Happy New Millennium! Now that the holiday ‘chaos’ is over, it's time to make some Goals for the New Year! It would be great if we could ALL include educating others about FODs as one of our goals for this year. Several members of our Group have been active in getting information out to laypersons and professionals about the benefits of FOD Newborn Screening. State Legislatures are beginning to take notice, but we still have a lot of educating and advocating still to do. Examples of letters to write to your legislators or state health boards/committees have been in past newsletters as well as on our web page. Wendy Nawn (wendy@savebabies.org) has been extremely busy writing and talking with newspaper, magazine, and TV people, as well as sending out letters to family, friends, and professionals. It’s that kind of activity that will get these disorders brought into the forefront. The KEY is to spread the word to anyone and everyone!

There are many different ways of doing that ~ telling your own Family Story, for example, at your local hospital's Pediatric or OB/GYN rounds for new physicians is one way to make these disorders REAL to professionals that haven’t heard about them yet. It puts a face ~ YOUR face ~ on a medical reality! Every effort to get the word out will benefit our present and future children! Speaking of Family Stories, it would be great if those of you that are dealing with some of the more rare FODs (i.e. SCAD, HMG, Carnitine Transport etc.) would share your personal experiences. Some of our newer families are searching for practical information from families already dealing with these disorders, but they have not been able to find much. So please think about writing your own Family Story. It would not only be informative, but emotionally supportive. I'm sure all of you can relate to what it felt like when you first got an FOD diagnosis! Please send your Stories by May 1, 2000.

Professionals (MD, PhD, RN, RD, MSW, etc) ~ we don't want to leave you out! Your support is vital too. Letting us know updates on your FOD research or clinical work with families will help us all understand FODs better. Please send any updates/articles to me (DLG) by May 1, 2000 and we can put them in the printed newsletter and/or on the web page. We need your help, so please consider sharing your expertise with us.

Family and Professional Lists ~ PLEASE SAVE! In order to contain our copying and mailing costs, the January issue will be the ONLY issue that we send entire Lists. The July issue will only contain a small listing of NEW Families and Professionals and then they will be added to the major List the following January. I have also included in this issue our new FOD Pamphlet and Card ~ please feel free to copy and/or share the information (*in 2000, our pamphlet is in a pdf file on our website under Online Forms).

While we are in the process of changing to a new web address (www.fodsupport.org) our ‘old’ address will still be in effect for several months. Once again, THANK YOU Sigma-Tau Pharmaceuticals, Inc., for helping us to finance our online and printed materials!
Our Email List has been extremely successful as another avenue of support. If you haven't already signed up, please do. Support of all kinds is INVALUABLE! The many responses families have had truly show that

‘We Are All In This Together!’

Take care…
Deb and Dan Gould, Co-Editors
fodgroup@aol.com
336-547-8682

Letters to the Editor
(Letters/Articles from Professionals/Researchers are ALWAYS welcome too)

Dear Deb:

Sorry this letter has taken so long to write, my oldest daughter Fallon suffers from Cystic Fibrosis, and she has been home now for five weeks with a serious chest infection. Reuben also had his first hospital admission a fortnight ago, since his MCAD diagnosis in September 1997. We couldn't find anything wrong with him, he just didn't want to eat or drink for two days so they had to put a drip in. He's recovered fully, and now back to his old bubbly self. It's been wonderful reading your newsletters, and realizing how many others are in the same situation as us.

We couldn't get very much information in New Zealand and we were very worried about what the future held for our son. We certainly feel a little happier now, which makes us better to cope with most day-to-day situations. A few things we don't understand are, why Carnitine is continually taken by a lot of children in the newsletter and why a low fat diet is recommended? Reuben was given Carnitine for one year. We have been told the normal level in his blood test should be 23 to 60. Reuben's was 14 but unless it drops under 10 they will not put him back on to Carnitine. The only thing we were told to do as far as diet went was to make sure Reuben eats more carbohydrates than normal. He has a carbohydrate powder in his milk called Polycal at bedtime, which we think helps keep his blood sugar level over night. We have noticed his stools looks very fatty but we've never known why. Maybe if we lowered the fat content of his diet this may help. I'm sure in time to come, we will learn more and more. Looking forward to our next issue.

Karen Gill
42 Litchfield St
BLENHEIM New Zealand
gill.construction@clear.net.nz

Dear Deb:

I was reading the July newsletter today when I read Megan's story. I noticed that her parents mentioned not being able to find riboflavin in liquid form. My son, Tristan, takes riboflavin also. The way to get it into liquid form is to find a compounding pharmacy in the area. Our compounding pharmacy is a franchise of the Medicine Shoppe. You can ask your doctors if they know of a compounding pharmacy. Our pharmacist has been very helpful. I hope this information is helpful to those using riboflavin. Thank you.

Beth Morley
tbmorley@naxs.com

New Metabolic Disorders Foundation formed in memory of James William Lazzaro (VLCAD)

Wednesday, August 13, 1997 started out just like any other day except that, our youngest son,
James William happened to awaken before I left for work. Little did I know that the kiss I gave him that morning would be my last. Later, at lunch, my heart skipped a beat as I recognized the number on my pager to be our special code for a household emergency. As I raced home, the knot in my stomach just kept getting worse as I pictured what our babysitter had just told me – our precious 8-month-old baby was fighting for his life.

When I got home, I couldn't believe my eyes, as our little baby lay lifeless on the living room floor, still in his pajamas, being attended to by the paramedics. The ride through our small town seemed to take forever as the ambulance made its way to the emergency room. The attendant at the hospital told me that “it didn't look good,” and my wife, Jamie, immediately broke down. After what seemed like another eternity of prayers, and more tears, a team of people came in and gave us the news that our little baby Jim was dead. When the minister asked if we had a preference for funeral arrangements, I felt as though somebody kicked me in the stomach, and was standing on my chest. How could this be happening? Jim was a healthy, normal 8-month-old.

The days and weeks that followed all blur into a painful collage as friends and co-workers cautiously tried to comfort us as we started to try to resume our shattered lives. Certainly, there was little time to feel sorry for ourselves, with a 2-year-old running around, but my wife and I were still searching for some sort of answer. After about 3 months, the pathologist called to review the autopsy findings. While her thoroughness and professionalism were obvious, the conclusion of Sudden Infant Death Syndrome (SIDS) still left my wife and I feeling empty.

The autopsy showed some fatty lipid deposits in the liver, but since SIDS is an exclusionary diagnosis, there was nothing to conclude. At this point I began to research anything and everything associated with SIDS, and stumbled over an article on a fatty acid disorder known as MCAD. The symptoms seemed to match with Jim’s autopsy, and what was worse, there was a 25% chance of recurrence in siblings if this was in fact the cause of death. I phoned the pathologist and questioned her about all of the information. Although she was unfamiliar with Fatty Acid Oxidation Disorder, she was more than willing to see if this was, in fact, the real cause of death.

Subsequent tests at Baylor Medical Center confirmed that Jim had died from VLCAD. This finding was even more significant to our family as we were expecting our third child, and the very thought of losing another baby at times was overwhelming. Thankfully, our daughter, like our oldest son, was born without the genetic defect that took her brother's life.

In memory of our son James William we have established a non-profit, private foundation to reduce the number of infant fatalities due to genetic metabolism disorders by focusing in four key areas:

- **Awareness**: According to a poll conducted by the ABCD group, as many as 450,000 children could be suffering from undiagnosed metabolic disorders since many physicians and parents are not aware of the symptoms or conditions. The foundation will work closely with the medical community to raise awareness without creating undue stress for expectant parents.

- **Diagnosis**: Since many of the Genetic Metabolic Disorders are easily treatable with diet modification and/or various supplements, discovery of the defect at birth is critical to reducing infant fatalities. The JWL Foundation will partner with such organizations as the Fatty Oxidation Disorder (FOD) Family Support Group to promote mandatory newborn screening.

- **Treatment**: As highlighted above, many of the Genetic Metabolic Disorders are treatable through diet modifications or supplements. The cost-effective distribution of special infant formula and support for parents of children with metabolic defects will also be a priority for this foundation.

