that he was off to a slow start. From the time we brought him home from the hospital, Kyle was very fussy. He cried inconsolably several times a day.

Within 3-4 weeks of life, he began the "projectile vomiting." It seemed that it would come up by the bucket full. Soon I learned that Kyle could not be laid flat on the floor or in bed because it would cause him to cry and vomit. His Pediatrician gave him medication for Reflux. Kyle spent the first year of his life sleeping half the night in a car seat and the other half of the night in an infant swing. Sometimes, he would cry all through the night.

It was obvious that he was having pain because he would pull his legs as close to his chest as he could. The Pediatrician wasn't concerned. In fact, she told me that Kyle was a "high needs child" and that I should wait 4 years before I had another baby.

Kyle had his first seizure at 9 months of age. I walked in the kitchen to find him blue and limp on my floor. The Pediatrician told me that it was breath holding. It was apparent that Kyle was lagging on his developmental milestones. When he was 12 months old, his pediatrician referred us to an early childhood intervention program. Kyle did not walk until 18 months old. His speech was seriously delayed. Kyle began receiving speech therapy at 2 years of age and continues to receive this service.

The "breath holding" continued and worsened. He was having "episodes" several times a week. It got to the point where he would turn blue, fall to the ground, make a funny noise while having a wide-open dead stare. Finally, when Kyle was 2 1/2 years of age, a new Pediatrician referred us to a neurologist. As we suspected, the diagnosis was seizure disorder. The EEG showed definite seizure activity. Kyle was immediately put on Depakote, which did control the seizures.

Continued on page 9
Family Stories - Kyle, SCAD (cont’d)

Doctors told us that the developmental delays, hypotonia and seizures were not in any way related to the other. He was diagnosed as having idiopathic developmental delay and seizure disorder. We continued to ask questions and press physicians. It didn’t add up that his problems were not related and we were not going to accept it as an answer. After all, there was not a history of developmental delay or seizure disorder in either of our families and how could a child have so many problems, none of which were related.

We noticed that Kyle fatigued very easily. He was our “couch potato.” When Kyle was 4, we went to a developmental physician for a second opinion. By this time Kyle had developed a very noticeable tremor. As a result of seeking the second opinion, Kyle’s neurologist told us about metabolic testing. Immediately, we said we wanted it. The neurologist told us that he was certain that Kyle did not have a metabolic disorder, as they are so rare and that the testing would be very expensive. We went ahead with the testing and one month later, on my birthday, the neurologist called to say the tests indeed indicated a metabolic problem.

Kyle’s alanine level was very elevated. As a result, the doctors believed that he had a mitochondrial disorder possibly involving the respiratory chain. When we finally found out that Kyle had a metabolic problem, we were 3 months pregnant with our third son so we were also very worried about whether or not he would be affected. Kyle had an open muscle biopsy and skin biopsy at Oregon Health Sciences University. Approximately one year later, we found out the diagnosis ~ SCAD. Kyle’s skin cells had been sent to Denmark, Amsterdam and Ontario and the diagnosis was confirmed.

Normal childhood illness such as colds and flu would last for weeks and leave Kyle with no energy. His tremors were more noticeable, and we would see an increase in seizure activity. School would report during these times that Kyle couldn’t function at school.

Kyle has always suffered from stomach problems. He was weaned off his reflux medicine when he was a year old. We thought the problem had resolved. Last summer at age 6 we had a pH probe done (insert probe down nose to tip of stomach) and found out that Kyle continued to suffer from severe gastrointestinal reflux. Because he was eating solids, which were heavier, he wasn’t throwing up like he did as a baby. Kyle had a Nissen procedure done and a gastrostomy tube placed in August of 2000.

Out of concern for Kyle’s declining health, I looked for other options. I found Dr. Roe’s study at Baylor University. Kyle began participating on my birthday in August of 2000. Upon admission, Dr. Roe found Kyle’s liver to be enlarged, his muscles weak, his eye movement abnormal and his body fat percentage to be abnormally high. Since initiation of the treatment, we have seen an increase in Kyle’s energy, and he is better able to fight infection. Kyle continues to suffer developmental delays. He is in regular education but receives special education services to help with academic tasks and he receives speech, occupational and physical therapy. Kyle is learning to read and enjoys playing with his friends. Kyle continues to suffer from occasional seizures, tremor, hypotonia and stomach pain.

I believe with all my heart that if Kyle’s disorder were detected earlier, through expanded NBS, Kyle would not have the severity of the symptoms that he has now. I am a strong advocate of NBS. In fact, the hospital in the town in which we live has agreed to pay for every baby born in that hospital (at no charge to the family) during the next year, to receive newborn screening, to be conducted by Baylor.

