



FOD

fatty oxidation disorder
communication network

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The **FOD COMMUNICATION NETWORK Newsletter** was created and is currently edited by Deb and Dan Gould ~ 805 Montrose Drive, Greensboro, NC 27410 (336) 547-8682
Any questions or comments should be directed to them.

From The Editor

It's hard to believe that another New Year is upon us already. I'm sure 1998 has been a busy year dealing with family, work, and play, probably more so the first two than the last! Some of us need HELP with the play part at times! Several families in our Network across the country have been working hard promoting awareness of FODs and advocating for them to be included in Newborn Screenings. A few states, such as NC, PA, WI, MA, and CA are either considering initiating some kind of a supplemental program or have already begun pilot and/or actual mandated testing. If your area is in that process, please let us know so we can inform the entire Network.

FAMILIES: Please help us out by completing the enclosed 'Inherited Metabolic Disease Survey Cost Benefit Analysis' (*in 2000, see the Results on our current website on the Newborn Screening page) by April 1 so we can SHOW the Insurance companies and state legislators that Newborn Screening and Early Diagnosis of these disorders are beneficial to ALL involved! See the Medical Update section for details. We are hopefully working toward having ALL states involved with FOD testing on some level (*in 2000, many of our Families are working with the Tyler for Life Foundation to advocate NBS for 30+ disorders, including FODs ~ see www.tylerforlife.com for more details and what your state tests for) ~ possibly through future REGIONAL TESTING CENTERS, which may be more economical and effective in the long run; considering the equipment, instrumentation, and training for individual states can be quite costly. So keep us posted on what all of you are doing to further FOD AWARENESS across the country!

We have printed several **Family Stories** in the last few issues ~ please **KEEP THEM COMING!** Sheryl Gerstl of WI (Family List, Unsure diagnosis) is interested in hearing from those of you whom are dealing with LEARNING DIFFICULTIES or DISABILITIES, and other 'symptoms' such as speech/motor delays, mood swings, obsessive compulsive disorder, anxiety disorders, and autistic symptoms that impact your child's learning and family life. She is also wondering if other parents have noticed EXCESSIVE SWEATING with their child and how you've dealt with it. If we get enough Stories on these concerns, maybe our July issue can feature those LEARNING DIFFICULTIES and how families can positively cope with them. **Professionals** ~ please feel free to also offer your expertise if you've dealt with an FOD child with these concerns.

This issue is not only packed with more Family stories, but some very strong Editorial comments. Please let us know what you think about our newsletter and its content ~ what you like, what you want to see more of, and what you don't like. We're trying to provide practical and medical information that families/professionals can REALLY use, as well as comments/information that make you think about different issues. We may not always

agree on certain issues, but one thing that can be agreed on is this ~ **‘We Are All in This Together’**



Take care...

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Editorial

I receive many calls from frustrated and anxious parents trying to advocate for their children when seeking both an accurate diagnosis and effective treatment. Several weeks ago I received my most frustrating call from an overseas MCAD mother, where professionals usually do not promote treatment with the L-carnitine that many of our FOD children/adults are prescribed as part of their daily treatment.

This mother's newborn was in the hospital when she called me and was VERY concerned that her son's floppiness was not improving even though his glucose levels had returned to a higher level and they were going to send him home the next day. She had shared with her Drs the newsletter information that I sent (MCAD Family Stories, about their children taking the L-carnitine, treatment protocol sheets, etc.), as well as the drug information pamphlet provided by Sigma-Tau Pharmaceuticals, Inc., the makers of this particular drug, Carnitor® (levocarnitine). She inquired whether her son would be treated with this drug. She was informed that L-carnitine sometimes had side effects of "diarrhea and/or vomiting" and that it would not be prescribed for him or their other son that also has MCAD and has several residual effects from previous episodes only treated with glucose IV. **She could not understand that IF this L-carnitine was available and it has helped many children, WHY wouldn't it be prescribed for HER children? GOOD QUESTION!** Additionally, **her Drs DID NOT INFORM her that L-carnitine was even an OPTION for these disorders ~ SHE was the one that told THEM about it!**

Here's my question: **WHAT IS MORE IMPORTANT ~ a little diarrhea or SAVING THE LIFE OF A CHILD FROM UNANTICIPATED INFECTION and/or a METABOLIC CRISIS???**

L-CARNITINE is AVAILABLE and SAFE with these disorders, as well as many other metabolic disorders (*there are rare cases where some have discontinued carnitine use for various reasons), and it can **MAKE A DIFFERENCE** in the life of a child/adult by not only increasing muscle tone, but also removing deadly metabolic toxins from their bodies.

One of my suggestions to this mother was to **show her Drs the list of children in the newsletter's ‘LOVE MESSAGES.’** Almost all of these children **died BEFORE a SPECIFIC FOD DIAGNOSIS was made and BEFORE EFFECTIVE DRUG AND DIET TREATMENT could be started.** MOST of those families have gone on to have other children, some of the children **ACCURATELY DIAGNOSED** with an FOD and MOST of whom (MCAD) are treated with L-CARNITINE as part of their multidimensional treatment.

As an MCAD parent and **ADVOCATE for NEWBORN SCREENING, EARLY DIAGNOSIS, and EFFECTIVE DRUG and DIET TREATMENT, I DO NOT WANT TO SEE MORE CHILDREN ADDED TO OUR ‘LOVE MESSAGES’ LIST!**

My CHALLENGE to ALL PROFESSIONALS ~ PLEASE FULLY INFORM PARENTS of ALL THEIR OPTIONS!

Deb Lee Gould, Director

Letters to the Editor

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

Now that life is returning to normal, I am catching up on my FOD news. Deb, I just visited the website and read your article on guilt. It prompted me to share something with you. I feel terrible for a parent that would feel guilty because their child died as a result of an FOD. It is devastating enough to experience the loss of a child, but to feel as if one did not do enough to prevent their own child's death is unbearable.

Sean's first episode occurred when she was 6-months-old. Within 2 hours of the onset of vomiting, she went into a comatose state. Thank God (and I'm not using that phrase rhetorically) we were in the ER when it occurred. I got her down to the ER in time... right? I should give myself a big pat on the back, right? WRONG! So much of what occurred that day was downright LUCK!

I decided to ignore my doctor's office and take Sean to the ER. But, what did I do? I passed by the ER and went to get my mother. I doubted myself. When I got to my mother's house, my sister was there and said "That's a sick kid." My sister is the type who sends her kids to school as long as they have a pulse, so that hit home. I grabbed my mother, told her to hold Sean and shake her periodically, got everyone into the car and ran every red light to the local hospital.

At the ER the triage nurse began her paperwork on Sean. I was frantic and she kept giving me one of those "Oh-No-Another-Nervous-Mother" look. A friend of mine happened to be working in the ER that night. She took one look at Sean and practically yelled "My God, what is wrong with her? She doesn't look right!" The triage nurse stopped the paperwork, took a good look at Sean and whisked us inside. The timing was incredible, just as the nurse got us on the table and the staff came rushing in, Sean went out...totally unresponsive to painful stimuli.

An IV was started...with a solution that provided enough glucose only because Sean was an infant...had she been bigger it would have been insufficient. Luck, Luck and Luck. It terrifies me to no end, the luck that was involved in Sean's first and second incidents.

Sean's second incident occurred when we were supposed to be vacationing in St. Thomas. I just happened to cancel the vacation because something that Sean was doing reminded me of her first incident...an incident that I was told would probably never occur again! Less luck is involved now. I know that Sean will be triaged properly at the hospitals that we use regularly. I know that she will be given the appropriate treatment. I know the signs to look for. But, even though less luck is involved, I cannot forget that it is still a factor. Many of her incidents occur in the middle of the night...and Sean has a tendency not to awaken. Somehow I have managed to awaken each time and find her. Most likely it is a sound she makes or something that she does that rouses me from my sleep in time. But it feels like luck to me...as much as I hate to, I have to sleep. I just hope I continue to awaken in time...I just hope I continue to be lucky...because, especially in the beginning, so much is luck.

