FOD Support
fatty oxidation disorder communication network
FOD All in This Together Fatty Oxidation Disorders Family Support Group

In this Issue

The FOD Communication Network Newsletter was created by and is currently edited by Deb and Dan Gould, 805 Montrose Drive, Greensboro, NC 27410 (phone) 336-547-8682 http://www.fodsupport.org
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From the Editor

THANKS to Sigma-Tau Pharmaceutical's continued financial support, as well as to those Families that participated in the Pampered Chef fundraiser (coordinated by Sheri Merrill), we are able to provide this issue in printed form to US and Canadian members, as well as online for our overseas' Families and Professionals. We are also able to provide **updated Family and Professional Lists ~ please review your listing** and if there are any mistakes, **send your corrections to me (Deb)** at the above email/address. If you DON'T see your name listed on the Family List then you didn't return your SIGNED Family Questionnaire that was enclosed in the new Family Packet I mailed when you signed up for the Group. We have a copy of that Questionnaire on our website, left sidebar, Online Forms. **Please date and SIGN the Questionnaire before mailing it to me** and I will add you to our next updated List**.** This may be the only printed issue until next July so **HANG ONTO YOUR LISTS!**

We are in the process of discussing our **next Metabolic Conference (possibly for summer 2004)** with Trish Mullaley of the National Coalition for PKU & Allied Disorders. Trish coordinated our last conference in October 2002. Although there are no definite plans yet for where it will take place, **we are looking for ideas for SPEAKER TOPICS as well as SPEAKERS you would like to hear.** If anyone has any suggestions PLEASE either email me at deb@fodsupport.org or snail mail me at the address at the top of this newsletter. I KNOW it'll be another WONDERFUL get-together with FOD Families and well-respected and knowledgeable experts in the areas of FOD (and other metabolic disorders) diagnosis, research, and treatment. We had @ 35 members at the Orlando October 2002 Conference ~ hopefully we'll double that for 2004!

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Thank you to Dr Ibdah from Wake Forest University School of Medicine for sharing his knowledge about Trifunctional Protein Deficiency/LCHAD. There is so little information out there for Families that we are grateful that Dr Ibdah has taken the time to write a family-friendly article. Thanks also to Lynne Wolfe, Metabolic Nurse Practitioner at Baylor, for her article on the new privacy regulations (HIPAA) ~ they can oftentimes be SO confusing, but Lynne helps us understand how they might impact all of us.

Thank you also to our Families that shared their struggles and challenges with us in this issue by way of their stories. **We welcome ALL of your stories and pictures** and we will try to either print them in the newsletter or place them on the ***Family Stories, Newborn Screening, or Love Messages*** page on our site. We would especially like to **encourage families dealing with some of the less common FODs (i.e. HMG, SCHAD, Carnitine Acylcarnitine Translocase, TFP, CPT 1&2 etc.)** to share their experiences. We're also always looking for more low fat recipes, poems, and pictures. Be sure to check our website every now and then as we add new Stories or other special items.

Professionals ~ PLEASE let me know if you'd like to share your knowledge and expertise. We can always use more **information and research articles or ongoing FOD studies** on our website as well. Additionally, THANK YOU to all the Professionals (researchers, dieticians, counselors etc.) who returned the ***'Professional Questionnaire for FOD Referral Purposes.'*** If you haven't already please **complete this one-page questionnaire on our website (Online Forms)** so we can update our files, **even if you are already listed on the printed Professional List.**

The fight to SAVE NEWBORNS continues ~ MN just passed legislation for their mandated screening, BUT expanded NBS for California is now in jeopardy! Their pilot study positively screened 41 infants but politicians are now saying the state budget cannot handle the screening on a mandated basis (but a family from CA mentioned in late June that they MIGHT be able to fund it through an alternative source)! We have Families all over this country mobilizing to **MAKE EXPANDED NBS HAPPEN** ~ it definitely helped get the legislation through in MN so let's get out there and help CA and ALL the other states that are still not mandating this **IMPORTANT SCREENING** because…

'We Are All in This Together!'