This is a step in the right direction. I know we are truly blessed when I say that my other two children are without symptoms of the disorder. Because the phenotypes for SCAD are not known, we are not able to determine if the other two children are carriers or unaffected. Hopefully in the future we will have that valuable information.

Michelle Miller
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FOD Recipe Book

Sharon Fisher is compiling low-fat recipes for an FOD cookbook, which will cost $6.25 each plus shipping and handling. Each book sold will help donate $3.00 (non-tax deductible, however) to the FOD Family Support Group and will help us with copying and postage costs. We still need more recipes as well as pictures of FOD kids to be used throughout the book and on the cover. Please include the following with your picture or recipe:

Name
Relationship to FOD person
Type of FOD
State or Country you are from

If you do not wish to have your name and information in the book it is still okay to send recipes and we will just write down anonymous contributor.

Contact Sharon Fisher for more information – 402-463-5699 or tyandsharon@hotmail.com
1. **Remember that your student has an FOD and is not an FOD child.** Just like all disorders, FOD does not make a child. As a teacher it is important to remember not to place labels on those students that have any kind of diagnosed disorder.

2. **Learn about your student.** As an effective teacher it is important that you learn everything that you can about your students. With a child who has an FOD this is even more important. Because every child may react differently to low blood sugars it is important to know what symptoms the child in your class may present and then watch for them.

3. **Remember that time is the key.** Because of the nature of these disorders, it does not take a child with an FOD very much time to go from well to deathly sick. If you notice a child beginning to show symptoms of an illness contact your school nurse and the parents. It is always better to be safe then sorry.

4. **Always have snacks on hand.** It is very important that a child with an FOD eat regularly. It is not always possible for the child to return to their desk and get their snack. By taking snacks along on field trips or outings away from the room you can be prepared even if the child is not (however parents should provide daily snacks to keep on hand in the classroom as well).

5. **Learn how to test blood sugar.** This is not an absolute necessity for all children with FODs as most are able to do this task on their own. But in some cases a child's sugar may become too low for them to perform their own testing. By knowing what to do you will not only feel more confident but you will also assure the parents that you care enough about their child to learn.

6. **Be flexible.** Because some children with FODs have a hard time recovering from illness they may miss more school than other children. Being flexible with these absences is key to helping these children succeed. Meet with the parents prior to the school year and develop a plan for such instances. If you are willing to work, then the child most likely will be too.

7. **Be respectful of illness.** Because of the nature of these disorders, it is very important to keep children with FODs as healthy as possible. By encouraging other parents with sick children to keep them at home you are not only protecting your students with FODs but also yourself and others.

8. **Be understanding.** Remember that a low blood sugar can make it difficult for a child to concentrate. If you notice a child having difficulty with concentration maybe suggest a snack before you write it off as daydreaming.

9. **Keep a medical protocol letter on hand in case of an emergency.** Every child with an FOD should be given a medical protocol letter to give to doctors in case of an emergency. This letter not only explains a little about the FOD that your student has but also gives specific directions for treatment. This letter can mean the difference for a child where time is the key.

10. **Don't have unrealistic expectations.** Instead of setting specific or unrealistic expectations of the children in your classroom offer encouragement and challenges. This way a child can achieve and succeed.

Sharon Fisher (mom to Isabelle, unclassified FOD, and author of *FOD and Me*)
Hastings, NE
[tyandsharon@hotmail.com](mailto:tyandsharon@hotmail.com)

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Welcome to new babies!

Jaiden Ann Alexis, sister of Alyssa (undiagnosed MCAD) and Austin (MCAD), was born on July 25, 2001. Proud parents and grandparents are Jennifer and Bill Boucher and Dorene and Ron Bellaire.

Elijah Joseph Cooper, brother of Noah (undiagnosed LCHAD) and Rebekah (unaffected) was born August 16, 2001. Sandy and Jon are very proud parents too!

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Deb’s Updated Email Address

Please update your address book —

My email address is deb@fodsupport.org or fodgroup@hotmail.com

And I also have an AOL Instant Messenger Screen name, which is FODGroup

In the next few weeks my AOL address may no longer be active.
It's a Tuesday morning and all is quiet in the house. Our 11-year-old, Nicole, has her own room and the 6-year-old, Taylor, should be sleeping in the bunk bed with her sister, 3-year-old Ashley, but because Ashley has MCAD I allow her to sleep in my room since I need to watch her so closely.