- **Prevention**: Like any disorder, the most effective treatment is prevention. The fourth and last component of the JWL Foundation will be to help fund research related to Genetic Metabolism Disorders. It is our sincere belief that by focusing on these four elements, and, with your tax-deductible support, the James W. Lazzaro Foundation for Genetic Metabolic Disorders will successfully establish mandatory newborn testing for metabolic disorders. While nothing will bring back our 8-month-old son, in his memory we can prevent more families from suffering the terrible loss of a child. For more information, contact us at:
Ysabel’s Story ~ LCHAD

What an incredible miracle girl we are blessed to have in our lives! And we are also blessed to have the love, support, and prayers of each of you, especially through the last seven weeks. This is Ysabel’s story, a story of God’s faithfulness and answers to each of our prayers.

Ysabel was born at 3:17 PM on September 26, 1999. Of course we were thrilled and could not wait to take our daughter home. We arrived home on the 28th and by the morning of the 29th we knew something was wrong. In the middle of the night/early morning, Ysabel was breathing very strangely and wheezing. On the advice of Kaiser, Gonzo took her into a steam-filled bathroom. This seemed to help. He was advised to keep her upright and to call at 7:30 am for an urgent care appointment. For two hours, Gonzo held Ysabel on his shoulder while she slept. She was sleeping so well that he almost put her back into her bassinet. Something kept him from doing that, even though he was starting to fall asleep himself.

About 6:00 AM, he felt a "gurgling" in her back. He started to pat her, thinking this would help her cough up whatever was causing her problems. Nothing came out, but she did begin to cry. At this point, Lisa heard her and told Gonzo that she needed to breastfeed. Ysabel was so weak she could not latch on. She was also lethargic, floppy and very cold. Her head felt like ice, despite the fact she had been wrapped in a blanket. After another call to Kaiser, Gonzo decided that she needed to be taken to the ER. At this point, we were still feeling like paranoid new parents. We arrived at the ER within 4 minutes. The staff took one look at Ysabel and put oxygen on her. She started to turn blue before our eyes and was whisked away by the nurse. A few minutes later, the nurse came in and suggested we call our minister. That is how we found out we might lose our baby girl we waited so long for.

A lot of the next few hours is still a blur for us. We were told that Stanford had been called and that they would transfer her to Lucille Packard Children's Hospital (LPCH) as soon as she was stabilized. It took close to two hours to stabilize Ysabel. CPR had to be performed and veins had to be found for various medications and blood. Once she was stable enough for the ride, she was placed in a special transport unit with a respirator, and various monitors for heart rate, blood oxygen level, etc.

We were able to quickly say good-bye to her while she was in this box with tubes and blood on her. It was the last time we saw both eyes open for the next six days. The drive to Stanford was terrifying for us. Neither of us were allowed to ride in the ambulance and we were unable to follow because of the speed they were going. Several of you received frantic phone calls from us during this ride. We arrived at LPCH to hear our daughter was critically ill and no one knew what was causing her to be so sick. The staff discussed possibilities such as a severe infection, heart and lung problems or a metabolic disorder.

We were finally allowed to see her once they had everything set up; an oscillating ventilator, monitors and several IV's was any parents’ worst nightmare. Ysabel had to be artificially paralyzed so that she would not fight the ventilator she was on. We felt so helpless, powerless and of course were blaming ourselves.

In the ensuing days, Ysabel was having seizures, and having trouble regulating her own glucose level. She was heavily medicated on painkillers, three seizure medications and antibiotics. She was having chest x-rays every six hours and required at least one blood transfusion, as well as constantly having blood drawn to check levels of all kinds. By the 3Oth we had a preliminary diagnosis back from Duke University.

There was a chance that Ysabel had a very rare genetically passed metabolic disorder called LCHAD ~ LCHAD is acronym for Long Chain 3-Hydroxyacyl CoA Dehydrogenase Deficiency. In a nutshell, Ysabel’s body does not produce the enzyme that metabolizes long chain fat within the mitochondria of all body cells for energy production. This diagnosis was confirmed the following week, and further confirmed when our DNA was tested. The three of us have the common mutation of the gene. LCHAD is passed to the child from both parents. We each have one good copy of the gene and one bad. Both our bad copies are in Ysabel.
By October 3rd we felt comfortable enough with Ysabel's condition that we actually slept at home that night for the first time since the nightmare began. At this point Ysabel was on a respirator and just beginning to show signs of waking up — we were thrilled by her movements and responses to stimuli. On the 4th, she opened her eyes for us. On the 7th, she pulled out her own respirator tube and on the 10th she nipplefed from a bottle for the first time. Ysabel's MRI on 10/9 was abnormal. It showed that damage occurred consistent with a lack of oxygen for an unspecified period of time. What that means is only time will tell about her development.

Currently we are thrilled because she has normal infant behavior. On October 13th Kaiser transferred Ysabel to their facility in Santa Clara. While she was stable enough to transfer we still had obstacles to overcome. The doctors needed to find a formula that Ysabel could tolerate in quantities large enough for her to grow. Also she had three detectable seizures the first week she was at Kaiser, so her Phenobarbital dosage was increased. Finally, by October 22nd Ysabel was started on a formula that she could tolerate. The formula contains 25% breast milk, Provimin, Polycose, MCT oil and water (Lisa makes the day's formula each morning. It requires some fancy measuring and a variety of kitchen tools).

Over the next week the amounts were gradually increased and she was weaned off the IV nutritional supplements. Also during that week there was no visible seizure activity and the EEG done two days before she came home also showed no seizure activity. Finally on October 28, we were able to bring our baby girl home! Yes, we were terrified the first night but we have finally settled in. Ysabel remains on Phenobarbital as we don't know if the seizures were due to the insult to her body and brain or are part of the metabolic disorder. If Ysabel remains seizure-free, the doctors will try to wean her off the medication by letting her outgrow her dosage. Ysabel is also taking carnitine, a supplement that helps the body with fat in the mitochondria and removes toxins. The primary treatment for LCHAD is a modified diet, low fat, high-carbohydrate, with no sustained periods of fasting. The other element vital to insuring she does not have problems is to keep her "well", i.e. avoiding as many viruses and infections as possible. If Ysabel gets sick this could affect her appetite or the way that her body metabolizes her food. We have already been warned and have faced the reality that if Ysabel does get sick she will be hospitalized. Ysabel is one of the youngest children to be diagnosed with LCHAD; her age of presentation" is atypical — yet another reason for her doctors to track her closely.

We have now had her home for three weeks. We are in awe of her will to fight, how far she has come and her smiles! To look at her now, you would never believe what she has been through. She brings us joy each day and we are having a great time being new parents. Coming so close to death really makes us appreciate each little cry, sigh, hiccup and breath she takes. For all of this we must give the glory to God! For the future, we hope there will be that in God's hands, Some LCHAD children, as they reach their teens, have developed Retinitis Pigmentosa, a degenerative loss of eyesight. We hope to enroll Ysabel in a study that is testing the use of DHA to arrest this problem (*see our website under Medical Info for the DHA Study information).

Also, she is at risk for cerebral palsy. We have been told that any infant who goes through a traumatic event is at risk for CP. At present, Ysabel is showing no signs of developing neuromuscular problems. Due to the antibiotic and ventilator use it was necessary to test her hearing. She passed that test and will be re-tested in six months. Another MRI is planned for a few months down the road. As of November 15th, Ysabel weighs 9lbs 10.3 oz and she is close to 22 inches long. All her growth is right on track. Actually, the doctors never expected her to do as well as she has been doing. She is already lifting her head up while on her stomach, and she eats like a champ! In the words of her pediatrician, "she's doing GREAT!"

We sincerely want to thank each one of our family members and friends for the incredible love and support you have provided. Whether it was prayers, phone calls, meals, gifts, hospital visits or financial support, each effort means so much to us. We could not have survived without all of these elements. You all made a huge impact on our lives. Thank you from the bottom of our hearts and we pray that God bestow special blessings on each one of you. We hope to touch your lives as you have touched ours.
Our Angel Brett ~ VLCAD

I would like to tell you our story about our son, Brett. He was born a healthy baby on June 14, 1993. We were staying with my parents at that time. I was breastfeeding him, but my milk hadn't come in yet, so he was getting colostrum. On the second day, he didn't look too energetic, he was throwing up and I thought that he was spitting up. Later that day, I knew something wasn't right, so I called the pediatrician for an appointment for the next day. He never made it to that day.