I feel for anyone who experiences guilt. It is impossible to actually be guilty about something over which one has no control. What I feel is terror. I practice thought-stopping when my mind begins to review Sean's first incident. Because I know she could just as easily be dead as alive. Circumstances occurred in the proper sequence to support her life. Someone said just the right thing that prompted someone else to do the right thing, and so forth. Sean has been sick now for two weeks...asthma, sinus infection and a Difficile infection. I'm on alert, non-sleeping...hope I remain in lucky mode.

Eileen McMullin
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[Personal experience comments from DLG: Thank you Eileen for sharing your thoughts and feelings on parental guilt. Yes, guilt is a very common emotion ~ on conscious and unconscious levels ~ for any parent, especially those that have had a child/children die. As much as we would like to say that we shouldn't feel it, it's there, and part of it is because we place those unrealistic expectations, from ourselves and society, on ourselves that we should protect our children from ALL things~ even things out of our control! It sounds illogical, but emotions ~ any emotions ~ that are felt in such chaos and darkness, and even light, are not illogical to parents. They are just what they are ~ a deep longing to have their child/children back. That longing, over time, is hopefully worked through (*please note that thought-stopping may be suggested for certain thoughts/situations, but it is not recommended when one is working through or processing the intense emotions and thoughts of grief) and put in perspective and reality, but it is something that never fully goes away. It's kind of like what happens to some FOD children after severe and traumatic 'episodes'~ they may have some residual effects that may be temporary or may stay with them for a lifetime. For parents, a child's death is a loss that is NEVER gotten over, but with great work and perseverance can be learned from. Yet, there is what I call a 'residual grief reality' that you can't fully believe that a child of yours has died ~ it's a disbelief, a new reality ~ NOT a denial though ~ that will be with a parent in varying degrees for their entire lifetime. It's difficult to fully explain and even more difficult to understand. Yet, the more we educate families and professionals about FOD diagnosis and treatment, there will be fewer families that will have to come face to face with this difficult reality!]

Nancey's Story ~ Undiagnosed FOD

I have written a thousand letters to you, Deb, in my head over the last two years, but it has been very hard to make myself actually sit down and finally write you. My daughter, Nancey, died 8 years ago this July. She was a precious 17-month-old and mine and my husband's very best little buddy! I am just going to start at the beginning and get to where I am now and hope that I make sense throughout this letter.

My husband and I got married when we were 18-years-old. We waited four years and decided it was time to start fulfilling our dream of having a family. All I ever wished to be was a MOM. Only one month later I found out we were pregnant...we were more than thrilled. I was very sick for the first 20 weeks of the pregnancy but I rarely complained knowing of the greatest gift growing inside me.

The second half of the pregnancy went very smoothly and four days after my due date my OB/GYN induced me and I gave birth to the most beautiful baby girl I had ever seen... and best of all, God had given her to me and Andy. She was 8lbs 10oz, 21"... just perfect.

I was uncomfortable with breastfeeding at the time so I decided to bottle-feed from the hospital. She seemed to do fine until she was about 3-weeks-old. Slowly she was getting harder and harder to feed and by the time she was 5-weeks-old, we could not get her to take more than ½-1 ounce at a time. With our pediatrician working closely with us, we switched to several different formulas with no luck. Finally we were referred to a Pediatric Gastroenterologist. He was quick to diagnose Nancey with irritable bowel syndrome.

At the time that sounded like an answer so we went home with instructions to feed her diluted Enrich with fiber and give her Immodium. We tried this and she was still not improving... screaming and refusing to eat. So, then he admitted Nancey to the hospital to be tube-fed. She began throwing up large amounts of the formula... and still refused to drink from a bottle. They put her on a continuous drip and she seemed to be doing fairly well, gaining weight and developmentally on target. I decided that if all we were going to do was sit in the hospital, I could learn to take care of Nancey at our home. We went home with Nancey on the 24-hour continuous drip of the Enrich formula.

Soon the doctor wanted to wean her off the drip feedings. But as soon as we began the bolus feedings she would begin to vomit usually before the feeding was complete or shortly after. She then seemed to be admitted time after time to the hospital for test after test...but everything was looking at her GI tract. I remember the GI doctor being very concerned about Nancey's liver enzymes. They were in the 850 range the first time he tested her and he seemed to focus a lot of attention and tests on her liver enzyme level (SGPT/SGOT).

I continued to try to get Nancey interested in taking a bottle and felt such failure in trying to do so. Finally the doctor suggested we put in a gastronomy tube in her stomach and we agreed (she was 3½ months at this time). Still not doing well and having liver enzymes that were out of whack, the GI doctor decided to do a liver biopsy. Two weeks later it came up negative. We were relieved and were told she still just has irritable bowel syndrome and she will get better over time as her GI tract matures. (It is frustrating to write about this now that I know all I know, but I will continue ...the liver biopsy was a test for glycogen storage disease).

At six-months-old we finally met with a metabolic doctor (Dr. G Sherwood who worked with Dr. Andersen at Baylor Hospital at the time). Nancey underwent the 20-hour fasting test...and the information she gave me back at the time was that "she was storing her fats instead of breaking them down into useable energy." The GI doctor put Nancey on Vivonex T.E.N. and for a short while Nancey seemed to do pretty well. We were hopeful that this was our solution. At seven-months-old Dr. John Andersen called us and told us that Nancey had a carnitine deficiency and she was started on carnitine. Because we were in the middle of a political battle between the two GI doctors, our previous GI (Dr. Alan Strickland) convinced us that he could treat Nancey. (Andy, my husband, and I were young and naive and really trusted what the doctors told us at the time.) He had even gone as far to tell us that the other doctor had said he didn't know how to treat kids like Nancey. Nancey seemed to be at this point to be poked and prodded at more than ever ...not responding well to anything. She had pH probes, upper GI's, jejunum tubes, a different formula called Jevity...nothing was working! She began oral therapy because she was gagging at food when people ate around her. I was engrossed in doing what I thought would help Nancey get well and be a normal child some day.

In November, the testing was even more. They did everything from doing Barium drip feeds and taking an x-ray every hour for 10 hours to barium enemas to urine testing...at this point Nancey's liver enzymes had shot all the way up to the 1600's. We spent an

entire month at the hospital having test after test and Nancey often was picking up viruses at the hospital. She finally came home 2 weeks before Christmas. We so much wanted to have a normal life with our baby daughter.

She continued to be the cutest, most fun baby during the trauma she continued to go through. I never left her side and spent every night with her in between my husband and me. She vomited at least once during the night and we wanted to be there to help her at all times. I began to start treating Nancey as if she was normal...I joined a play group with other moms and their toddlers but was just very cautious about Nancey being exposed to illnesses because she seemed to get sick easily and often ~ lots of diarrhea, vomiting, fevers and tired a lot.

When Nancey was 14-months-old she went to Children's to see a metabolic doctor named Lewis Waber. He did a quantitative amino acid test and started her on a formula called UCD/80056. I believe at this time her amino acid test kept coming up inconclusive. We were fed up with testing and wanted our daughter to just be able to enjoy her life, so we set up a meeting with Dr. Waber to discuss "living with her carnitine deficiency" and how to maintain her health without a lot of testing.

On July 18th we had this meeting with Dr. Waber, and we left feeling very good about Nancey's future and that things would improve with her age and there would be a possibility of her having a skin biopsy in the near future to check for a missing enzyme. The next day Nancey had a fever and was very sleepy. I took her to the pediatrician and he thought she just had a virus. That night I called my husband to come home early because I was exhausted and needed his help to take care of Nancey. He put her to bed with us at 11:30 pm and I got up at 2:27 am to check on her and found her dead, lying between us, her continuous drip still going.

I jumped up and called the ambulance and when they arrived they could do nothing for her. I was in shock and throwing up...my whole life was gone...this was a nightmare...this could not be true...**I was never going to accept that I would never hold my child again...Nancey was our EVERYTHING!** We did everything for her...certainly someone was going to open my front door and let her walk back in and give me a hug.