At six months of age she was diagnosed after a severe episode of hypoglycemia that resulted in a coma. At the time we had no idea what MCAD was, nor did we know my husband and I carried the abnormal trait that causes it. Ashley spent a week in the hospital with no answers until her blood work was sent to CHOP in Philadelphia, where they determined she had a fatty oxidation disorder called MCAD.

Since that time she has had numerous episodes of low blood sugar mostly due to poor eating or illness. About a year ago we discovered she suffers from seizure disorder and this probably stems from her first episode when she was in a coma. She is a sweet child with the brightest eyes you have ever seen and when she has an episode it is frightening to see her eyes change and her body go limp like a rag doll. She is so full of life most of the time that nobody can believe she has any kind of disorder.

We have experienced so much with her and we feel so bad that she has to have her blood sugar checked regularly and have a ‘special diet’ compared to our other children. Not only is it difficult for us as parents but also for our other daughters who have to see their little sister being taken away in an ambulance or witness cops and emergency personnel in the house when they wake up in the morning. The stress of having a child with health issues is very real. It is so hard to help our daughters understand that she must have a special diet that includes lots of carbohydrates and low fat items. Overall, they are really understanding, but when I think about how much they have sacrificed because of it...I get sad. There are times when we have had to leave the beach because it was too hot to keep her hydrated or when we go away and she does have an episode it shortens the trip and they give up their fun for the sake of her health.

Ashley receives disability benefits because she has had numerous medical bills we could not afford. My husband is self-employed and the cost of medical insurance was really expensive, which we could no longer do financially, without some help. I am thankful that we now have found doctors who know about this disorder because in the beginning there were very few who actually knew about it. We take her to her regular Pediatrician for simple matters such as cold and flu but she also sees an Endocrinologist at CHOP and a Neurologist here in our local area. At one point we met with a Nutritionist to meet her needs on diet and she also follows up with a geneticist at Robert Wood Johnson Medical Ctr.

Ashley takes seizure medication three times a day and we have tried about four different types along with various dosages...at times I felt like she was not going to benefit from the medicine, but we finally have found one that does not change her moods too drastically and she will be attending Pre-K this year hopefully without any illnesses. The last three years were really difficult for our family to cope with since we almost lost Ashley on more than one occasion. That thought is always in my mind and I constantly watch her closely. Everything changes when your child is diagnosed with a disorder. I am more conscious of things I took for granted before...like watching what she eats and paying attention to her in extreme heat especially when at the pool because she can suffer a seizure while bathing or swimming. All of these things cause added stress in our lives but the greatest gift we were given was to be parents and also the gift of KNOWLEDGE because without a diagnosis her life would be at risk daily.

I am sad that she has to endure all the tests and hospitalizations, but the fact that we know what to do now is the most important thing. Maintaining her diet is a small price to pay for a child's life. Anyone dealing with this issue knows how hard it can be, but knowing that some parents have lost children because they did not have a diagnosis makes me feel very fortunate. Since starting this, the girls have woken up and we will start a new day...sure hope it is a healthy day and un-eventful.

Lisa Cajuste
MCAD Parent
Keyport, New Jersey
pclc910@monmouth.com
My husband William was frantically driving me to the hospital at 4am. He wasn't just nervous because he was about to be a father. He was trying to get us to the hospital in one piece. Caleb Patrick was born at 5:20 pm during a really horrible tropical storm. My son blasted into the world as the storm blasted by us.

I will always remember September 14, 2001. That is the day I became a first time mother. It was the happiest day of my life. My son was finally here. He made it through all of my infections, severe preeclampsia (I am waiting to find out if it was FLOP or HELLP) and having the umbilical cord around his neck twice. I was the proudest mother ever. I had a fighter for a son.

I was only able to hold Caleb for a minute before he was taken to NICU. Caleb was placed in the NICU because he was born at 35 weeks and his blood sugar had to be monitored because I had Gestational Diabetes. I was so afraid to hold him for the first time. I was afraid that if I touched him that he would get sick. So all I did was lift up his blue knitted hat to see if he had hair. I told him "I love you and I will see you soon."

I was on bed rest for the next two days so I wasn't able to visit Caleb in the nursery. The nurses did however sneak Caleb in for a thirty-minute visit. That was the only time I was really able to be Caleb's mommy. I didn't have any staff watching me or listening to what I was telling my son. During that visit I told Caleb all of my hopes for him. I looked at every inch of his body. And yes I counted his fingers and toes. They were all accounted for so I was happy and at peace.

Every day William would come and visit us. I would sometimes get jealous. William would come in and say hi to me and give me a kiss then say well "I gotta go see my boy." Now, I am jealous because William had more time with our son. I am thankful that Caleb received so much love from his father. Caleb knows that his parents love him so much and will never stop loving him.