Take care…
DLG

Editorial

Please Note: *The strong-worded comments below are not directed at any of our Families or Professionals ~* ***we ALL KNOW how important Expanded NBS is!*** *These comments are directed at anyone that calls this type of screening 'defect testing' and to those that are working toward keeping the screening from being mandated in various states. The 'defect' comment was made a few months ago by a health professional (that should know better!) and* ***this Editorial is MY personal outrage at that argument against expanding NBS.*** *DLG*

Although a lot has been happening on the NBS front across the country, things are moving rather slowly. The frustration comes when the political process and money and turf issues get in the way of saving lives. Yet, as shown on the TV News, there are **some times when things move faster than usual in that political process. An example is the NATIONAL MANDATING of the Amber Alert ~ it's GREAT that having that will SAVE LIVES ~ but SO WILL EXPANDING Newborn Screening!** Many of us in various organizations have been working out front or behind the scenes for YEARS on this issue but in only 3 months the Amber Alert was passed and MANDATED across this country. My question is: What makes that issue so much more important than expanding NBS that it can be moved through Congress so quickly and unanimously? A question that is probably not politically correct to even ask and one that we will probably not get a full answer to ~ but I feel is worth asking.

Of course, critics of expanding NBS will be quick to voice their opinion on this issue and then some. **For those critics of Expanded Newborn Screening that believe this type of screening is really 'un-consented genetic testing to create a database, track and research defective children'** (these are paraphrased comments taken from comments made by a health professional who tried to derail expanded NBS in MN) **~ it is NOT! I was APPALLED** at the news release this health professional from the Citizens' Council on Health Care posted on her website **calling NBS 'Defect testing' and calling our children that are identified 'LESS THAN PERFECT!'** If you've ever experienced the **ramifications (death or major LIFELONG medical complications) of NOT knowing** your child has/had one of the 30+ disorders that can be screened for using tandem mass spectrometry, **you would KNOW** that it's **NOT** a matter of testing in order for some medical/insurance/government group to track/exploit your family's medical history!

Expanded NBS is also not trying to ascertain (by genetic testing) whether someone will be predisposed to some disorder/disease process in the **FUTURE** because they may carry a specific gene ~ **it is a SCREENING for SILENT and INVISIBLE disorders that if not detected AT BIRTH may not give that infant a CHANCE TO LIVE beyond the first days of life if they are not treated immediately!** These are also not disorders that a person will develop because of environmental or behavioral factors (i.e. developing cancer due to asbestos exposure or smoking) ~ **they are BORN with these disorders ~ and whether or not Expanded NBS gets fully mandated across this country, these babies are STILL GOING TO BE BORN ~ and many will continue to die just because some critic(s) believe it's not right to screen for them because they think families shouldn't be subjected to such testing or that their screening card shouldn't be kept on file for several years.** They state that privacy is an issue ~ but if you read Lynne's article on HIPAA in this issue, you will learn that **privacy is taken into consideration and there are laws to protect it!** States that are currently adding the expanded screening are also implementing their own privacy regulations in addition to what the federal government has already implemented so **the privacy issue really isn't an issue in my opinion.**

As for consenting to expanded screening at birth versus having it mandated and being **automatically** done on all newborns (unless for religious objections) **~ I would have given anything to have had the medical professionals just do the Expanded NBS in 1983** (if it was available) when our daughter, Kristen, was born ~ unfortunately, the screening test wasn't fully developed at that time and our daughter died suddenly because we DIDN'T KNOW she had MCAD, one of the many disorders that can be screened for AND treated through diet and medication IF detected EARLY. And if her NBS bloodspot had been saved **we may have learned about her MCAD a lot sooner than 1 year AFTER her death!**