My parents were in London...I needed my mom...somebody had to be able to take this pain away from me. My heart ached as if I was going to have a heart attack and my arms ached and longed to hold my baby again. Hours went by...it was the longest I had ever been away from Nancey.

We had her funeral a couple of days later...it was all a blur. We waited for her autopsy results and they came up with death from natural causes. **I could not and cannot accept that a 17-month-old child dies of natural causes.** My grief was fierce. I let myself feel every emotion. I stayed at home and cried and cried and fantasized what it would be like if God would just give her back to me. I went through the "what ifs" (I still do) and I looked through her photo albums and dreamed of holding her, smelling her, touching her, singing to her. I longed for people to ask me about her or tell me a memory about her.

Somehow, some way, I learned to go on with my life...but it was a different life. I was not a naive little girl anymore and I had a big hole in my heart. I thanked God for letting me have her for a little while (even though I wished deeply it could have been longer than my lifetime) but if I had to choose whether to have known her for a short time or not to have known her at all...I definitely would choose being her mom for the short time we had together.

I knew that Nancey could never be replaced but I did long to fulfill my need to be a mother...against everyone's advice...Andy and I began to try to have another baby. People thought we were crazy to risk losing another child because there were so many unanswered questions about what was really wrong with Nancey. I had to go with my heart and not my head. I became pregnant two times and had two miscarriages.

I began to wonder if this life I had dreamed of to be a mother was just not ever going to be. I asked God "Why?" a million times. Pregnant women began to haunt me...I felt completely overwhelmed and bitter about this life I was being dealt. I went to a fertility doctor who diagnosed me with several different things that could have caused the miscarriages and in November of 1991, I became pregnant again. On August 8th, 1992, my beautiful son Peter was born. He was 8lbs 11 oz, 20.5" and perfect.

I was determined to breastfeed Peter, hoping I would not run into the same problems I did with bottle-feeding as I had with Nancey. The doctors were extra cautious and ran some cardiology tests on Peter before he came home. Everything looked completely normal. They sent him home on an apnea monitor to try to help me be able to sleep. It did the exact opposite and I stayed up watching the blinking lights waiting for the monitor to go off at any time. I finally asked if we could take him off the monitor and prayed to God to give me the faith and strength to trust that he would be all right while we were sleeping.

Slowly I was beginning to feel that things would be all right. At three weeks, Peter began to fuss and have reflux. I gave him supplemental bottles and just seemed to switch him back and forth between breast and bottle feedings. We took him to a different GI doctor than we had seen with Nancey. (I wanted things to be different this time...he was not going to have a thousand tests.) Dr. Prestridge gave him some drugs for his reflux and we continued until Peter was 7-months-old and Dr. Sherwood at Baylor did the metabolic fasting test and found out that Peter had a Fatty Acid Oxidation Disorder (no specific name). He did do a skin biopsy when Peter was 9-months-old and that test did come up negative. He did share with us at this time that Nancey had this disease too but they did not know how to read the test at the time.

They put Peter on a low fat/high carbohydrate diet with Carnitor® four times a day. Peter did very well on this diet. His reflux improved dramatically and completely disappeared by the time he was 10-months-old. I was told to call if he ever got a cold or a virus but I seldom exposed him to other kids and he never got a stomach virus as an infant. When he was 1½, I joined a playgroup and I began to try to treat him as normal as possible. (But I still am a little paranoid). Peter is now almost 6-years-old and quite the little genius. I also have Mallory who is 4...she was never tested but was suspected that she could have this Fatty Acid Oxidation Disorder. She had severe reflux, low muscle tone and some other problems but Dr. Sherwood disappeared so we treated her like Peter and she has done well. We also have a daughter Becky who is 21-months-old and has always been symptom free (***please be aware that individuals can be without symptoms and still have the disorder**).

I guess all of this is surfacing again despite my kids all having such good health right now because I got a call from a person named Betsy Furler and she has a 5-month-old with suspected FOD. I read your newsletters with envy of families that have a firm diagnosis of their kid's disorders and solid treatment plans. I hope sometimes I am not being too hasty in carrying on like my children are perfectly normal. Betsy seems like, through your help, she has gotten hooked up with the right doctors and thank goodness she has a support group to help her with questions that she may have. Do you have other families similar to mine that there is no firm diagnosis on what kind of FOD they have? (*in 2000, we have over 30 Families with an unclassified FOD).

If you have any feedback for me, I would greatly appreciate it. I thank you very much because it is so hard not to wish for second chances. It is hard not to look at our family portrait and not imagine what it would look like with Nancey's smiling face in it. But I have to believe that maybe **Nancey's gift to me (besides our precious 17 months together) is the 3 kids that I might never have had the opportunity to have if she were still living.** Peter, Mallory and Becky are all special and wonderful children and Nancey's short life has taught me to cherish every moment with them. Through all my tragedy, I still feel very blessed. Thank you for your time.

Temple Ketch
Dallas, TX

MCAD Times Two in Northern Ireland!

May we as a family take this opportunity to thank you for all your help and encouragement, particularly with the recent newsletters and MCAD information. It has meant so much to us as until now we have been given no information on this condition, even though Ben (and of the last three months, Josh) has been attending the Royal Victoria Hospital for Sick Children in Belfast for the last two years. Our experience since Josh has been born and has been attending the hospital has been very negative.

Anytime we asked how other sufferers were doing we were always panned off and told they were doing fine but we were never informed as to how to contact these people for support and advice. It wasn't until about a month ago that a friend of mine let me use his computer to surf the net and I found your address and wrote off to you. You have been such a source of encouragement to us and we want you to know that all your hard work and efforts are greatly appreciated.

Let me tell you about how we first found out about MCAD. Karene and myself were married on August 10, 1994. Like any other married couple we had plans for our lives together, which included having kids. You can imagine our excitement when we found out (around April of the following year) that we were expecting our first child. The first couple of months of pregnancy for Karene went okay until around four months (27 weeks) when things began to change. Karene began having contractions and was admitted to hospital where she was put on a drip to try to slow them down. The doctors gave her a course of steroids to develop the baby's lungs just in case he was to arrive. Thankfully the contractions stopped and after a week in hospital, Karene was allowed home and told to rest and take it easy.

The rest of the pregnancy was spent with similar periods in and out of hospital and as it went on, Karene's blood pressure got higher and she ballooned as more and more fluid filled her body (face, arms, legs, ankles etc.) She went from being 119 pounds to 203 pounds. She had pre-eclampsia which explained her high blood pressure. Eventually on January 5th, 1996 Ben David Thomas was born weighing in at 6 pounds. He was forceps delivery because he was in distress and born twenty days early. In spite of this he appeared no different than any other newborn. For the next few months everything was as it is (sleepless nights, making bottles, feeding Ben and changing nappies (diapers)). That was until one morning in November 1996.

The previous week, Ben had some diarrhea and a runny nose but appeared his usual happy self. When I went in to feed him his breakfast, he was lying in his cot very limp. I called over to him and he briefly opened his eyes and lapsed in and out of consciousness. He was very clammy and had a rash over his body. I knew that something was wrong and

so we headed off to the local hospital. On arrival they assessed him but couldn't find anything wrong although they knew by looking at him that something definitely was. They tried to get some blood from his body but his veins had collapsed, however eventually they were able to get some from his ankle, but by this stage he was unconscious. They administered some antibiotics and took him away for a lumbar puncture. He was then put on a glucose drip and within about an hour and a half had regained consciousness. They took out the drip and within about twenty minutes he lost consciousness again so they placed the drip back in and this time kept it in for the next 24-48 hours.

By the next evening, Ben was back to his normal self and crawling around in his cot in the hospital ward. He was kept in for about three days and we were told to bring him back if anything changed, but they were baffled as to what had caused this incident. They had initially thought that he had the c-strain of meningitis but that came back negative. We were told to ring the hospital back after the weekend to see if anything had changed.