I was released on my fourth day. That is when I found out that Caleb wouldn't be coming home with us. I was then informed that he was being kept because he was a lazy po (by mouth) feeder. I wasn't even informed of this by his doctor. I was informed of this when I went to the nursery to feed Caleb.

I was always left in the dark by the first hospital. That night William and I attended Caleb's 6pm feeding. The nurse in charge of Caleb that night told us that Caleb was floppy earlier so she tested his sugar. It was at 20. She sent the blood to the lab and it was actually under 20. She said Caleb received some sugar water in a bottle and that he perked right up. Both my husband and I asked if they were still monitoring his sugar. We were told that it was not necessary. It was just an episode for my having Gestational Diabetes. I went on to explain to the nurse that I have a family history of Diabetes. She still said that there was no need.

That night Caleb took 20cc of formula. Over the week William and I would make our daily visits to drop off breast milk and to feed our son. During these visits we were not allowed to hold him until it was time to feed him, then we would have to leave right when he was finished. Needless to say, we never really got to bond with our son. I will never forgive the hospital staff for that.

I was allowed to change Caleb's diaper a few times. I loved it. I was actually allowed to take care of my child. During this time Caleb became a lazier po feeder according to the doctors. Caleb was put on an NG tube on his fifth day of life. He would start off on a bottle for 20 minutes, then what formula was left, would be given by NG.

On Caleb's 10th day of life everything was looking bright. We were present for his 9 pm feeding. Caleb took 29cc in 10 minutes. I made a comment to my husband "watch Caleb will be home this Friday or next Friday by the latest." I said this on a Tuesday night.

The following afternoon at 1:30 I received a call from the NICU telling me to come to the hospital. I didn't hear any concern in the doctor's voice. I asked if Caleb was being released. I was told no that Caleb had gotten worse. Of course I said "What?" I was then informed that during Caleb's noon feeding he became floppy in the nurses arms and that he was given oxygen. I said how is he right now? "He's still floppy, cold and pasty white." I called William home and we arrived an hour later. I have never seen a baby so white. We sat caressing our son until 6:30 pm. That is when I ran out of the nursery while they were bagging Caleb. I came back after he was stable to tell him that I loved him and to fight for mommy. I wanted to wrap my baby in a blanket and just hold him. He was so cold.

During this time I was informed that he was being transferred to a level three hospital. Caleb was transferred at 9 pm. We were informed that we would not be able to see Caleb until morning. I kissed Caleb good-bye and told him that I loved him. The transport team went one way while we went another. As we were heading to the parking lot the transfer team were coming out a different door. A little boy was walking in front of us, he said to his parents "Awe look at that cute little baby." I broke down. Praying to God "Please don't let this be the last time that I see my son." William and I sat in our car until the ambulance pulled away. I cry whenever I hear sirens.

Caleb survived the transport. I called that night at midnight to see how he was doing. I was told that he was hanging in there. And that he was receiving a blood transfusion due to anemia. I could hear my baby crying. At this point whenever someone would touch Caleb he would cry. He knew that another needle was coming. I asked if he was going to make it through the night. I was told yes and that I can visit him tomorrow. I was up all night just staring at the clock waiting for it to turn to 9:30 am.

We arrived at 10:30 to visit Caleb. The doctor in-charge of Caleb that day spoke to us. We were informed that Caleb might have an inborn disorder that affects the heart. We were then told not to expect Caleb to survive the night. I kept praying that God would spare my son and if it was not meant to be to please let my mom make it in time from Michigan so that she can hold her grandson. William's parents flew in from Texas. They were able to welcome their first grandson into the world and they were able to be present when he passed.

During our first visit Caleb opened his eyes three times. It was if he was telling me "Hey mom, I am still here." That was the last time I saw Caleb's beautiful blue eyes. I also noticed that his umbilical cord came off. I made the nurse find it. William and I were waiting for the day that it would fall off. Will used it as an excuse for not changing his diaper. The excuse was now gone. The cord and a lock of my son's hair are the only physical reminders I have.

Continued on page 13
**Family Stories - Caleb, TFP (cont’d)**

The second visit that we were allowed that day I just kept touching him. I kissed any part of him that didn't have and IV in it. Caleb had six total. And he was on a respirator. By the third visit the IV's all started failing on him. A central line was ordered but the surgeon never showed. At 8:30pm I had my son Baptized. It was something I felt I had to do. I wanted my son to be able to enter Heaven to be with my father. This way I knew he would be in good hands. Everyone in the NICU sang “YES JESUS LOVES CALEB.”