So **to ALL the critics out there ~ This issue of expanded NBS is NOT about trying to place your NOT SO PERFECT LABEL on infants or about the government having a hidden agenda of creating a 'defect inventory' of families with genetic disorders ~ it's about SAVING LIVES ~ OPEN YOUR EYES and your HEARTS!**

Deb Lee Gould, Director

Family Stories - *Henry's Story, Unclassified*

Henry is our first child and was born after a rough pregnancy. I had no trouble getting pregnant, but had spotting/bleeding throughout the first trimester and high blood pressure and edema starting at the beginning of the second trimester. I went into labor at 28 weeks, but labor was stopped and I was hospitalized over night. I was on bed rest for the remainder of my pregnancy for either pre-term labor or high blood pressure. Finally at 36 weeks, my water broke.

The OB was very excited as he was planning on inducing early. My labor was long, but relatively easy and **we were so excited to welcome Henry into the world on February 6, 1998.** He was 6lbs7oz – a great weight for a 36-weeker. He was very alert, but was uninterested in eating. We worked hard on breastfeeding for 3 months. **Henry had one blue spell in the hospital** and seemed to be "growing" little round band-aids. I think his blood sugar was low or borderline, but I was told nothing.

We went home after 2 days and **Henry continued to struggle with eating.** Feeding him became my 24-hr/day job. I was determined to breastfeed so I fed him, pumped, and then bottle-fed every three hours. **It was exhausting, but I was so happy to have this baby!**

At 3 weeks, the Pedi Dr decided that he was failing to thrive and hospitalized him. There **the GI doc decided that I wasn't trying hard enough to feed him and that he would be fine it I just breastfed. After all, she breastfed and her kids were fine!**

Henry went into a coma-like state and my husband came in and squeezed formula into his mouth until he came around. That doctor eventually was asked to leave the hospital. We went home and continued feeding Henry as often as possible.

At 6 weeks, he stopped breathing and he was admitted for apnea. They came to the conclusion that **he had severe reflux (that was a correct diagnosis).** He came home on new meds and new formula and was somewhat better. At 3 months, Henry started screaming and would not stop. We went back to the ER. They decided that **there must something else wrong.** The next day **we saw a GI doctor that diagnosed Henry with an FOD.** He was put on a fat free diet and did much better. At 6 months we started checking blood sugars and he started Carnitor®. **He had low muscle tone, but once he started taking Carnitor® his muscle tone normalized.** He thrived on the Carnitor® and still does.

Over the years, **Henry has been through quite a bit.** His development has been excellent and **at five he is reading and learning to ride a bike!** He is very talkative and friendly. **He has food allergies and mild asthma that complicate his diet.** His blood sugar also fluctuates greatly. He is on a low fat diet and cannot eat beef, pork, or dairy products. He has some problems now with sugars especially from fruit. Lately he has had a lot of highs and we are in the process of having that evaluated. He has had a few seizure-like episodes and one that lasted about 30 minutes. He had presumed pancreatic insufficiency as a toddler and was treated with Creon5. We have continued to struggle with Henry's lack of appetite. He is now 40 lbs and 42 inches and although a little small for his age he fits in well with his classmates. Henry has behavior problems when he is "out of whack," but is wonderful when he is feeling well. He has been through two surgeries and several viruses and did fine on the IVD10.

We live in Houston, TX but Henry sees Dr Vockley at Mayo for his metabolic Dr. We have been very pleased with the care Henry receives there. It is well worth the commute. We have 2 wonderful pediatricians here in Houston who consult with Dr Vockley. He is wonderful at communicating information about Henry's care with us, as well as his Drs here in Texas.