When we rang back they asked to see us immediately. We were met by the doctor who sat us down and explained what they had found ~ MCAD. He told us that we had acted very quickly in bringing Ben in and that if we had left it any longer that he wouldn't have lived to tell the tale. They explained the little that they knew of this condition and wanted to take a skin graft from Ben's arm to double check that it was indeed MCAD that he had. This was sent off to Holland and was positive. Blood taken from Karene and myself also revealed that we were both carriers of the condition and that there was a 1:4 chance that any future children could have the same condition.

The next few months were spent getting on with Ben's condition and treasuring each moment with him. He had another couple of incidents in hospital over the following seven months, however he hasn't been in hospital for the last year and a half apart from visits to the clinic.

In July of this year, the second addition to our family Joshua James (J. J.) Ian Thomas was born weighing in at 6 pounds 3 ounces. Like Ben, he too was born prematurely and by forceps delivery. Because of the history with Ben's condition they were keeping a close eye throughout this pregnancy. Again Karene spent the last ten weeks in hospital due to severe backache. At birth his blood sugar was 1.2 (I'm not sure if our levels vary from yours but anything below 2.9 is considered low). After a feed it went up to 1.4 so they took him into the intensive care unit for newborns. He was closely monitored and various bloods taken and this time, sent off to Great Ormonde Street Hospital (London).

Four days later the results came back to confirm what we had already prepared ourselves for, that Josh too had MCAD. The one good thing to have come from it is that at least we know that he has the condition and how to control it from having looked after his bigger brother. With both of the boys we try to make sure that they eat enough at meals (with a newborn it isn't so hard compared with a 2½ year old) and would regularly check their BM's (*blood sugars).

We are so thankful to have been blessed by God with two beautiful boys and wouldn't change anything, When we read of others, we realize how fortunate we are to still have Ben and Josh. From what little information our Drs have told us, there are about six children in Northern Ireland with this condition (two of whom belong to us). We would love to hear from anyone in Ireland (North or South) who perhaps has a child(ren) with the same condition.

To you Deb and Dan, we feel like you are family. We cannot thank you enough for your information and suggestions and hope that we can be of some help in your support group. Please do not hesitate to contact us for anything as your logo says ~ 'We're all in this together!' All our love,

David, Karene, Ben and Josh
Dunmurry, Belfast, Northern Ireland

Sean's Story ~ VLCAD

When Sean was an infant, he had really bad reflux. He was on medicine for that, but it wasn't working. It was as if he had really bad heart burn all the time. It was even worse if he ate, so he'd refuse to eat. We had to do all kinds of things to get his bottle in him. If we could get him to eat, he'd be ok (except for his pain from reflux). If he wouldn't eat, he was ghostly pale with very shallow breathing. He looked dead.

We kept telling his doctors, and no one would listen. They acted like we were nuts.

When he was 7-months-old, he and his brother got the flu really bad and went to the ER to get hydrated. Colin got better and Sean continued to get worse after we got home. A few days later we took him back to the ER and after they gave him an IV with Dextrose in it, he got better.

A couple months later he got the flu again and this time they checked his blood sugar and it was low (32). It was then that they began the work-up. At that time he also had no ketones in his urine ~ Non-ketotic hypoglycemia. They found amino acids in his urine that suggested MCAD. The doctors were all excited. They consulted specialists from just about every body system. They found carnitine deficiency at that time (*which is often a secondary disorder to many of the FODs). MCAD tests came back negative.

All the doctors seemed to write him off as "we don't know what's wrong, but don't let him fast." All the doctors except one ~ Dr. Larry Kien. He is an MD with a PhD in nutrition. By this time it seemed like Sean was a science experiment and we had had enough. Dr Kien wanted to do a fibroblast study by cutting skin off his back and growing it in the lab. At first we said no, Sean had been through so much. He was still having trouble with reflux so getting him to eat was very difficult. When he didn't eat well or at all, he'd be pale, weak and grumpy. We didn't know what was wrong, but my husband Charlie and I knew that if Sean was going to stay alive we needed a way to feed him. When medical treatment fails for reflux, the next step is surgery. We never dreamed we'd do something like beg the doctors to do surgery on our child, but we did.

The medicine for reflux wasn't working and when he didn't eat he seemed like he was dying. After a few months they finally agreed with us that he needed "guaranteed nutrition." Thank God!!!! Speaking of God, we've learned to put all our faith and trust in Him. We thought we were already doing that, but every day we learn to do it more and more. Since we have done that, we have been doing a lot better and so has Sean. His first 14 months of life, Sean was admitted to the hospital 13 times (3 times for surgery ~ 2 inguinal hernia surgeries and Nissen G-tube surgery, once for wheezing (he also has asthma), and the rest were metabolic episodes (which at that time we didn't know what was going on).

As admissions went on, we discovered first that an IV of 5% Dextrose helped pull him out of these episodes, then later that **an IV of 10% Dextrose helped even more**. When he was 14-months-old he had Nissen/ GT surgery. They placed his button then, and after a few complications of surgery were cleared up, he came home on tube feedings. After a

few months we had him eating some and just getting his tube feeding at night and during his nap. We still do it that way now. **I forgot to say we did consent to the fibroblast study. It took forever to come back, but it showed VLCAD.** Dr Rhead in Iowa (*in 2000, in WI, see our website under Medical Info/Diagnostic Labs) said Sean has no activity in the VLCAD enzyme. Dr Rinaldo at Yale (*in 2000, at Mayo, as above see Diagnostic Labs) was doing blood studies and said Sean showed a heterozygous defect for SCAD. The fibroblast study said no, he didn't have SCAD. Who knows? Dr Kien is the one who is organizing all this research on Sean. Sean had Kawasaki's Disease in 5/96 and it can have cardiac complications, so he had a cardiac echo, EKG and exam which all came back normal.

Over the last year Sean has done pretty well. His tolerance for fasting has shortened from about 8-10 hours to 3-4 hours now. It used to be that fasting and illness were the only things that caused him to have metabolic trouble, but now any stress at all causes him to try to go into a metabolic attack (things like getting too hot, cold, tired etc.). A few weeks ago he had more color changes with his attacks (his toes turned purple and his body was mottled, he was also breathing very fast and didn't have enough energy to play or sit up). That seemed cardiac related to us. He went back to the cardiologist at Dr Kiens' recommendation. He had a cardiac echo and EKG that were normal. The cardiologist wasn't very personable and said come back when he shows more symptoms because once cardiomyopathy starts, each episode will be progressively worse until something such as illness puts his heart under too much stress and kills him.

Over the past few months Sean's been trying to go into these episodes with no apparent cause and it's getting harder and harder to stop the episodes. Until now we've been able to stop most of his attacks with a bolus of his formula through his tube or cornstarch dissolved in water given through his tube. He is on a low fat diet (6 grams/day orally). He needs 26 carbohydrates every 4 hours to prevent fat breakdown, and if he won't take this orally, we give it to him through his tube (usually in the form of corn starch). He also gets 10 cc of Canola oil a day to replace the essential fatty acids.

Dr. Kien thinks Sean might be having autonomic seizures. He was evaluated by neurology and his MRI and EEG were normal. Now they want to admit him and do a 72-hour EEG. We haven't agreed to that yet. Admission to the hospital is really hard on him. He usually catches something while he's there and ends up more sick. I am an RN and my husband is a Paramedic and it seems like what Sean is doing now is cardiac-related, but as has always been the case, the doctors think we're nuts until one of their tests prove what we've been saying all along.