Visiting hours were over at 10:30. I was kicked out at 10:45. Caleb arrested at 11:00. They were able to revive him. I was told at 11:30 that this happened. I told the doctor that if it happens again that I want to be there. "I was there when he came into this world, I will be there when he leaves."

I asked if I could visit with Caleb. I was told “No,” that they were trying to stabilize him. Around this time a nurse from the first hospital came to be with us. Caleb was known as their little ‘Mr.Grabow.’ He touched so many in such a short time. We were talking when the doctor informed us that Caleb stopped breathing. I ran back in. My son was already blue. I had them stop what they were doing. I said, “Let my son go in peace.” They unhooked the last two IV’s and wrapped him in a blanket and placed my baby in my arms. **Caleb took his last two breaths in my arms at 12:45 a.m. It was on Caleb's 14th day of life.**

Afterwards I gave my son his first bath and the nurse from the first hospital combed his hair. She was the first one to do it and I wanted her to have the honor of being the last. I put my son in a diaper and dressed him for his next journey into God's hands.

The following day was Friday. The day I thought my son would be home by for sure. It was the day I had to plan my son's funeral. The day I had to pick out his urn. **I finally brought Caleb home on October 9, 2001. No mother should have to put her child's urn in a crib.** I kept telling Caleb I couldn't wait until he was home sleeping in his own bed. I will be crying for the rest of my life. I am just grateful that I was Caleb's mommy for fourteen days. Caleb we love you BIGGIE MUCH.

William and Shelly Grabow
Jacksonville, FL
Boo1974bear@yahoo.com

There are little eyes upon you
and they're watching night and day.
There are little ears that quickly
take in every word you say.
There are little hands all eager
to do anything you do;
And a little boy who's dreaming
of the day he'll be like you.

You're the little fellow's idol,
you're the wisest of the wise.
In his little mind about you
no suspicions ever rise.
He believes in you devoutly,
holds all you say and do;
He will say and do, in your way
when he's grown up just like you.

There's a wide-eyed little fellow
who believes you're always right;
and his eyes are always opened,
and he watches day and night.
You are setting an example
every day in all you do;
For the little boy who's waiting
to grow up to be like you.

(from: *Chicken Noodle Soup for the Soul*)
~author unknown~

Recited In Loving Memory on 9-29-2001 for
Caleb Patrick Grabow
Sept 14, 2001 ~ Sept 27, 2001
‘Angel In Heaven’
Our son Jack was born February 1997. We had some minor pregnancy complications in the beginning and then a rough C-section, but we were delighted to have him! He weighed 8lbs 2 oz and seemed very healthy, except for jaundice.

From the first night in the hospital, he seemed to have this cry that was a bit unusual. I remember him bringing me to me at night. He was sleeping in the nursery and was brought to me for feedings. I would awaken to this baby screaming and then a minute later, a nurse would bring in Jack. I remember thinking to myself, “Wow that poor, Mom.” Later, I figured out that poor mom was ME! It was just one of those cries that hits you like nails on a chalkboard!

We came home from the hospital still a little jaundiced, but otherwise no complications. Once or twice a day his cry would get like those nails on a chalkboard and I just attributed it to a little colic or just plain old baby cries. Jack would cry about an hour after being breastfed, and I just thought he was one of those babies that needed to suck. I would try and feed him and he wanted to suck, but was not hungry. He still was not sleeping through the night and my husband and I had many conversations about letting him tough it out, or at least not trying to feed him in the 2-3am wake-ups. And while he didn’t seem overly hungry he always ate and went back to sleep. Life went on and he grew quickly.

He walked at 8 months and was very alert. He babbled like normal but wasn’t interested in making many other sounds. When he was 1 year old, he was on bottles and table food, but we would have some mornings that Jack would wake up and just shudder with hunger. This continued for about 5 months. We spoke to the pediatrician many times about this unusual behavior. Our daughter who is 5 years older than Jack never had any of these issues so we were not sure. Our pediatrician assured us that maybe it was just a phase, but took some blood tests anyway. At one point he would guzzle milk out of a cup (almost 2 cups of it) and would be shaking, then ½ hour later throw it up. This happened maybe 6 or 7 times in a couple of months.