Henry will be in a regular kindergarten class in the fall. **We are expecting our second child in January of 2004** so we have a lot of changes coming up! Henry is excited about having a sibling and promises to help out with the baby. We are lucky that Henry has done as well as he has. **He is our little miracle.**

Betsy and Eric Furler
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Family Stories - *Nanci's Story, Adult onset CPT2*

I thought it might be interesting for some of the adults out there to hear another adult's story. **My problems began in infancy, in the early 1950's.** During my first year of life, I suffered what my parents thought must be polio (this was before the vaccine). I apparently became rigid and didn't move for about three days, following a fever. Of course, the pediatrician didn't know what to make of this, and everyone was relieved that it was NOT polio!

As a child**, I suffered many bouts of what I now know is rhabdomyolysis**, usually in connection with some infection or overexertion. Each time this would occur, I would be laid up in bed for several days, mostly unable to move and in pain. After it would run its course, I would recover and be okay for awhile. And, of course, there were the dreaded physical education classes! I was always the slowest, weakest, worst in everything! Gym teachers always told me to try harder, get in shape, and don't be so lazy!

I always knew there was something not quite right. No one else I knew had these bouts of what we termed "muscle stiffness," but the doctors never seemed to pay much attention to this. I was determined to maintain an active life style that included swimming, tennis, and skiing, and for the most part was able to do this.

As I grew into adulthood, the incidences of rhabdomyolysis became more severe. Sometimes the triggers were be fasting, menstruation, stress, or illness. I still maintained an active lifestyle, but sometimes I would get "attacks" that would seem to come out of nowhere and last for days. I never found a doctor who could explain any of this, nor did I realize the seriousness of the situation.

In my mid-thirties, the situation worsened as I aged. I got a new job (I'm a special ed teacher) teaching emotionally disturbed adolescents who often acted-out physically and required physical restraint for safety reasons. After about seven months of performing these restraints, **I fell gravely ill with renal failure.** I had just recently gotten married, moved to a new community, and started this new job, so stress levels were high.

One day I started running a high fever (not unusual for me) and the 'rhabdo' set in. By the time my new husband got home from work, I had the telltale dark urine, which at the time I thought was blood. I didn't know what was wrong with me, but knew I was really ill. My husband insisted on taking me to the hospital and I agreed (much to his surprise, for I had always shunned hospitals before). **After examining me, the emergency room doctor took my husband aside and in a confidential tone explained to him that there was nothing wrong with me ~ that it "was all in my head!"** How he came to this conclusion, I will never know. **Luckily my husband insisted we see another doctor, who was able to diagnose the renal failure.**

I must say that **the following two weeks were the most physically painful I have ever experienced.** I received around-the-clock dialysis that was excruciating. I suffered a seizure and hallucinations. **The doctor treating me had no clue why the renal failure occurred.** When asked why, he simply replied, **"Don't worry, it will never happen again."** I always wondered "why not?" I was lucky, because my kidneys eventually began functioning again and I was able to resume a "normal" life.

Looking back at this time, I wonder why the nephrologist didn't seem interested in investigating the cause of the renal failure. In the meantime, **I informed my employer that I could no longer perform these restraints. My employer informed me that I would therefore be fired, due to incompetence (I had a flawless teaching career)!** What followed was an ugly lawsuit, which I eventually won. But, **the idea prevailed that because I "looked fine," there must be nothing actually wrong with me.**

The quality of my lifestyle continued to decline as I approached my 50s. I was still determined to remain active, but found that I could do less and less with each passing year. Long, beloved hikes in the beautiful Catskill Mountains became shorter strolls. I skied less and less, and **often experienced muscle pain and weakness when doing nothing much at all!** Even going up the stairs started becoming a chore, as I would need to hold on to a railing and pull myself up. I started to get scared. The kidney failure convinced me that whatever was going on was serious. **I knew I should do something, but what? Should I see a muscle doctor? Is it an autoimmune problem? I really didn't know.**