Our questions at this point are: 1) What symptoms have the children with cardiac involvement shown? 2) Is there anything to prevent cardiac trouble? 3) Does anyone know what to do to stop an attack if cornstarch or formula doesn't work ~ anything besides an IV ~ we use 2 tablespoon of cornstarch in water? 4) What kinds of formula have helped other children? Sean is on Isomil with one tablespoon of polydose per 2 ounces of ½ strength formula. 5) Does anyone know a metabolic doctor who specializes in VLCAD? 6) We want to know about other children's seizures related to metabolic disease. Sean's neurologist said a lot of kids with metabolic disease develop seizures because of the hypoglycemia and hypotension they experience during the episodes. They suspect that Sean is having seizures because many of his episodes are unexplained. 7) Where do we find literature on VLCAD? 8) Does anyone use canola oil or safflower oil (or some other kind of oil) to replace the essential fatty acids? If so, what and how much?

If anyone knows a good doctor who might have suggestions for us by reading Sean's story, please share it with him or her! It's really nice to find support ~ it's been a very

lonely road so far not knowing anyone who understands! Thank you so much! We would like to hear other children's stories and provide support in every way we can.

Charles and Kathy Borden
Columbus, OH

Veanna's Story ~ Undiagnosed FOD

Sorry it took so long since we last talked. We are still trying to find the answer about Veanna. At around 4 months she started throwing up. She had what I thought all of us had...a really bad cold. I nursed her. I called her my Ms. Piggy. After I nursed her she threw up every time so I took her to the doctor's office. They just told me it would pass and that it was just a spit-up and over-worried parents and she will get better in a few days. She had baby shots not too long ago and I had a Deprovero shot for birth control.

People were talking. One or another people were saying it was the baby shots or family said it was the birth control shots. It made me so mad and upset. We knew something was wrong. She's my second child so I wasn't that nervous. The doctor said to just nurse her a few minutes at a time but she got really bad. We called 14 times and went in 8 times and they told us the same thing ~ "she'll get better in a few days, it is only a cold" ~ until her last visit. She threw up in their faces!

She was gagging and just throwing up without having eaten anything. She lost about 2 pounds in 3 weeks. **I spent 3 weeks trying to tell the doctors that something was wrong. You'd think we would know our own baby the best.** They took her to the hospital. She was in the hospital 3½ weeks. I tried everything. She got a G-tube because the doctor told me to nurse her a few minutes at a time. That was not working. She had a muscle biopsy, blood test and a lot. The tests only showed that she was low in carnitine. She started on carnitine 2.5 cc 3 times a day.

When she came home it was hard. A lot of night feedings until she started eating on her own. She is seeing a geneticist and metabolic doctor and others. They increased her carnitine to 4 cc 3 times a day and they are still looking for answers. Veanna now is sitting with support but doesn't walk or crawl. The geneticist said she is very intelligent and that she is 75% of her age. When I pick her up you can feel her bones and I'm afraid I will hurt her. The geneticist also said that she had more fat on her (overweight) than muscle. We hope soon they will get the answer we want. We also got a wheelchair for her. It's made for her. The arms come down and fold in one big box. It's too big for our car and we're still looking for help for a van. I wonder if there is anyone out there with a similar story or that knows what is going on with Veanna...

Dana Crawford
Lakeland, FL

Amber's Story ~ VLCAD

It has been almost 9 years now. This is still very emotional for me to write. Remembering brings all the pain back so strong. Seeing where mistakes were made and wondering if it could have been different. Wondering if we could have been better parents. There is never enough of "only if."

Amber was born on Father's Day, June 18, 1989 ~ Father's Day will never be the same for us anymore. After she was born Joseph had gone home to get some rest. When they

brought Amber to me to feed. She had a knit hat on. They explained she was having trouble keeping her temperature up. I had rung for the nurse during her visit to me. I thought Amber had stopped breathing on me. That was the last time they brought her back to me. A doctor had stopped into my room to ask me if 'sugar' runs in our family. I told him diabetes runs on both sides of our family and who they were. Didn't say anymore or ask anymore. Just left as fast as he came. Seems so minor now. I worried they were going to tell me Amber was going to be a diabetic. Just to think I was going to have to stick my child with a needle. Could I do it?

When Joe called to see how we were doing, I broke down crying and told him to get back down to the hospital fast because something was going wrong. When I went to check on Amber, she was not in the nursery. I started to panic and they pulled Joe and I into the room where they had taken Amber. I remember the tour of that unit before Amber was born. That was where they took the sick babies. She had wires, bags and oxygen on her. They had informed us they were airlifting her out to another hospital about an hour away. Only God knows how we got there safely that day. We were in some kind of daze. Don't even know if we went through any red lights or not.

The doctor came from the neonatal ICU and told us we had a sick little girl and thought he had some kind of infection going on ~ no that's not a mistake. The doctor kept calling Amber a he/him and we finally asked if he was talking about our child! Her liver was enlarged and we learned later she also had a enlarged heart. They put her on three different antibiotics. That night they had chased her by trying to bring her potassium down chemically. The doctors asked us many questions the next day. I remember one being "Are you two related to each other then besides being married?" We said "NO!" It was like something was puzzling them. But no one would actually talk to us.

Amber was released from the hospital two weeks later. All they said to us was if she comes back we'll know there is something wrong ~ not explaining the comment. So we went home thinking the worst was behind us. Little did we know that within two weeks it would start all over again. Amber would only drink 2 ounces of formula. By the time I got all 4 ounces into her she would throw it back up on me. I called a couple times to the hospital that she had been airlifted to earlier and was told that I was over feeding her. I called another doctor and they said the same thing ~ that I must be over feeding her. I knew babies drink more then 2 ounces.

Seemed like no one wanted to hear me. So I changed her formula on my own. It was a little while before she started throwing up. We had gone out-of-state for Joe's grandparent's 50th anniversary party. Amber got moody and crying. Nothing seemed to calm her down. Call it mother instinct or whatever. I felt that she was getting really sick. Joe's aunt thought she was teething. She was throwing up her formula we fed her and wouldn't take anymore for us. Then back to sleep she'd go.

When we got back home I took her to the doctor and he gave her baby shots. I was upset about him giving her the baby shot when she was sick. The doctor told me "If you wait till a baby is well to give them their shots, they will never get their shots." That night seemed like the beginning of a nightmare. She cried something awful. Joe had to go to work early that morning. He told me before he left that I was to keep her up more that day. I let her sleep too much and that was why she not sleeping at night. After he left for work I called my mom and asked her to stop up to look at Amber. Since I was a new mom I didn't know if they acted the way Amber was acting to the baby shots. By the time my mom got there Amber had settled down. My mom took one look at her and said "Oh my God." We ran her back to the doctor (who gave the baby shot) without calling we were coming. He took one look that her and admitted her into the hospital. I had called

him from the hospital and told him if he didn't know what she had he had better send us elsewhere where they would know. So he sent her back to the other hospital.

She had no color whatsoever. We almost lost her that day. She was so close to death. The doctor told us he gave her three months to live. Her heart was so swollen it had fluid around it. That was why Amber would drink only 2 ounces of formula ~ because it would tire her to drink a bottle. Our hearts were broken that day. They ran test after test. They even ran a fasting test on her to see what would happen. It was only to last 10 hours and it had gone on longer than that. Knowing what we know now makes me feel as if they were using her like a guinea pig. Then Amber was put on a special formula with just enough fats in it for her brain to develop. **They told us they thought it was Long chain acyl-CoA dehydrogenase deficiency. They even did a skin biopsy and sent it away to make sure that's what she had.** Still today I can't pronounce the illness. Amber wouldn't bottle eat the formula. She would plead with her eyes for me to help her and the nurse would force Amber's head so she couldn't see me.

The doctor came back from the weekend and told the nurse it (the formula) wasn't meant to be fed by mouth. They had requested me to feed her other foods by mouth. I have pictures of her holding her tongue out for long periods of time with baby food on it. They taught Joe and I how to put the NG tube down to give her the formula (NG is a tube that goes from the nasal to the stomach). They also put her on digoxin to regulate the heart and Carnitor®. Amber responded well and we got to take her home for her first Christmas. Of course we had a heart monitor that gave us some peace at night to sleep. We figure we could save her if we had to.