One Monday in October 1999, Lauren (by then 7 years old), came to me at 5am to inform me that her earring back had fallen off. Now this was unusual because Lauren is a great sleeper and normally would not wake up for such a thing. So I got up and escorted her back to her room to make sure she wasn’t sleep walking. On my way back to my room I heard muffled sounds coming from Jack’s room. Normally I would not dare open the door for fear that this child would rise for the morning and his sleep would be done, but something made me check. When I entered, the room was very dark (it was still dark outside) and cold. It was hot when we went to bed and the window had been open about 2 inches. I went over to feel Jack’s head and he was wet, like clammy wet. He was mumbling. I tried to talk to him, but no answer…just more mumbling.

This was very unusual. I lifted him out of bed and he was limp like a noodle. At this point I realized that something was wrong, so I took him into our room and woke up my husband. My husband put Jack on his chest. We could not get Jack to wake up. He had a cold so we thought maybe he was having difficulty breathing. My husband noted his clamminess and asked me to get the thermometer. I grabbed the Braun ear thermometer and tried it in Jack’s ear. It came up with an error. I told my husband that the batteries must be dead. He said no since he put it in his own ear and it worked just fine. So we put it on infant and sure enough it came up 93.7’. Just then he started to twitch and assume a fetal position. My husband said, “Call 911! He is having a seizure.”

When we arrived at the hospital, they drew lots of blood and were getting ready to do a spinal tap, when his glucose test came back. His glucose was only 20. So they started a glucose IV. The day held many other tests and a lot of waiting for us. Our son was conscious after a long-needed nap, which the doctors informed us was very normal after a seizure. He was released the next day, but the releasing Doctor consulted with an endocrinologist before letting us go. She ordered a couple more tests which were performed the next morning since Jack had to fast. Those tests came back with abnormal results. We really struggled to find a Doctor to help us. We were not getting very far with the Endocrinologist that had ordered the tests. We had to wait a month for appointments and Jack was still having some nighttime issues, sweats, and his temp dropping. The pediatrician just kept saying that the endocrinologist was handling it.

Thus, we called upon a great friend who is a pediatric ICU nurse at Fairfax. I faxed all the info to her from all his labs. She called upon some of her colleagues and their friends who finally put me in touch with a Dr who had worked with Dr Roe. She told me to go to my pediatrician and demand Jack be seen another Doctor. She called my pediatrician and spoke to her and told her, “This needs to be addressed. He might have MCAD.”

Ten months later, after his seizure incident, we got a clinical diagnosis. It had been almost a year since the first incident. We had to have a skin biopsy sent to Dr Roe at Baylor and they diagnosed Jack with MADD/Glutaric Acidemia Type 2.

What seemed so unusual to us was first, finding a Doctor that had any experience with these things and second, all the doctors were confused at how late he presented with MADD (2½ years old).

Jack takes Riboflavin 3 times a day that we have compounded at a pharmacy and Carnitor® 3 times a day. Other than making sure he has a heart-healthy diet and his glucose levels stay high, he lives a normal life. We have to be careful of course when he becomes sick and when we travel we monitor him much more closely. We just make sure we have lots of Gatorade and jelly filled granola bars for him to snack on. We are truly blessed that he has a mild case and that there was no permanent damage done with his one incident.

Thanks for reading our story and for your support,
Jack, Lisa, Lauren and Mark Spraggins
markandlisaspraggins@home.com
Love Messages

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

Sandy and Jon Cooper

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

David and Amy Deshais

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

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

Shelly and William Grabow

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

Nicole and Chris Gulinello
Alec - Birth Feb 21, 2001  Death Aug 24, 2001

Michael and Nicole Gumiela
Michael - Born March 28, 1998 Death April 4, 1999

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

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

Ralph and Angie Hedrick
Chelsea - Birth Jan 11, 1995 Death Apr 3, 1996

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

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

Brad and Kim Holmes

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

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

Lisa and Pete Leonardi
Devlin - 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

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

Randy and Misty McDonald

Christine and Mark McFarland

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

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

Lori and Jeff Michaud

Simone and Michael Miller

Mike and Sheryl Mulhall
Justin - Birth April 22, 1990 Death April 22, 1990

Verna Parker

Diana and Kevin Patterson

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

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

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

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

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

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

Brian and Cherryl Rosenberger

Janice and Steve Rowland

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

Jackie Shears

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

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

Anne and Gary Stitt

Lisa and Doug Tennyson

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Coping & Healing

Our Connection in Spirit...

How important is it for humans to be connected with others while facing challenges in life? That may seem to be a simple question with a simple answer, but let me tell you, it’s far from simple.

Many of us strive to connect with other families and professionals across the world dealing with these disorders, but there are some that choose not to. I won’t guess at the reason for that or sternly judge anyone if they don’t reach out to others, but what I CAN say is that SUPPORTING EACH OTHER IS VITAL in ALL aspects of life, not just when dealing with a rare disorder. Yet, you can’t be supported or even perceive that others are offering support if you’re not open to receiving it.