While my parents were watching Brett, Mike and I went to his sister's house to pick up a few things and we would be back in time for visitors. When we got back, no visitors showed up and I thought maybe they will be a little late. When we walked in the door, my dad was on the phone with 911, and my mom was on the floor doing CPR on Brett. I went hysterical! The ambulance came and took Brett to Scottish Rite Children's Hospital and we followed them. The nurses put us in the waiting room. Minutes seemed hours. I just couldn't believe what was going on. My son was only 3-days-old. My mom said that when she was holding him, she felt cold air blow on her face and she looked down at him and she said that he was pale and she knew something wasn't right. While we waited, my husband and my mom made phone calls. I sat on the couch wondering what did I do wrong. The paramedic asked us if we wanted to see him again, so we did. We held him and had him baptized. He looked so peaceful. I kissed him and said goodbye.

He was sent to the medical examiner for an autopsy. The autopsy results were Metabolic Storage Disease. They said that his liver was yellow and enlarged. I wondered everyday what did I do wrong. I know I didn't do anything wrong. My husband supported me through the grieving process and he said that we would get through this together. Brett is an angel now.

Six months later, I was pregnant again and my husband and I decided that this baby would stay in the hospital longer for testing when it was born. Emily Michelle arrived on September 29, 1994, weighing 7lbs 12oz, 20” long. She was a gorgeous baby. We had her tested and she was diagnosed with MCAD. We met with the genetic doctors and they explained very well what MCAD was. Dr. Rani Singh is her nutritionist and put Emily on a special diet. Her diet was Similac with iron, provimin, L-Carnitine and polycose. We've had to prick her toe every other day for her blood sugar levels and feed her every 2-3 hours. She has had 2 or 3 episodes, but pulled through. I recommend the flu shots because it saves us a trip to the ER. Emily takes Carnitor® and Polycose mixed with skim milk and she is a very healthy eater.

Today, she is an active 4½ year old and she is a proud sister to a healthy brother, Parker Riley, who arrived on April 26, 1999. He weighed 8lbs 13oz and 22”. We had him tested right after he was born and he is a carrier, but will not be affected by MCAD.

I am very happy and I still think about Brett. He will always be my angel. I am trying to figure out how to write to the Atlanta Journal about metabolic disorders and Newborn Screening. I would like for someone to help me out – give me some advice on what to write (*see article below and also on our website under Newborn Screening). We should go on national television, like "Dateline" and let the people know about these metabolic disorders. I am going to try to get the state of Georgia to pass a law that requires newborn screening. I want to save lives of infants and prevent parents from going through what I went through. It is a horrible tragedy. I am a stay-at-home mom and I take care of things. I love getting mail so anyone who reads this story, please write. I am hearing-impaired but I can talk like anyone else.

Darci White
2379 Holly Springs Road
Marietta, GA 30062
Advocating FOD Newborn Screening
‘Things You CAN Do to Promote Newborn Screening and FOD Awareness’

People write in wanting to do something so all newborns can be screened for FODs, but people don't always know what to do or where to start. I've been working on this very cause for a year, and I'd like to share plenty of ideas. **There is no greater power to induce change than personal testimony that touches people's hearts.** My experience has been that most people are more willing to talk than they are to read. When you can, it is **better to call than it is to write.** At least that's been my experience.

I suggest contacting the following:

- **Me** – so that we can work together. [wendy@savebabies.org](mailto:wendy@savebabies.org), [www.savebabies.org](http://www.savebabies.org)
- American Academy of Pediatrics, advise them to have pediatricians inform parents newborn screening with tandem mass spectrometry (MS/MS) EXISTS and is optional but is a GOOD IDEA. I've contacted them and they said they will do something, but they still need to hear it and hear it often, until screening is done religiously.
- Contact various pediatric practices, particularly large ones. They need to inform parents that the screening exists!
- [Coalition4PKUAD@aol.com](mailto:Coalition4PKUAD@aol.com) and [www.pku-allieddisorders.org](http://www.pku-allieddisorders.org) ~ Trish Mullaley runs a coalition that also advocates newborn screening. (877) 996-2723.
- Visit [www.junkscience.com/database/medrpt.htm](http://www.junkscience.com/database/medrpt.htm) or call me for the info. There are 10 pages of medical reporters and their phone numbers, across the country. Start calling! Everyone cares about newborns.
- Cal Thomas, a syndicated columnist who takes up miscellaneous causes.
- Marilyn Brooks, a syndicated columnist who does medical stories at [www.WTAE-TV.com](http://www.WTAE-TV.com), an ABC station.
- Robert Bazell at NBC, a medical reporter.
- Don Imus, who does a morning show on MSNBC. He raises money for kids with cystic fibrosis. I’ve written him, asking him to raise AWARENESS of FODs and to make the newborn screening AVAILABLE!
- Melinda Gates, who runs the charitable foundation with husband Bill Gates. I’d make a plea for ALL babies to be tested at birth. What’s $20 per baby to the Bill Gates Foundation?
- Reye's Syndrome organizations, SIDS organizations, the National Center for Education in Maternal and Child Health in VA, and the U.S. National Library of Medicine (deals with diagnosis and treatment of specific disorders) in Bethesda, MD. Deb has already done all this, but until every baby is screened religiously, it helps to constantly remind these people that we FOD people COUNT and have VALUABLE INFORMATION they need to LISTEN TO and LISTEN TO OFTEN.
- Read about SIDS and Reye's Syndrome, and send info about FODs to the authors. They should expand their writings, and they will if they're given the information often enough.
- Early Intervention programs or Zero to Three, a non-profit health organization that professes stressing good health for children 3 and under. This is about the time when FODs typically strike.
- Nursery schools, daycares, and elementary schools. Teachers should forward your information to parents. Have school children raise awareness by creating a public interest program through their school. Children are often imaginative and what better cause than FOD screening? A school in Virginia created an MCAD Awareness Day.
- Childbirth education classes, including the main organization, Lamaze International, in Washington, DC. Let's have them advise newborn screening and proper feeding during the first few days of life, so that unsuspecting parents can have knowledge and prevent unnecessary heartache.
- Malpractice lawyers who represent hospitals. Contact me – I'll get a listing of these lawyers in any area you want. In your communications, I'd use the Hershey Medical Center case in PA as an example. Presently, I know of 5 such cases in various states.
- Churches. They see expectant couples and babies all the time.
- Network with pharmacists. They may know parents with babies and may advise them of the testing.
- Celebrities. They all have attraction qualities, and some have a heart too. Perhaps they'll attract awareness.
- Ann Landers, Dear Abby.
YWCA, YMCA
- Planned Parenthood and other midwives. Midwives often like to take an interest in patients that doctors often won't.
- Prevention magazine does a lot of preventive-type articles. Newborn screening applies here.
- Pay attention to junk mail. Sometimes you'll receive mailings from charitable organizations with an interest in childcare, or from women's health organizations. It wouldn't hurt to give them some info.
- Pay attention to circulations from children's stores. Toys R Us, for example, has a publication called "the University for Parents." They run articles of interest to parents. Maybe they'll run yours.
- Insurance companies. The 4-page yellow insert from the last newsletter shows it is cheaper to screen all babies at birth than it is to do nothing and let them get sick. Insurance companies will only listen to bottomline dollars. This is best handled by a contact with someone inside an insurance company.
- Senator Ted Kennedy. His state, Massachusetts, screens newborns but how about the rest of America! Senator Kennedy is a healthcare advocate and has a lot of pull. How about mandating regional screening so that babies living outside Massachusetts can have their lives saved too with proper treatment and early detection!
- A letter, where each person needs to sign a petition for Ted Kennedy.
- www.ivillage.com, an information resource for parents and women, and similar sites. Let's educate them!

The following are suggestions for the breastfeeding issue. FOD babies fast the first few days of life before mother's milk comes in, if they are EXCLUSIVELY breastfed and NOT supplemented. Newborn screening may not help these babies because test results take at least a few days to come back ~ parents need to be advised to supplement in the first place with sugar water or formula.