Getting home was the best thing for Amber. I worked with her to try to get her muscles strong. She was 6 months and couldn't sit up on her own yet. When I got her in her walker she did better in it than Christopher (our second child after Amber passed on). She had EKG's and many visits to the doctors. We had one heart doctor say to me "Had I not known what she had, I would say she was normal." Joe and I were the happiest parents in the world to hear those words. We were finally heading in the right direction.

In April the other heart doctor said Amber had an NG tube infection. They put her on antibiotics and set us up to talk to someone to put a J tube in (a tube that goes from outside of stomach to the inside of stomach). Joe and I didn't really want this because we feared Amber going under the knife. Could she survive it? But we never made it to that appointment. We were told to watch out for virus. They (viruses) were what was going to send Amber to the hospital and when she did she would be put on a higher sugar IV than you or I would get because this illness Amber had would cause her sugar to bottom out. She would be in the hospital for about a week till she was over it. I guess we could handle that.

One night she had a high fever and we were scared to death she was getting sick on us. So Joe and I ran her through the emergency room. They ran some test and said she only had an ear infection and gave us what she needed and sent us home. Within a day I called down to see how much Tylenol I could go up to. I knew that I had to get the fever down because of her heart. A day later I called a doctor and told him that Amber was squinting like she was in pain. He told me to get her to the hospital and they admitted her. We informed the nurse all we knew of Amber's illness. The nurse played it off as if they have a lot of cases like Amber's. Before we were told it was rare. I had just gotten her formula down when a lady doctor came in to look her over. Amber learned not to trust the people in white. The doctor got her so upset that she threw up what I had just gotten down her.

That night all they seemed to be worried about was getting a spinal tap to see why her fever was so high. We had to remind them that her digoxin was over due. Which was when they informed us they put them on their own schedule there. They didn't get an IV on her like we were told that they would.

Within 24 hours Amber died of cardiac arrest on May 17, 1990. Her potassium was sky high and her sugar was 9 when she died. One heart doctor called us to say how sorry he was that Amber had died and that if only the doctors the night before would have realized how important it was for Amber to eat. Still we wonder WHY? We thought they knew what they were doing.

On December 14, 1991 we had a son, Christopher. We had him tested before and after he was born. We have come to the conclusion that he is a carrier like us. In 1997 with the help of Denise Needham of Cincinnati, Ohio and Dr. William J. Rhead, then of the University of Iowa, Division of Medical Genetics, **we know that Amber had VLCAD** ~ remember the skin biopsy? We have had two miscarriages since Christopher was born.

I don't know if anyone else has this trouble or if it's because we live in a rural area. But we have the trouble of making doctors here understand the seriousness of this illness. In 1997 when I miscarried, the doctor who we had picked (problem pregnancy) to deliver that child wouldn't even take my information on the illness to inform himself of it. Just wanted a copy of Amber's test results. We hope to have another child one day before we get too much older. The doctor I have now, said the cards are not stacked in our favor.

Thank you, Deb, for the newsletter of the past to the present. I have not made my way through them all yet, but they have given us strength. The fear of having another child with VLCAD disorder is so overwhelming. To think of losing another child to any illness again terrifies me, but the newsletters have had this calming effect on us. We will still try for another child and keep whatever God may give us and do the best we can with your help and support. God Bless you all for giving us strength to carry our cross. My logo idea would be a teardrop with a heart inside of it ~ Meaning the Tears of Love.

Barbara Brown
Dunshore, PA

Robert's Journey ~ Unsure Diagnosis

Our son Robert was born on July 18, 1990. I thought it was going to be a joyous event, having a healthy baby boy. It turned into a whole new experience our family had to share together.

Our son's first diagnosis was a shock ~ it came as a surprise to all of us. He was diagnosed with characteristics of Down Syndrome. We had many things to deal with as a family but we decided to give parenting a step in a good direction even though it was a path into the unknown. As time went by, all my son's tests were negative for Down Syndrome. What a relief! Robbie's Dr said he would be "normal" just somewhat delayed. Well, at that time, we were involved in Early Intervention and our caseworker, Suzy Klein Berndt, kept asking his doctor about sending Robbie for a neurological evaluation. The day our pediatrician gave in to our request was a blessing in disguise. Another medical diagnosis was discovered. Hydrocephalus ~ water on the brain. At 11 months he had a shunt implanted by Dr. Thomas Kaye, a neurosurgeon.

So time went by and things seemed to be looking up. We were expecting our second child on June 23, 1992. Courtney was born and everything was pretty normal until she

had a CAT scan that showed she had microcephaly, cerebral atrophy, almost the same as our son. Luckily, she did not have to have a shunt.

Well, as 2 years went by, my son kept getting hospitalized for what seemed to be a virus. The doctor kept telling us to change his diet and give him more potassium. The straw that broke the camel's back came in March of 1995. Robbie's Dr hospitalized him for a week because he was in excruciating pain. He thought it was some strange virus. It turned out Robbie's shunt was completely blocked and he needed an emergency replacement. There were many times we didn't know which end was up. At times our son looked as if he was almost near death.

When Robbie needed that shunt replacement, we felt it was time for a new pediatrician. In April of 1995, we found a new pediatrician. Dr. Rosner took all our medical history and he was right on the ball. Robbie was hospitalized for more episodes at our new home away from home. In June 1996 we were sent to see a Genetic Specialist in Boston named Dr. Ampola. She in turn gave us the greatest gift possible ~ our son was hospital-free! Although we had a couple of shaky moments, things are great now. My son takes 4 ml Carnitor® every day and 1 T Canola oil to make up for the fat that is needed. Now we are looking into cerebral lactic acidosis. Our geneticist said this might be the reason for our children's disabilities. We are waiting to have Courtney tested as well. We thank God and our family for their support and also that our children are healing. Whatever is dealt to us in the future we will keep you updated.

Kathy and Jay Tesini
Westfield, MA

David ~ Adult onset CPT II from 'Down Under'

A few years ago, (Christmas holidays 1994 to be exact) I was enjoying a cycling holiday around Tasmania, which is an island located at the bottom of Australia, and planning my next holiday which was to be another cycle trip to New Zealand. After that, my plan was to tour parts of Europe and England. Cycling is (was) a great way to get around and you see and appreciate much more when traveling slowly. I am by no means a fitness fanatic, not like my brother who was at the same time cycling from Manchester, England to Madras, India, which is a distance of 10,000 miles. However, I was somewhat proud of my legs, which were all muscle and could carry me 100 miles in a day against a headwind.

I had never heard of mitochondria or any of the dozens of enzymes that service them and would have never have guessed what the future could hold. Sure, there were a few instances when I had heart palpitations and even a couple of related seizures but any test I had showed everything was fine. The heart tests showed I had a heart, the brain scan showed I had a brain (and no sign of epilepsy) and the blood tests showed I had plenty of blood. "There's nothing wrong with you" I was told. Over the years I figured out that if I ate certain foods (fatty or oily) I would have palpitations and at worst, the ticker would be tired for a few days. Try explaining to the doctor at the emergency room that your heart was trying to kick its way out because you had over indulged on Mom's carrot cake.

One medical text says that it is speculated that a sudden rise in triglycerides may cause cardiac arrhythmia. Since several others with CPT problems have the same symptom, I would think that the CPT connection to this symptom is very strong. Apart from the odd cold or flu, I had no problem until the end of '95 when I was working with a colleague who liked to make life hard for everyone. I won't bore you with the details, but after several months of putting up with his antics, I started to get very tired. By mid '96, my

legs were starting to ache. This was rather alarming, as I had never had this problem apart from aches and pains associated with a cold or virus. I started visiting doctors as my condition became worse and ended up taking 10 weeks off work as I could hardly walk from the pain. I would get breathless even walking to the front door. During this time, my joints were aching, muscles aching and I kept forgetting to breathe (which is a bit of a worry when sleeping) and my lower back was killing me. I think I visited about 20 doctors and specialists over the next several months and had lots of tests and was given lots of blank looks when I tried to explain my symptoms.