The FOD Family Support was not created to provide ALL the answers or invincible and proven suggestions to families. Although we were painfully ‘birthed’ due to NOT having knowledge and answers about FODs, we chose to focus on offering the intangible and often ‘inarticulateable’ dimension of connection and emotional support…a connection that crosses all boundaries of space, time, and all other human characteristics and systems (i.e. race, religion, politics etc) ~ a connection that is also presently being demonstrated so poignantly by men, women, and children across the world after the September 11, 2001 terrorist attacks…PEOPLE NEED PEOPLE!

[As a personal aside…It was encouraging, as well as inspiring to see so many men, whom often may feel in our society, that their masculinity is threatened if they express the emotional part of themselves, to allow themselves to fully feel and to hug another experiencing such horrific pain instead of cognitively ‘staying in their head’ in order to cope with the totally incomprehensible events. It was and is an extraordinary example of how HUMAN SPIRIT and FAITH, HOPE and LOVE can be the salve that will ‘heal one’s fractured heart’ over time and with much grief work. I HOPE that men, as well as women, boys and girls, continue this ‘breaking of the shell’ ~ it makes for a more REAL and TRUTHFUL and FULFILLING LIFE!]

Just as there is a wide and varied spectrum of how children/adults MEDICALLY present and respond to an FOD, there is a similar spectrum associated with how individuals/families EMOTIONALLY cope when faced with a rare metabolic deficiency.

A while ago, I spoke with a new FOD mom that wanted to hear that things definitely would be okay and I couldn’t 100% guarantee that. All I could offer was that most of the time once a diagnosis and specific treatment begins, and IF it’s earlier rather than later, then the kids usually do okay and create their own kind of ‘normal.’ Then on the emotional level, it’s up to the families to work hard at living and coping WITH this challenge instead of fighting AGAINST it and themselves.

I mentioned the spectrum of how these kids present and gave examples of how some responded when diagnosed/treated from birth versus later on (i.e. refer to our Cost Benefit Analysis on our website’s NBS page), some experiencing residual medical complications with seizures, gtubes etc., and the anxious mom said, ‘I don’t need to go there’ ~ meaning she didn’t want to hear anything other than positive things…she didn’t want to hear REALITY!

I wish I could be ALL ‘positive’ when talking with new families, but that’s not real life! Yet, being positive is a very relative state depending on your own context – to me, just GETTING a diagnosis is positive compared to having none!

Life is full of challenges and having a child (or yourself) with a disorder can present a family with spiraling ups and downs over time. Hopefully, you’LL CHOOSE to PROACTIVELY take on these challenges rather than be frozen in denial and wish or pretend they didn’t happen to your life or your child’s life!

Soon after talking with this mom, I had another call from a new family that demonstrated this proactive approach. They had just gotten a diagnosis the day before and were already researching information on the Internet and making calls to other FOD Families with the same disorder.

Continued on page 18
Coping & Healing (cont’d)

I also mentioned to them the wide spectrum of presentation and they acknowledged that possibility instead of dismissing it as the other family had… I have yet to hear back from that first family. That’s frustrating, but I can’t make decisions for people… they have to CHOOSE for themselves how they are going to cope and deal with what’s presented to them. Some unfortunately prefer the ‘head in the sand’ way over the I Am/My Child IS WORTH ADVOCATING FOR attitude!

It’s not about me, or anyone for that matter, stating cold hard facts in an in-your-face way (i.e. “Your child has a FOD, deal with it!”) – it’s about CONNECTING COMPASSION and REALITY… I call it Compassionate Realism.

It’s about discussing a family’s particular situation and what the facts are (i.e. results of tests, definite diagnosis, medical concerns etc), as well as combining that with ‘listening for and hearing’ not only the spoken emotions and fears, etc., behind the diagnosis and ‘coping’ with it, but also the unspoken ones…the ones that carry a lot of power…at least until they are voiced and validated and expressed in a constructive way.

This is where the importance of SUPPORT comes in… instead of struggling in silence, children/adults can connect and share those emotions, thoughts, and doubts etc., with other families walking a similar journey ~ diffusing some of the power of those unspoken fears. It can’t take away your own personal and family challenges, but it sure does help to NOT FEEL ALONE on your journey.

For those that are ‘closed’ at this time to support or connection or facing reality TOGETHER rather than alone, I will say… there is always HOPE that someday you will feel safe enough to face your/your child’s challenges HEAD ON and TOGETHER with others in our Family Support Group or with your family and friends… just KNOW that we will be here when you are ready to experience that vital and vitalizing connection in spirit!