- American Academy of Pediatrics. They have a policy dated December 1997 that advises breast-feeding EXCLUSIVELY and NOT supplementing. This policy can be life threatening and brain damaging to the 400 babies born each year in the US with FODs. This policy can also be harmful to the 200 babies born each year in the US with MSUD (maple syrup urine disease), citrullinemia – acute neonatal, arginosuccinic synthetase, and argininosuccinic aciduria. A pediatric endocrinologist can verify this. AAP needs to change their policy so that it helps babies and doesn't hurt them!
- The nursery committee in hospitals. They need to be educated on newborn screening and on breastfeeding.
- Childbirth education classes, including the main organization, Lamaze International in Washington, DC. They follow current American Academy of Pediatrics policies so they need education.
- Contact various breastfeeding organizations. Perhaps the hospital's nursery committees can refer you to such organizations.

Some of the most powerful things we can do take a huge emotional toll, a huge amount of time and aggravation, and are not for everyone. That would be mandating, and suing. Suing the hospital and/or pediatrician for not having children screened in the first place will open the eyes of other hospitals. When a win occurs, lawyers representing other hospitals will have their eyes opened by a headline that says, "Hospital ordered to pay $X million judgment because $20 test was not performed." I know a lawyer here who can give a referral, in or out of state, to anyone considering litigation.

Mandating is also an excellent way to get babies screened. Visit www.savebabies.org to learn what your state presently mandates. Above all, I stress doing SOMETHING. People like to give you the impression that "you can't do anything." This is untrue – if that were true, no child would be screened today for PKU. Obviously, you CAN DO LOTS OF THINGS. I've been doing things for the past year, with some successes. I believe if I have to call hundreds of places before one life is saved, that's fine because I've done SOMETHING. I'm sure the family of that one life I save would agree with that. Please do something and contact me with any feedback at all.

Wendy Nawn
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(This article is also on our website under Newborn Screening)
Our story starts on Thursday, October 7, 1999. At 7am I woke up Savannah and she made a funny comment to me ~ “My leg and my tummy hurts.” I thought that she was just being her normal dramatic, imaginative self. She had a little scrape on her knee from falling during hide and seek the night before, but other than that she looked fine. As I turned to get in the shower, I told her, “I think you're gonna live.” And with that she began to vomit on my bed. About thirty minutes after she vomited, I tried to give her some water but she began to violently vomit again. I started to worry but thought that we would go to the doctor, find out what was wrong, and get through this like we do all childhood ailments. Once again, at 8am I gave her another couple of sips of water and she could not hold it down.

As soon as her doctor’s office opened, I called and told them that I had a young child that could not hold down fluids and needed to come in as soon as possible. They told me to be there at 11am. While we waited to go to the doctor, Savannah was able to keep down a blue Otter-Pop. We went to the pediatrician's office and they did the standard CBC test and Rapid Strep test. Her pediatrician said that Savannah had strep throat but that her white blood count seemed to be elevated even for having strep throat. He told me that her white count was 26,000 (normal= 10,000-15,000). I began to get very concerned and told him that once when Savannah was four months old, she had a similar white blood count and was admitted and treated for septic shock. The previous time and this time, she had these high counts but no fever and I found that very odd, so did the doctor. He told me that I was being overly concerned and that Savannah was no longer an infant but a big girl and she would be just fine. With that, he gave her a shot of penicillin and a phenergan suppository and sent us on our way with instructions to keep her hydrated by giving her an ounce of 7-Up every 30 minutes.

We got home and Savannah started dry heaving again. You see, Savannah had been very moody the night before and had not eaten her dinner, by now she had nothing to vomit. I called the doctor and told him that we were still having problems. He told me to continue as instructed and call him at 4 pm. Meanwhile, my husband came home from work and I left to go to work for about an hour. When I was done at work, I told my boss that I would be in the next morning and that my husband was going to stay home with Savannah. Then I rephrased it and told him that I would be in unless something drastic happened and we ended up in the hospital. I picked Michael, my eight-year-old son, up from day care and he wanted to go out or do something that night and I remember telling him that we wouldn't be able to because his sister was very sick and I thought we might actually end up at the hospital. I don't know why I felt this way other than the experience we went through when she was a baby. I guess it was just mother's intuition.

We returned home at about 6 pm and Savannah was napping on the couch and Dennis had just given her 7-up. He said that he and Michael were going to grab some dinner and check the post office box and be right back. At 6:30 pm, while Dennis and Michael were still out, I tried to wake Savannah up to give her a drink. She wouldn't wake up. She was like a rag-doll. Her eyes were rolling back in her head and drool was coming out of her mouth. I also noticed that she had her hands in fists in front of her and they were shaking just like a shiver. I decided that something wasn't right and I was going to call my friend Cindy. Then, I decided not to because she was at work and it was a really busy time for her right now. Just when I decided to call her anyway and reached for the phone to call, it rang. I looked at the Caller ID and it was Cindy calling me! I didn't even say hello. I said, "God must have known that I needed you right now!” I told her what was happening and she said to call 911. I told her that maybe she was still drowsy from the phenergan and that I was going to page her doctor. She said that she would keep in contact. After that, I tried to get a hold of the doctor's office and the answering service put me on permanent hold. Frantic, I called my dad in California and he said to calm down and call 911. I called 911 then called Cindy to tell her we were leaving, and left a note that read, "called 911, at hospital" for my husband.

When the fire department and paramedics showed up, Savannah and I were in the front yard, in the grass and she was still unconscious and began to vomit. The paramedics quickly assessed her and informed me that we did not have time to get to Texas Children's Hospital. We went to West Houston, which was two miles from my house. When we got to the hospital, they determined that her blood glucose was 14 and immediately began IV glucose. They had a hard time getting an IV started on her due to the dehydration. They finally got one in her neck. After about ten minutes on the glucose, Savannah opened her eyes and
said, "Mommy, I'm all better now." Little did I know, we had yet to see the worst. They stabilized her and transferred us to Texas Children's Hospital. We had to go through the emergency department there as well. I thought that we would be admitted for the night to watch her and give her some IV antibiotics and then go home. By this point, her white blood count was 40,000 but still she had no fever. The emergency personnel at Texas Children's began assessing Savannah and shortly after we arrived, escorted my husband and I to the family waiting area. We decided that we would rather wait on a gurney parked near the room Savannah was in.

As we waited, and waited, we began to get more alarmed. Staff members, both nurses and doctors, were coming and going, coming and going from her room. Someone finally told us that they were sedating her and intubating her. While we waited another resident came and asked about three pages worth of standard questions looking for some kind of indication as to what was wrong with her. **Nothing could have prepared us for what we saw when we walked in to see her.** Savannah was lying on the bed with tubes in just about every possible place they can go. Her face was swollen and her hands and feet were swollen, blue, and cold. I immediately asked the doctor what we were dealing with and he said that she appeared to be in septic shock and was also having trouble maintaining her blood pressure on her own. I told him that we had done this once before but that it was not this severe. I told him we spent a week in NICU and then went home. The doctor told me that, unfortunately, he did not know if she was going to make it this time. I told him that I understood that he had to give me possibility but that now I wanted to discuss probability and statistics with him and he said that he could not at this time. **I never wanted to scream and hit somebody like I did then. This wasn't right! God couldn't take her from us!**

When I finished up with the doctor, my mother called and I told her what he had said. She said that she was on her way to us. They transferred us up to the PICU at around 5:30 am. My husband and I were sent to the family waiting area for that unit where we sat at a table and waited. Although all of this was very traumatic to me, all I could do was try to be strong and deal with it. My husband, who is usually not a very emotional guy, laid his head on the table and just sobbed. That, in and of itself, broke my heart. As soon as we were allowed, we went to see Savannah. The critical care pediatrician informed me that they might be giving Savannah a blood transfusion. That threw me off because she wasn't losing any blood or anything like that. The doctor informed me that because her white count was so high she had slacked off in production of red cells, which oxygenate the body and keep the tissues alive, that she may need the transfusion. I then understood why her hands and feet were swollen and blue. Her tissues were damaged due to the lack of oxygen and she could not absorb the massive amounts of fluids being pumped into her so she was filling up kind of like a water balloon.