I found it was best not to even mention the work situation as many doctors try to avoid anything that may involve workers compensation. Eventually **I was advised to see a local doctor who believed me (thanks Dr. Cathy!) and kept sending me for tests until the problem was found. I have a 90+% Carnitine Palmitoyltransferase deficiency.** Apparently, I have always had a few enzymes missing but the system could cope quite well until overloaded. When the body is exercising for prolonged periods, cold, ill, fasting or stressed, it requires the utilization of lipid (stored) fats for energy. This is where I come unstuck.

I soon found out that there was not much information available and there was no treatment except that patients are advised to observe a low fat diet and to exercise in moderation. Fortunately, my other brother is a doctor and he sent for some information from NORD, which was very helpful. This provided me with several clues to more information and I started writing letters to all corners of the globe. One address was the FOD Family Support Group. I received some excellent information and a package from Deb Gould, which had me puzzled as I poured over the pages looking for CPT links. This was a great education for me as I started to understand the complex mitochondrial system and how all the enzymes work and what happens when they don't.

After corresponding with a few of the CPT patients on the list, I took a doctor's advice and connected to the INTERNET. Wow! There is so much information on the web that I now have a thick folder full of information. I have also discovered the power of email and over the last few months have been in constant contact with others with the same problem. **I am not alone! It is great to be able to share your problem with others who understand what you are going through and to help each other with support and share ideas.**

Diane Martinson, whose symptoms are 'atypical' like mine, has developed a great website for CPT patients (* www.spiralnotebook.org) and Barbara Seamen has now published our very own CPT newsletter in conjunction with Dr. Vladutiu who is an active CPT researcher and has recently discovered a 15th mutation for this problem! **The number of mutations is probably why some people have problems only when they have exceeded their limits and others are in pain all the time.** It may sound bad but once you realize your limitations and concentrate on other things, you can still enjoy life. Fortunately, this is a very rare problem (less than 250 cases documented in medical literature over the last few decades) although **it is thought some patients with chronic fatigue symptoms may actually have a CPT deficiency.**

Unfortunately most doctors have only limited reference to mitochondrial problems as everyone with an FOD problem will know. However, as many CPT patients are now in contact with each other, new links are being made to symptoms that were thought to be unrelated. Perhaps the medical fraternity will listen to a group of patients rather than individual cases. My motto ~ 'there's safety in numbers!'

Being an adult with an FOD, it is bewildering to come to terms with such a problem and so hard to get anyone to listen to you and I can understand how frustrating and frightening it is for children and their parents when things go wrong. We would like to thank the Goulds and all the FOD staff for all their hard work because without their effort, all the CPT patients they have helped bring together would still be feeling lost and alone. P.S. I did go to New Zealand last Christmas and even walked the Milford track, which I always wanted to do ~ even if it was only the first 2 miles!

David Killey
Chesterhill, NSW Australia
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Questions and Answers

[Please Note: This question and answer column is designed to answer questions, both medical and practical, on FODs and their treatment. Answers to questions are solicited from those who have had firsthand experience dealing with an FOD. These include physicians, parents of FOD children and children/adult FODers themselves. It is our hope to provide general guidelines in responding to questions posed as opposed to specific foolproof solutions. Additionally, it is especially important to note that our Medical Advisor, Dr. Charles Roe, formerly of Duke University Medical School and now Medical Director of the Institute of Metabolic Disease at Baylor in Dallas, has read and approved responses to all medical questions. However, because of the individual nature of each case, it is always important to discuss these guidelines with your physician before making any changes.]

Question: What is the difference between an Amino Acid and an Organic Acid?

Answer: Amino Acids are the building blocks from which proteins are made. They are chemical compounds that have an acid group and also a basic amino group that is why they are called amino acids. The proteins in our bodies and our diets are made up of various combinations of about 20 different amino acids. There are about another twenty amino acids which are not usually part of proteins but have other roles in metabolism. An example of an amino acid disorder is PKU in which the amino acid phenylalanine cannot be metabolized properly.

Organic acids include a wide variety of chemicals that have an acid group but not an amino group. Included in the organic acids are fatty acids that come from the breakdown of fats and oils. An example of a fatty acid metabolic disorder is MCAD deficiency. Some organic acids also come from the metabolism of sugars. An example of an organic acid disorder related to the metabolism of sugars is lactic acidemia. In the metabolism of many amino acids, the amino group is removed to yield an organic acid. So there are several organic acid metabolic disorders that are due to defects in the metabolism of amino acids. An example is isovaleric acidemia that is due to a defect in the metabolism of organic acids derived from the amino acid leucine. To make things more complicated, the disorder called maple syrup urine disease has both elevated amino acids and elevated organic acids derived from these amino acids.

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Pharmaceutical Update

Sigma-Tau Pharmaceuticals, Inc., makers of Carnitor® can be reached at 800-447-0169 or on their web page www.sigmatau.com.

Medical Update Supplemental Newborn Screening Update

As many of you may know Supplemental Newborn Screening is an exciting new procedure that has been developed to identify inherited metabolic defects. The screening is usually performed by Tandem Mass Spectrometry using whole blood, dried on filter paper, that has been collected from newborns while still in the hospital. With this testing as many as 30+ disorders of Fat Oxidation, Organic Acidurias, Urea Cycle and other Amino Acid defects can be detected within the first few days of life. Those of us with children affected with inborn errors of metabolism know how very beneficial this testing could be.

Early diagnosis and appropriate treatment could greatly reduce a number of the problems associated with these disorders, **the greatest of which being unexpected death.** The best way to insure that every child is screened for these additional (above and beyond the routine state screening) metabolic disorders would be to have states mandate this testing ~ either done by a Regional Testing Center (*advocated by the FOD Family Support Group) or through the state's own Health Department Lab.

Unfortunately this is usually a long and very time-consuming process that needs to be done. I would like to share with you some information I have obtained by professionals in the supplemental newborn screening field. Although the automated technology utilized in performing this screening has enabled the Laboratories to offer this testing rather inexpensively (usually \$20-\$25 per child) the idea/benefit must still be 'sold' to the state or individual hospital. Most hospitals have numerous managed care contracts and receive a 'case rate' for each labor and delivery. This case rate is usually substantially lower than the actual cost incurred by the hospital. So while the medical community believes this testing to be beneficial, hospital administrators have problems with the inability to be reimbursed by insurance companies. Also of note is that managed care contracts quite often include a clause stating something to the effect of "no other out of pocket expenses for testing or services are to be offered to our patients." Thus presenting a stumbling block for offering this testing as optional.

Medical Directors from several large insurance carriers have been briefed on Supplemental Newborn Screening and its benefits. While all agree this testing is quite impressive and will definitely benefit patient care, they all expressed a need to show their administrators the 'cost benefit' to their companies for allowing (or paying) this charge. This is mainly due to the rarity of these disorders (although researchers believe the incident rate of one very treatable disorder, MCAD, may be as high as PKU which is already mandated testing in all states).

For two children brought to my attention, one MCAD and one VLCAD, the medical costs were staggering. One family's provider paid \$250,000 over a 10-month period, which included a 67-day hospitalization. The other child, first affected at 4 months old, required a 4-month hospitalization prior to obtaining an accurate diagnosis. The insurance company paid out \$750,000 for this child. With these two children alone, 1 million dollars (enough money to screen 40,000 newborns) was paid by the insurance companies.

Given this information I spoke with Kathy Stagni, Executive Director of the Organic Acidemia Association. We felt that with our network of families and the two support groups working simultaneously we could put together some desperately needed data. So **enclosed in this newsletter you will find a survey. The information requested would be used to compile a COST BENEFIT ANALYSIS for insurance companies.** This same information could also be used to prevail upon state legislators to mandate this testing. **All surveys will be kept completely confidential!** We hope you will lend a hand in supporting this worthwhile project by taking the time to complete and return this survey. Please return all surveys by April 1, 1999.