Deb Lee Gould, Director
FOD Family Support Group
September 2001

Pharmaceutical Update

Sigma-Tau Pharmaceuticals, Inc. (makers of Carnitor® ~ www.sigmatau.com)
New Fax: 301-948-2049

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

Resources

Mercy Medical Airlift (Ed Boyer, President) recently arranged a very deep discount airline ticket program that CAN BE USED FOR CONFERENCES (concerning your/your child’s medical condition etc). This can save a lot of money: IF TRAVEL TO THE CONFERENCE IS LESS THAN 1,000 MILES, the travel can be for free through the Angel Flight America network (service provided by private aircraft). Access to this type of help through the National Patient Travel Helpline on 1-800-296-1217 or www.raredisseasetravel.org.

For patient travel, the number to call for help with free or deep discounted (for over 1000 miles away) airline tickets for patient travel (in the National Patient Travel Center) in Virginia Beach, VA, is 1-888-675-1405.

Also, the National Association of Hospital Hospitality Houses (1-800-542-9730) provides lodging assistance for persons receiving medical treatment away from home.


NBS Updates, Email Discussion Lists, and State-by-State Networking ~ see Save Babies Through Screening at www.savebabies.org and the National Coalition for PKU and Allied Disorders at www.pku-allieddisorders.org. The Coalition has recently formed Email Lists for almost all of the states so families of various metabolic disorders can network face-to-face and possibly work together on special issues regarding their own state (i.e. insurance coverage for formulas, NBS, etc.). It’s just another way for individuals/families to connect for not only emotional support but for raising awareness and promoting PRO-ACTIVE ACTION on these very important issues.

Next Metabolic Conference jointly sponsored by the National Coalition and Exceptional Parent’s World Congress on Disabilities will be October 3-5, 2002 at the Marriott in Orlando, FL. FOD Families and Professionals are invited! Stay updated by going to www.pku-allieddisorders.org.

5th International UMDF Symposium on Mitochondrial Disease for mitochondrial specialists, general practitioners, and families will be June 6-9, 2002. Registration begins in Feb 2002. Families contact UMDF office at 412-793-8077 for Family meetings (June 7-9) and medical professionals contact the Univ of Texas Southwestern Medical Center at Dallas at 1-800-688-8678 or www.utsouthwestern.edu.
Kids Korner

Nicholas Baer, SCAD

Zachary and Nicholas Leonardi, unaffected brothers to Devin, undiagnosed MCAD

James Raccosta, TFP

Lucy and Abbie Sims, UK MCAD

NBS Update

Mayo Clinic is now performing expanded Newborn Screening. They also perform FOD Diagnostic testing and consultation. All inquiries for Lab testing/interpretation for an FOD (they test for other disorders as well) should be directed to Dr Piero Rinaldo and Dr Matern (and staff) at 800-533-1710. Those seeking a clinical evaluation at Mayo for an FOD can contact Dr Jerry Vockley or Dr David Whiteman at 507-284-8198.

Mayo Medical Laboratories
Mayo Clinic Biochemical Genetics Laboratory
Supplemental NBS Testing/ Diagnostic Testing/ Consultation
Phone: 800-533-1710
Fax: 507-284-1759
Co-Directors: Dietrich Matern, MD, FACMG (matern@mayo.edu) and Piero Rinaldo, MD, PhD, FACMG (rinaldo@mayo.edu)
Website: www.mayo.edu/bgl (click List of Tests then ‘S’ for Supplemental Newborn Screening)
Family & Professional Donations

**Family Donations:** Julie and Dave Wolin in honor of Jacob (MCAD), Chris and Tom Doughty in honor of Scott, Libby, and Jill (all MCAD), Connie and Barry Immink in honor of Jenny (MCAD), Robin and Vince Haygood in memory of Ben (undiagnosed MCAD), Pauline Hill in memory of Rosemarie Rees (adult daughter, undiagnosed MCAD), Wendy and Chris Nawn in honor of Alex (MCAD), and Pamela and Eric Wilson in honor of Jacob (SCAD).

**Professional Donations:** Scott Palubiak, Vice-President of Marketing and Sales representing PerkinElmer (makers of tandem mass spectrometers).

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

Thank You

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.

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

Reminders

**Families** - Please send **TYPED** stories by **May 1, 2002.** 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, 2002.** Also, please return to Deb the enclosed **Professional Questionnaire** even if you are already listed on the printed Professional List.

‘Without a sense of caring, there can be no sense of community.’

~ Anthony J. D’Angelo ~