As if all of this wasn't enough, Friday night she developed pneumonia and her right upper lobe collapsed. We waited, prayed, waited and prayed more that God would perform a miracle for our little girl. I have to say that with the few people who knew Savannah was sick, she sure had a lot of people praying for her. She was on prayer chains in California, Texas, Colorado and who knows where else. One thing we all noticed **while Savannah was sedated was that she could in fact hear us. When we would talk to her, her little heartbeat would go up and she was actually trying to wake up.** They would of course up her doses of sedative, but at least we knew she was trying. **That gave us hope.**

Saturday and Sunday were both hard too. Savannah kept on thrashing in the bed and really seemed to want to wake up. It was hard to see her like that and not really be able to help her. There were times that it took three of us to hold her down. Sunday, they decided to stop her sedation cold turkey and extubate her. The doctor gave strict orders that someone was to be in the room with her at all times. Although it was a hard long process bringing her out of sedation, it was nice to know that we were making good progress. As she was waking up, she would respond by facial expressions and nodding her head to questions. I would ask her if she could hear me, and she would nod her head "yes." Once, I asked her if she wanted that icky tube in her mouth and she frowned and shook her head "no". She couldn't really open her eyes that much but you could tell she was trying her hardest. It was nice to have her somewhat back. We got through the extubation and she was weaned off of the dopamine for her blood pressure and sent to a regular floor for the remainder of her antibiotics on Monday night. Things went fine from there and she regained her strength and started walking short distances on Wednesday.
We weren't diagnosed with MCAD until three days after we left the hospital. You see, the first time she was hospitalized, nobody gave me an explanation and I thought it was just a freak thing and was past us. This time, I told the hospital to compare the charts and that I was not going to be satisfied until they told me why my little girl could be fine one minute and knocking on heaven's door the next. Thank God they found the reason. I know that my story is very long but I do have to say this in closing ~ God is Good. My husband and I are not very religious people, but I know in my heart of hearts that Savannah would not be here today without His intervention. Had we gone to bed that night at our usual time and just carried her to bed, she would not be here today. I would also like to say to all of the parents that have lost loved ones as a result of an FOD that while my heart breaks for the pain you suffer, I admire the strength that you have to go on. I hope that if I am ever faced with the death of a child, with God’s help, I can be so courageous.

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‘No Magic Pill’

We see it ALL the time ~ take this pill or use this machine and you will lose 30 pounds in 1 week, feel more energy and be happy, or look and move like a well-conditioned elite athlete. Sounds good...BUT...life doesn’t work that way! Even though we may know in our heads that those things won’t work overnight or over the long haul, we’re hoping we’re wrong and that they WILL work and make our lives easier or more fulfilling. Sometimes when things get difficult we grasp for just about anything to ease our pain ~ whether it be physical, mental, emotional or spiritual pain.

The same principle applies to our situations as FOD Individuals/Families dealing with a chronic disorder. It would be nice if once we knew which disorder we were dealing with that our child(ren)/we could take a pill and everything would be cured and episodes would never occur. Unfortunately, magical thinking only complicates how we, and our children, might practically and emotionally deal with the disorder. As much as we wish it to be true there is NO MAGIC PILL or 'one size fits all' prescription when it has to do with losing weight, gaining muscle or being happy ~ just as there is no one symptom description or treatment protocol that covers ALL FODs for ALL children/adults. There may be some common symptoms/treatment suggestions, but you can't make a blanket statement that ALL present the same way and should be treated the same way. And that goes not only for the practical aspects of medically dealing with these disorders but also for how our children/we emotionally cope with them on a daily basis and through the years.

There is not just one way that works for everyone. There are too many factors and characteristics that are unique to EACH individual and family to make the bold and totally unrealistic assumption that what works for one will automatically work for another and will continue to work over time. So if we can't 'do it' all the same way year after year, then where does that leave us ~ what CAN we do, as parents of an FOD child or as an adult FODer, in order to cope with these disorders over time and how can we help our children with constructive suggestions when they have uncomfortable or strong feelings or behave in a difficult way and don't know where and how to channel their energies?

First of all, when you/your family get an FOD diagnosis, acknowledging and understanding that many of the assumptions you might have had about how your life SHOULD BE and how every child born to your family SHOULD BE that so-called ‘perfect’ baby are shattered ~ and that shattering most definitely will touch every aspect of your being ~ emotionally, physically, cognitively, behaviorally, socially and spiritually. Yet that shattering of assumptions does not automatically mean that life is going to be miserable. Within the context of the bigger picture, it presents us with the OPPORTUNITY to make some changes ~ not only in our own lives, but in our family's lives. If any of us stayed in that shattered state ~ I wouldn't be writing this right now and you wouldn't be reading it! So...we ALL have a CHOICE.
Unfortunately, for some, that choice includes denial or running away from the situation, which doesn’t help our children/ourselves in the long run. Yet, for most, that choice will hopefully entail putting those fractured pieces of our hearts back together ~ most likely in a new and different way ~ yet in a way that helps us to move forward in life in a positive and proactive way and to help our children KNOW and BELIEVE that they are just as SPECIAL as they are ~ with or without an FOD! And also for us to KNOW and BELIEVE that we are good persons/parents and NOT horrible people for being carriers of these rare disorders!

Going back to the weight loss scenario, cutting calories and certain types of foods is not the only focus for making that change ~ it also takes exercise, relaxation/meditation, motivation, lots of water, and being around others that will support you and not sabotage your program, as well as many other factors to really help in the process if it’s going to be a healthy process. If you chose to cut out eating completely that would not only be self-destructive and dangerous, but it would most likely cause you to gain even more than what was lost when you begin eating again. So working with all the factors as an important part of the whole picture will ultimately make an impact on your goal of seeking a healthier lifestyle and body weigh/image.

The same goes for dealing with FODs ~ seeing them in a multidimensional way and not just in one way will benefit ALL in the family. A challenge to all of us is learning how to work WITH these disorders and not AGAINST them, and work with them as an individual and as a family. A first step for many of you (probably after being THRUST into chaos after the diagnosis!) was becoming part of this Support Network where you KNOW that you are NOT alone in your struggles and in taking on all these challenges! It may not be your typical support group in that we don’t meet face to face, but it’s the immediate connection that we make with other FOD families experiencing similar concerns that makes this Group ‘work!’

In taking on the challenge we are acknowledging that our child(ren)/we have a rare genetic disorder that has short- and long-term ramifications in ALL dimensions (i.e. medical, emotional, mental, etc). Acknowledging that fact propels you to learn more about FODs so all will understand (although my brain can only hold a small amount of biochemistry!) what’s happening metabolically. Yet it may also give rise to many fears about the present, as well as what the future may hold. Gaining medical and practical information and utilizing it over time may alleviate some of those fears and the stress that often accompanies those fearful thoughts. A key to coping with these disorders is to be DETERMINED to NOT have it break your spirit or your child’s spirit.

One way of activating your spirit is to embrace and constructively express ALL of your feelings such as fear, guilt, anger, frustration, vulnerability, and grief. Feelings just are ~ they are not good or bad ~ what CAN be bad is what happens when feelings aren’t expressed. Trying to suppress them will not only give your children the wrong message about dealing with emotions, but they will most likely come out in other not-so-positive ways at some point in time (i.e., acting out, non-compliance, use of substances to numb themselves). Help yourselves and your children by learning to normalize and validate your feelings and thoughts in order to be able to go with the ups and downs and the flow of life's difficult challenges.

It’s important to help our young children to label their feelings ~ they may not be able to articulate exactly what they are feeling inside (depending on their developmental and cognitive levels). So having them draw a picture or play with puppets or toys may help you and them get a feel for what they are experiencing. Encouraging them to tell you about their picture or ‘talking’ through a puppet will further uncover specific feelings.

For older children, it may be even more of a challenge since some adolescents don’t want to talk directly about what they are feeling. Sometimes doing an activity with them such as taking a walk, listening to music, watching a show/movie, playing catch etc., can be a good way of connecting. Yet, telling them to just ‘snap out of it’ isn’t going to help the situation. They are not only medically dealing with their disorder, they are dealing with the normal concerns and learnings of adolescence and that can oftentimes be a rocky ride.
LISTEN to what they have to say and WATCH their behavior and try to reflect their feelings and thoughts back to them without telling them that they shouldn't feel the way they are feeling (i.e., angry, sad). Many times there are feelings underneath that anger and sadness ~ possibly a fear or sorrow that they might not be accepted by their peers because of their medical concerns and restrictions in activities or foods or that they might have some embarrassing episode in front of others or that they may never have the opportunity to run or play a sport. These are all REAL to them and they need to be normalized, validated and expressed in constructive ways.