[Note: This article and survey project is a collaborative effort with others who are strongly advocating expanded Newborn Screening]

Resources

National Parent Network on Disabilities: 1200 G Street NW Suite 800, Washington, DC 20005. Fax: 202-638-0509 Email: npnd@cs.com Web: www.npnd.org

Toys R Us Toy Guide for Differently-Abled Kids: To receive a guide or to be put on mailing list, call 800-732-3298 or TDD/TYY 888-859-8011 Web: www.toysrus.com

KIDZ Kard: A little card that has a microchip embedded directly into the card that contains a comprehensive file of your child. The reader machine is in the area hospitals and can allow information to be immediately available to assist professionals. The service is national. Web: www.kidzkard.com

There is a **new CPT (a Fatty Oxidation Disorder) online and printed newsletter support:** CPTnet is open to patients, families, interested physicians and researchers. To register contact Diane Martinson at dianem@pmlink.com for details. To get information about the printed newsletter, contact Barbara Seaman, 312 Wisconsin Street, Lawrence, KS 66044.

Love Messages

(Please see our most current online issue)

'You can't turn off the darkness, but you CAN turn on the light'
~An old saying

'Finding the Light in the Darkness'

I began thinking about this article a while ago when I heard about a professional that was thinking about not telling an FOD family about our Group. I heard that she thought the newsletter was too depressing and that seeing all the children that had died listed in the 'Love Messages' might be too hard for the family.

I would take a guess and say that professional had never had a child die! It's a matter of perception ~ she **CHOSE** to see the darkness ~ and I **CHOOSE** to see the **LIGHT!** Every one of those children may have died physically, but their lights are still shining for their families! **And it was OUR CHOICE when my husband and I started this Support Group to honor ALL of those children for what they have given (and still are in another dimension) to their families ~ and I see NO DARKNESS in that!**

Throughout my entire life I have tried to see the 'light' around me no matter what 'darkness' has been presented to me. That was never so evident, and most challenging, than when we experienced the sudden death of our 21-month-old daughter, Kristen, from undiagnosed MCAD 13 years ago. I can attest that it hasn't been an easy road, but one filled with many 'bumps' and tears and life struggles and MORE tears.

Anyone that has had a child or children die can probably relate to that darkness ~ that ripping apart of oneself, one's family, and one's life ~ emotionally, cognitively, spiritually, socially ~ all in a split second...the second you realize that your child has REALLY died. As excruciatingly painful as that is, however, **we DO have a choice about how we will work through the chaos of that darkness.**

We can either stay in it and 'die' ourselves or allow ourselves to embrace the darkness and work through the grieving process in order to get through to the other side ~ to not only see the light but to enhance that light the rest of our lives! For me, it's been a union of Kristen's light and love with my own unique light ~ which has 'birthed' this Support Group and changing my career from teaching to Grief Consulting.

And for those that know me well, they can tell you that finding that union has been extremely painful and confusing, yet at the same time releasing and exhilarating. It has been a learning over and over and over again that pain and joy CAN co-exist and it is the experiencing and expressing of both those human emotions and everything in-between that promotes an active grief process.

The grief process is not just a one-time event or 'inside head job' ~ it is a lifelong journey that requires A LOT of Faith, Hope, and Love ~ and a lot of ENDURANCE and PATIENCE! Many will say I'm still working on the PATIENCE part! Anyway, in order to make MY process ACTIVE instead of passive, I found that it was, and continues to be, necessary to work internally as well as externally ~ individually, with family and friends, and within my own community and beyond.

My daughter may not have been on this earth very long, but HER LIGHT shines far beyond myself and my family. This newsletter and Support Group is a reflection of what she meant to all of us ~ Love, Light, and a great deal of JOY! Yet, getting to that point of finding meaning in Kristen's death AND life and a renewed purpose for my own life did not happen overnight. Working through her death occurred over many years and it intertwined with 'recycling' earlier losses and personal issues. As quickly as many would like us to 'get over it' ~ IT DOESN'T WORK THAT WAY!

Over the years I have read many books on grief, but there are two grief professionals that have helped me understand what the Grief Process entails (for all loss issues, not just from death). The writings of Dr. Therese Rando (**Parental Loss of a Child; Treatment of Complicated Grief**) and Dr. William Worden (**Grief Counseling and Grief Therapy; Children and Grief: When a parent dies**) helped me grasp the many interlocking aspects of a process I really didn't want to have to go through but knew I HAD to in order to **work toward a new wholeness and a 'healing of my fractured heart.'**

Understanding this multidimensional process was one thing ~ ANIMATING it was another story! Those of you that are in the middle of this process right now KNOW what I am talking about. Some days it felt like I was spiraling and on a nonstop treadmill at the same time! When was the pain going to STOP? I can write pages (maybe in a future issue) on explaining the "6 R's of the Grief Process" (Rando) or the "4 Tasks of Grief" (Worden), but that is not the purpose of THIS article. **My purpose is to say that WE**

ALL HAVE CHOICES and DECISIONS TO MAKE and that I hope others won't try to make them for us out of their own fears or issues! The family I spoke about at the beginning of this article almost had that done for them!

Part of our Mission as a Family Support Group is to do just that ~ **SUPPORT CHILDREN and PARENTS and FAMILIES ~ NOT try to protect them from hearing or reading about similar stories of pain.** It is through identifying with those stories that the 'healing' often begins. It is only when you go through the pain that you can feel the joy again ~ as strange as it sounds, **the pain is like a healing balm for our hearts and lives.** So that over time and with much grief work we can reach some sense of wholeness once again.

So my plea is this ~ Family members, Friends, and Professionals ~ **please ENCOURAGE families to work through their grief in their OWN WAY and TIME!** And one IMPORTANT way to do that is to inform them that **THEY ARE NOT ALONE** and that **WE ARE HERE ~ FOR THEM!** Help our FOD children and parents to EMBRACE their loss and follow it through to a new meaning and purpose in life ~ **EACH AND EVERY ONE OF OUR CHILDREN'S LIGHTS IS LEADING THE WAY!**

Deb Lee Gould, Director
October 6, 1998 Kristen's 15th Birthday
[January 1999 FOD Communication Network Newsletter]



Kids Korner Scrapbook



Marinus Smit — MCAD



The Ketch Family: Andy,
Temple, Mallory, Peter and Becky
see Nancey's story page 2



Griffin Grace
Decker — MCAD



Ashley Leann
Cajuste — MCAD



Linnea Fossum — LCHAD
with her
big sister Julie



Brett Revinski —
VLCAO

The Singer Family: Shelly,
Howard, Madison, Ashley & Austin
Undiagnosed FDD



Donations Received

The FOD Family Support Group would like to thank recent contributors: Chris and Wendy Nawn, Leonard Oberman, Nick and Gloria Kallelis (to NORD), and Linda Steinkrauss, RN CPNP for their generous donations. We greatly appreciate donations to help with postage and copying fees. Please be aware, however, that we are not a non-profit organization so donations are not tax-deductible at this time. Checks can be made payable to Deb Lee Gould. Please note on the check that it is for the FOD Family Support Group.

FOD Notables!

Scott Schulte, dad of Doug (LCHAD) and Taylor is proud to announce the spring release of his novel **Out of the Woods**. It can be purchased at LDS Bookstores or directly through Hatrack River Publications of Greensboro, NC. (336) 282-9848
Website is www.nauvoo.com/hrp

Thank You!

Thank you to Erika Wallace (Mailing Lists), Jeff Schmidt (text only Website*1996-2000), and Shelli Craig (newsletter) for all your hard work and a special thanks to Sigma-Tau Pharmaceuticals, Inc., for their continued financial support.

Reminders

Family Stories (Please TYPE) ~ especially seeking those involving Learning Difficulties, etc., for our next issue. Please send by May 1, 1999. Please also **return a SIGNED Family Questionnaire** or write out your information as on the Family List to be listed on the Family List.

Professionals: Please let us know about your research and/or clinical work with FOD families.

‘Freedom is what you do with what’s been done to you’

Jean-Paul Sartre

January 1999

Volume 9 Issue 1

[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