Being able to do these things gives us a sense of control over what we might think is a totally uncontrollable and stressful situation. Actually, it's the holding in and down of our emotions that makes the situation even more chaotic/stressful. So MODELING the “Naming, Claiming, Framing, and Taming” (meaning expressing constructively) of our feelings and thoughts (terms that I read years ago in a Family Networker journal) is vital to actively coping with these disorders in a HEALTHY way.

In past articles I have mentioned a variety of ways to express emotions and deal with stress (i.e., journal/write stories or poetry, exercise, listen to music, hit a pillow, and volunteer, just to name a few). Any of these can be done alone or as a family. Expressing emotions symbolically can also be ‘healing,’ such as lighting a candle on special memory days or holidays or volunteering your time/money/toys at a children’s hospital. The important thing to remember is to find ways that work for you and your family.

Another important aspect of healthy coping is being ASSERTIVELY PERSISTENT when running into obstacles, whether it be a lack of answers/diagnosis, insensitivity of patronizing people that tell you to stop being overprotective and go home to enjoy your child while you can (that HAS been said to several of our families!), or difficulty in obtaining needed services at school/home/work for your child/self. It's a FAMILY AFFAIR when receiving an FOD diagnosis and many adjustments (i.e. meds, diet, specialist appointments, decision whether to work or stay home, finances/insurance, making sure non-affected sibs get their needs met too) need to be made inside and outside the family ~ so NOT GIVING UP is imperative to making those adjustments and making them manageable for your family situation. Additionally, open communication with other family members and professionals is vital. If you don't feel you are getting what your child/you need, then seek out other emotional support or medical help.

Just because we know there is ‘NO MAGIC PILL’ or quick fix doesn't mean we give up hope ~ there IS HOPE ~ hope that as individuals and families we will find meaning in our life challenges, hope that researchers will develop more sensitive/effective diagnostic tests so EARLY DIAGNOSIS and TREATMENT can begin before a severe episode unnecessarily takes another child’s life, hope that our children/we will INTEGRATE these disorders into their lives as they get older, hope that they also learn that they are NOT their disorder and that they grow to be who they are to be, and hope that those families that may experience a death in the future have the strength and support to work through their difficult times ahead.

Even though many still have not heard of these rare disorders, it is MY hope that through our efforts as individuals and as families within our local communities and even around the world, more professionals will become aware of and knowledgeable about FODs so that each child/adult with an FOD will receive the earliest diagnosis and most effective treatment so they can live a full and meaningful life!

A goal for the new millennium may be to live EACH day to the fullest because none of us knows for sure how long our mission on earth will be. Strive to Love, Live, Work, and Play with PASSION and PURPOSE! There is ‘NO MAGIC PILL’ to make any of that happen easily ~ but you can bet when they are done based in LOVE and COMPASSION, life will be much RICHER!

Peace, Joy, and Love in 2000 and beyond...

Deb Lee Gould, Director
Our LCHAD Story

My first pregnancy was textbook. Everything was going perfectly. On Thursday June 24, 1993, in my 32nd week, I discovered at work that I was spotting. I also realized that I was having small contractions I hadn't noticed before. Upon examination my doctor found me to be dilated 3 cm. When my husband, Pat, and I arrived at the hospital, I was taken immediately to labor and delivery. We were both worried, but also excited. We knew we were having a boy – Richard Patrick. We wondered what he looked like. The fetal monitor went on to locate the heartbeat. After 30 minutes and 3 different machines we were told unfeelingly that there was no heartbeat. Our Richard would be stillborn. Things happened very quickly after that, with no time for pain relief. At 4:35 pm, June 24, 1993, our son, Richard, was stillborn. The cause of death was a knot in the umbilical cord – a one in a million occurrence according to my doctor.

We agreed to an autopsy just in case there was anything else wrong. There wasn't. For almost a year after Richard's death, Pat and I kept trying for another pregnancy to fill our void. About 3 months after Richard's birth I developed cervical dysplasia. I had a large growth removed in Dec. of '93. Convinced that I could no longer bear children, we gave up hope. That was June of 1994 the same month we found out I was, again, pregnant. Our happiness was to be short-lived. During a routine dating of the pregnancy ultrasound, it was found that I was carrying twins. It was also found that they had died at about 10 weeks. I had what is termed a "missed abortion" and was sent home to wait the weekend to see if things would happen naturally. They didn't. On Monday, July 25, 1994, I went to the hospital for a D & C. These events only reinforced our desire for a child. Most people, at this point, were certain there was something wrong with me and offered advice like, "There's always adoption."

When we announced yet a third pregnancy in January of 1995 it was greeted not with joy or happiness, but with fear and trepidation. My pregnancy went well. I was sent for every test possible along with weekly planning scores (Ultrasound combined with NSTs after 30 weeks). No one wanted any surprises this time around. At 35 weeks I started having contractions and was discovered to be leaking a small amount of amniotic fluid. I was hospitalized for observation and went into labor a week and a half later. The birth was going wonderfully until it was discovered the baby was now presenting face first. There were a few tense moments when the doctor told us he would be unable to assist due to the presentation. At 10:25 pm, August 13, 1995, our beautiful baby girl, Rachel Leigh was born.

Most people would expect this to be the end of "Our Story", the "happily ever after." This was not to be. After she was born, I had taken her to the ER a few times because she seemed 'not quite right' to me and to her grandmother. I was looked at like I had rocks in my head and sent home with a "There, there." December 12 we took her to the Doctor's for her 4-month check-up and needle. On December 19, 1995, we took Rachel to the ER of the local children's hospital, going on nothing more than vague symptoms and a mother's intuition.

After about five hours of blood tests, spinal taps, etc., we realized something was very wrong. With amazing speed, yet calmness, Rachel was taken to trauma where the doctors worked to stabilize her. Two hours later, she was taken to pediatric intensive care. During her 10 days in PIC no one could figure out what was wrong with her and she was getting progressively worse. Pat and I kept a bedside vigil day and night. Family and friends constantly surrounded us with love and support. On Friday, December 29, at 9am, the head doctor for PIC drew Pat and I aside. With Rachel on artificial respiration, twelve pumps constantly dripping medication [including adrenaline to keep her heart beating], kidney failure beginning, and seizures occurring almost non-stop, he told us they had reached the top level of medical science. There was nothing else they could do for her. With incredible calmness that shocked even myself, I informed everyone that Rachel was to be taken off life support and to please come to the hospital as quickly as possible. There were ten of us around her as she was baptized. I settled into the rocking chair with Pat at my side, prepared for our last grim role as parents. As the nurse gently nestled Rachel in my arms, I thought, it was only fitting that, as we had brought her into the world, we would also usher her out. I started singing softly to her and everyone joined in. Twenty minutes later, at 12:20 pm, December 29, 1995, Rachel died while I was holding her.
Tissue samples were sent to Washington Hospital in St. Louis for DNA analysis and to Baylor College in Texas for enzyme analysis. Autopsy findings showed she had LCHAD. Pat and I still wanted a child, but were waiting to find out if there was a prenatal diagnosis for this particular disorder. If there was, as you know, we still had a one in four chance of having an affected child, which would mean (for us) termination of pregnancy. With all the heartache in our past and possibly in our future, we still had not lost hope that one day we would have a healthy child. It took the labs in St. Louis and Texas 9 agonizing months to confirm her diagnosis and identify my mutation. My husband’s was the most common and the last I heard mine was a new mutation they had never seen before that was very difficult to locate.

In October of 1996, I became pregnant again. Testing was performed December 30, 1996, at 11 weeks via CVS (chorionic villus sampling). Apparently, we were told ours was the first prenatal DNA diagnosis of LCHAD in the world. One week later, test results came back positive for LCHAD and I terminated at 12½ weeks. Determined to have a healthy child, 5½ months later I was again pregnant. The CVS was performed on August 12. We received results three weeks later. To our extreme disappointment this baby was also affected with LCHAD. Once again, for us, termination was inevitable. After each D&C, we agreed to donate the placenta and baby to Washington and Baylor for further testing and research. Emotionally scarred, but not broken, our quest continued. In January 1998, the home pregnancy test was positive. Our reaction was hope, joy, and lots of fear. CVS was again performed on March 10, 1998. By this time our thinking was very negative. I was beginning to wonder if there were other factors affecting our pregnancies. Two agonizing weeks passed before we received the results. Our phone rang at 6pm, Wednesday, March 25, 1998. Tears, joy, happiness and relief are the emotions we felt as our geneticist told us the baby was an unaffected, non-carrier. Two days later chromosomal results returned normal.

Danielle Leigh was born Sept. 10, 1998, at 38 weeks, weighing 6 lb 12 oz. That was one of the most wonderful days in my life. She was re-tested at 7 months and is 100% LCHAD-free. I am happy to report she is now a thriving 25 lb 10-month-old.

In retrospect, I now know that the immunization meant to protect Rachel was probably what threw her body into metabolic crisis. At the time of her death, no one in Halifax, Nova Scotia, had ever heard of LCHAD, let alone how to treat it. I see many stories in the newsletter of children and families living with this disease and wonder if Rachel might be alive today if she were diagnosed earlier. I can't thank Deb and Dan enough for conceiving this wonderful newsletter. I'm sure I don't have to explain the empathy we all feel. To all those families coping with their child's condition, Good Luck! I'll be reading for updates.

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**FOD Web Page Fun Facts**

[http://www.fodsupport.org](http://www.fodsupport.org)  ~ Our site is linked to by more than 50 different sites around the world. Linked to 3 different college professor's chemistry classes: 2 at the University of California-San Diego and 1 at Bowling Green State University, Ohio. The Alta Vista search engine will dynamically translate our pages into German, French, Italian, Spanish and/or Portuguese.

**Pharmaceutical Update**

Sigma-Tau Pharmaceuticals, Inc., makers of Carnitor® can be reached at 800-447-0169 or on their web page [www.sigmatau.com](http://www.sigmatau.com).
Medical Update
‘Research Study: An LCHAD Family’s Perspective
Columbia University Cardiomyopathy Study’

In previous newsletters, Dr. Bergmann has stated his need for patients with deficiencies of fatty acid metabolism to participate in a PET Scan study at Columbia University, New York City. My son, Adam, participated in this study in May of 1998 and our family wants to act as a spokesman for this very necessary study.

The study involves analyzing the process of fat metabolism in the hearts of children with defects of fatty acid oxidation. Hopefully this study will provide some answers as to why children with these defects so often die of cardiomyopathy. It is very difficult for Dr. Daphne Hsu and Dr. Steven Bergmann to get participants in their study probably because most children who frequently have to experience invasive procedures and hospitalizations aren't willing to voluntarily participate in yet another medical procedure. My son may be an exception but most medical procedures that even to some adults are trying are 'no big deal' to him. Maybe this is because he has gone through so many since his diagnosis of LCHAD at 5 months or it's just his personality. We are very grateful to his relaxed attitude because it has actually eased the stress for him and his family when he is hospitalized or needs to have one of his many necessary tests.

For Adam the PET Scan was really a simple test and Dr. Bergmann was wonderful in bringing us in the day before to explain the procedure and let Adam 'play' with the equipment. Basically all that must be done is an IV administered with a very low radioactive solution (the same as one would be exposed to from a year of background radioactivity) and then the patient lies in a tube like machine (looking much like an MRI) while the metabolism is studied, about 1 hour. Dr. Bergmann was great at administering the IV~ one try right in, and Adam fell asleep through the test. All he commented on is that his back became sweaty. Maybe they could put a blanket (one of those hospital thermal ones) underneath the patient so it would be more comfortable.

Benefits to you!!! We stayed several days at Columbia University's expense at the beautiful Ronald MacDonald House in New York City visiting such sites interesting to 7-year-old Adam as the Statue of Liberty, top of the World Trade Center, Central Park, the Natural History Museum, riding subways and taxis and of course the plane ride there. Actually this turned out to be one of our favorite vacations and Columbia University was very kind to cover most of the expenses such as airfare, transportation, accommodations, and some meals. It isn't often that because you have a rare, hard-to-manage, sometimes-life-threatening, inherited disease that you are rewarded. But this is a reward AND you are helping science! Any questions about the test from a parent's point of view you can contact us.

Don and Valerie Fulton
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Love Messages
(Please see our most current online issue)

‘We are each of us angels with only one wing…and we can only fly embracing each other’

~ Luciano de Cresenzo
In the Kitchen: Recipes for you to try

**Potato Bombs**

**Ingredients:** large potato, salt, other seasonings, fat-free ham, minced low-fat cheese, MCT Oil, breadcrumbs

Boil potato with a little salt. Mash the boiled potato without adding milk. Season it with nutmeg or other seasonings of your choice. Mix in minced fat-free ham. Add one tablespoon of MCT oil. Mold the mashed potatoes into “bombs” ~ ping-pong ball size balls. Coat the balls in breadcrumbs or seasoned breadcrumbs. Bake in a non-stick roasting pan until they are golden. Children can help mum to cook this, making different forms. Juan loves to help and make biscuit forms, sticks, little balls, etc. Perhaps you would like to add a little pepper to the mashed potato. I remember when we lived in Rochester ~ everything has pepper! You can dip them in ketchup, barbecue sauce or Tony Roma's sauces. Try them! They are almost fat free and high in carbohydrates. If you boil more potatoes you should use more MCT. And you may also leave out the MCT oil if you don't require it.

Maria Martha Fernandez
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**Decadent Fat-Free Chocolate Cake**

**Ingredients:** 1¼ cups flour, ½ tsp baking soda, ½ cup unsweetened cocoa, ½ tsp salt, ¼ cup cornstarch, 1¼ cup sugar, 1 tsp baking powder, 3 egg whites, ½ cup light or dark corn syrup

Preheat oven to 350°F. Spray 9x9” pan with fat-free cooking spray. In a large bowl, combine flour, cocoa, cornstarch, baking powder and salt. In a medium bowl with a wire whisk or fork, stir sugar and water for 1 minute. Add egg whites and corn syrup and stir until blended. Gradually stir in dry ingredients until smooth. Pour into pan. Bake 35 minutes or until toothpick inserted into middle comes out clean. Cool. Makes 16 servings. Each serving includes: 140 cal, 2g protein, 34g carbohydrates, 0g fat, 0mg cholesterol, 160mg sodium.

Kimberly Pennington
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**Vanilla Angel Food Cake**

*from “Controlling your Fat Tooth”*

**Ingredients:** 1 dozen egg whites, 1½ tsp cream of tartar, ¼ tsp salt, 1½ cup granulated sugar, 1¼ cup sifted cake flour, 2 tsp vanilla extract, ¾ tsp almond extract, ¾ tsp lemon juice

Preheat oven to 350° F. In large mixing bowl, combine egg whites, cream of tartar, and salt. Beat just until very soft peaks form. Turn mixer to lowest speed and add sugar 1 tsp at a time. Keep mixer on lowest speed and add flour 1 tsp at a time. Do not over-mix. Pour in vanilla, almond extract and lemon juice. Using a spatula, transfer the batter to a non-stick angel food cake pan. Run the spatula down deep to break any air pockets. Bake on bottom rack for 45-50 minutes or until cake is golden and cracks on top feel dry. Invert pan and let cool. Remove from pan onto serving platter. Serve with fresh raspberries, strawberries or peaches. Can also use glazes. Nutritional Information: Total Calories: 148, Fat: Trace, Percent of Calories from Fat: 1%, Carbohydrates: 33g, Protein: 4g, Cholesterol: 0, Sodium: 99mg

Kelly Decker
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Resources

Nutrition Info on the Internet:
Tufts University Nutrition www.Navigator.tufts.edu
Mayo Health Oasis www.mayohealth.org
Consumer Information Center www.pueblo.gsa.gov
Special Diet Recipes www.mealsforyou.com
American Dietetic Association www.eatright.org
Diet Analysis Site Links www.nutrition.about.com

Please remember to discuss with your doctor or nutritionist before making any changes to your child's or your diet.

Other Resources:
CPT2 web page: www.spiralnotebook.org
Pen Pal Program ~ Children's Hopes & Dreams, a non-profit organization has a pen pal program that is serving 9,000 children in 53 countries. The program is FREE for all children age 5-17 with any type of chronic or life-threatening illness or disability. The kids are matched by age, sex, and category of illness. Call (973) 361-7366 or write Children's Hopes & Dreams Foundation, 280 Rt. 46, Dover, NJ 07801

FOD Notable

Christian Parents of Exceptional Children (CPEC) founded by FOD Parent Lynda Eads, mom of Dakoda (still being tested), lke_md@familyconnect.com, http://home.beseen.com/support/dzmom/CPECandEadsEditions (may be under construction)

Family and Professional Donations

The FOD Family Support Group would like to thank recent contributors: Don and Theresa Nawn and John and Michelle Nawn in honor of Alex Nawn’s (MCAD) First Birthday. Tom and Cheryl Knight. Mary Thorson in honor of Wendy Thorson (adult LCHAD). Dean and Marcine Patterson in memory of grandson, Trevor Dean Patterson. We greatly appreciate donations to help with postage and copying fees. Checks can be made payable to Deb Lee Gould. Please note on the check that it is for the FOD Family Support Group. Please be aware that because we are not a non-profit organization, donations are not tax-deductible at this time.

A Big Thank You!

The Fulton Family would like to send a big Thank You and Hug to those of you who took time from your busy schedule to email or write us with your suggestions and experiences in managing the care of your children with LCHAD and similar conditions. We received a lot of very helpful information and will discuss with Adam's doctors, the implementation of some of these suggestions. Again, thanks a bundle.

Valerie Fulton
vallchadmom@yahoo.com
Kids Korner Scrapbook

Emily (MCAD) & Parker White

Dakoda Eads (still testing) with mom, Lynda

Reuben Gill (MCAD)

Wendy Thorson (adult LCHAD), front and siblings
FYI: Health Insurance Update

Note: This informative article was sent to me by Wendy Nawn (MCAD Parent) after researching various health insurance issues. Additionally, comments located within the [brackets] were provided by Eileen McMullin (Unsure Diagnosis Parent), a Disability and Elder Services Consultant. She wrote about Insurance and Disability in our July 1996 newsletter.

I checked with a friend of mine, who is a VP of Human Resources for a healthcare company, and I'd like to pass along some information about health insurance. [HIPAA (Health Insurance Portability and Accountability Act of 1996) became effective for most insurance plans July 1, 1997. Under HIPAA, you should not be denied coverage for a pre-existing condition if you had coverage under a group health plan (including COBRA), Medicaid, Medicare, numerous state sponsored or public health plans unless your coverage lapsed for 60 days. If you did lapse for 60 days or more, you can be denied coverage for a pre-existing condition for 12 months and, in some instances, 18 months.] This is a federal law.

[If benefits are going to be discontinued due to termination of employment or reduction in hours, the employee or employee's dependents can opt to continue those benefits under COBRA (Consolidated Omnibus Budget Reconciliation Act). These benefits can continue for 18-36 months (the length of time depends upon the circumstances (i.e., dependents under a certain age, surviving spouse or Social Security disability involvement).] Depending on the size of the employer, their cost can sometimes be less than what you'd pay by yourself. I had this situation myself when I left my job and became self-employed. The cost was about half as much under COBRA as it was later when we had to pay the whole thing ourselves.

[There are states that have unemployed parent programs. These programs help qualify the entire family for Medicaid benefits. In addition to offering Medicaid benefits, these programs sometimes cover or subsidize the cost of the recipients' insurance plan. It is more cost effective for the state.]

For those who are self-employed in small companies, like Chris and me, some insurance companies have pools you can join. You have to have at least 2 people on your payroll getting coverage to get the benefits of the lower cost of the pool. [Another way for the self-employed to obtain an affordable group health plan is to join the local Chamber of Commerce. Frequently they have enough small business owners to get a good group plan.]

I also checked with Healthy Baby/Healthy Kids Helplines in Pennsylvania at (877) KIDS-NOW. This is a federal phone number and they can direct you to the appropriate number for your state. It's a program where kids get free or low cost health insurance, depending on income and family size. For example, in PA, a family size of 4 with income under $39,245 gets low cost health insurance for kids and the same family size with income under $33,400 gets free health insurance for the kids.

[You don't have to be very low income to get assistance, however. Often the cost of medical bills (including premiums) offsets the income, making one eligible for assistance even though one's income is above the stated income limit.]

[CHIP (Children's Health Insurance Program, also known as Title XXI) was passed as part of the Balanced Budget Act of 1997. This program provides federal funding to each state for health care coverage for children. All states must provide health coverage for children. For some wonderful information on CHIP visit these sites:


You can even access, online, some of the individual state plans directly. Contact information is provided for each state listed. You can also call the department that administers Medicaid in your state for further information. The name of the department varies from state to state. In some it's called the Department of Human Resources, and in others the Department of Social Services.]
There was an interesting lawsuit in the news recently against Blue Cross. If anyone has paid high deductibles under an 80-20 plan, a portion of that amount may be due back to you. That's because the consumer has been paying their deductible based on the amount BILLED, whereas insurance companies have been paying based on the amount ALLOWED, which is considerably less than the amount billed. For example, let's say you have a baby and the bill is $10,000. If you have an 80-20 plan, you'll be expected to pay 20%, or $2,000. In reality, the amount allowed is not $10,000 but only $2,500. That means insurance pays $500 while you pay $2,000. In other words, you've actually paid 80% and not 20% like you're supposed to. Finally, someone is suing over this. To see about a refund in PA, Pittsburgh area, call (800) 295-2111. I don't know if they can refer you to your area, but if it were me, I'd make the call because I'd want to find out about getting money back.

Finally, I checked with a law firm near Philadelphia. They appeal social security income cases that have been turned down based on medical conditions. If your child was turned down for medical reasons, they can help. They appeal cases in the Philadelphia, NJ, and DE area only, but they may be able to refer you to someone in your area. This law firm, as well as many across the country, will take these cases on a contingency basis—you pay only if they win a favorable decision on your behalf. If interested, contact me (Wendy) for the law firm's phone number.

[If anyone is applying for OASDI or SSI (Social Security benefits), I would be happy to help you from the very beginning of that process so as to hopefully not have to get to the point of litigation. I can help FOD Families fill out the application in such away that highlights information pertinent to a favorable decision. I can also provide examining MDs with the information from the Disability Handbook. This way, when composing the report, the MD will have a "recipe" to follow. So if any FOD Families need some help with this process, contact me (Eileen). I do not charge for my services.]

I hope this information helps someone having a difficult time with health insurance.

Wendy Nawn, wendy@savebabies.org
Eileen McMullin, eiei@mailcity.com

Reminders

Families: Please send TYPED stories by May 1,2000. To be listed on the FAMILY LIST, please return the SIGNED Family Questionnaire or hand-write your information as seen on the current Family List and sign and date it. Continue to spread the word about FODs and the need for screening ~ it will SAVE LIVES!

Professionals: Please let us know about your research and/or clinical work with FOD Families. Send articles by May 1, 2000.

Thank You

Thank you to Erika Wallace (Mailing Lists), Jeff Schmidt (Text Web page 1996-2000), and Shelli Craig (Newsletter) for all of your hard work. Special thanks to Sigma-Tau Pharmaceuticals, Inc., for their continued financial support.

‘There is a marvelous story of a man who once stood before God, his heart breaking from the pain and injustice of the world. “Dear God,” he cried out “look at all the suffering, the anguish, and the distress in the world. Why don’t you send help?”
God responded, “I did send help…I sent you.”

~ David J. Wolpe
[Please Note: Our Group began in 1991 as the MCAD Family Support Group ~ in 1996 we expanded to include all of the Fatty Oxidation Disorders (FODs). Please be sure to read the most current newsletters to get the most updated information on FOD diagnosis, Newborn screening, treatment recommendations, research, and names of FOD researchers/Labs.]

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.]

Medical Advisor for the FOD Family Support Group is Dr. Charles Roe, Institute of Metabolic Disease at Baylor in Dallas. Email is cr.roe@baylorDallas.edu