About this same time, my husband and I were trying to have a baby. **Several miscarriages later, we gave up and adopted our son, who has become the light of my life!** About five years ago, I was out hiking with my family, when my muscles just gave out, and I could walk no more. Luckily, my husband was able to find a forest ranger that rescued me with his jeep. This experience left me more scared. **Then about two years ago, I think I hit bottom.** I took my son to camp and had a few hours until I had to pick him up. This being the first day of summer vacation, I was jubilant as I began a short hike alone in the lovely mountain morning. **About a half hour into the hike, my muscles gave out again, and again I could not walk! I was terrified. No one was around, and I could not get back to my young son** who would be scared when his mommy did not reappear. All I could do was to lie in the dirt and yell for help. Eventually, I was again rescued. This experience left me rattled. I knew I had to do SOMETHING or my life would become more and more limited. But, I still didn't have a direction.

I think there must have been divine intervention when that evening, my husband leafed through an old Merck manual and found a listing for McArtle's disease! Now we were getting somewhere! I checked on the internet, and it felt like we were at last on the right path. I was **also lucky enough to find Dr. Slonim that day on the internet, who specializes in FODs.** I was in heaven the first time I spoke to his nurse, Linda, and she didn't treat me like an alien! **Dr. Slonim and his wonderful staff worked diligently with me, and within a matter of months, I had a diagnosis!** I understand this is actually pretty quick, but it took 50 years to get there!! We started on nutritional changes and a trial run with alanine, an amino acid.

I went through lots of stuff when I finally received a diagnosis. **At last I had a name to put to what I had been experiencing my whole life ~ CPT 2!** And, I realized there were others that had similar experiences! On the other hand, the diagnosis seemed to make this a "disease," and I had to adjust to this.

Last year Dr. Slonim suggested **I might be a good candidate to participate in Dr. Roe's study and I was accepted.** Imagine my delight when **I was suddenly surrounded by an entire staff of experts on CPT 2! Six months into the treatment, I have had amazing results!** Gone is the persistent muscle pain and stiffness. I skied all winter and feel like **I am getting my life back again!** Of course, there are still rough spots. On my last visit (a month ago) I was plagued with high blood pressure and unable to complete exercise and strength testing. I am confident, though, that this will be overcome and that I will be able to continue my participation. I think the **most important part of all of this has been the HOPE it has given me.** No longer do I feel like the alien that emergency doctor saw when he looked at me**. I have found strength in knowing I have at least some control over what's happening in my body,** and that I can hopefully look forward to a happy, active life! To all of you reading this, may you be well and happy!

Nanci Perrin, Saugerties, NY, vegiepal77@hotmail.com

Family Stories - *Jenna's Story, Canadian MCAD*

At 4:04 p.m. Feb 17, 2002, our beautiful baby girl came into this world. Jenna weighed 8 pounds 7oz and she was 19½ inches long. Right from the start **I knew there was something special about this little girl.** She had vibrant red hair and beautiful blue eyes. I asked the nurse if there had been a mistake. Had someone switched our baby? My babies are supposed to have dark hair and dark eyes. We all laughed and the nurse reassured me that this was our baby. Two hours later, Jenna and I were settled in our room.

Jenna was a very sleepy baby. I had a difficult time trying to get her to breastfeed. The nurses reassured me that this was all very normal because of the delivery. After many attempts I finally got her to nurse.

The next evening my pediatrician came to the hospital to examine Jenna. He listened to her chest, moved her legs and declared that she was a "healthy" baby. He told me to call his office to book her two-week check up. Then he was gone. Later that evening the nurse took Jenna from me in order to do her heel prick test. (i.e., PKU and Hypothyroidism). **Little did we know how important this test could have been for Jenna's very survival.**

We were discharged from the hospital the following afternoon. I was ecstatic. Everything seemed so great. Our circle was complete. We were truly blessed.

Jenna was a strong baby from the day she was born she could hold her head up. It was like she didn't want to miss a thing. By the time she was two months old she had learned to roll over on to her tummy. She would not sleep on her back no matter how many times I tried to keep her in this position for sleeping. At four months she had already cut two teeth. At six months she had started on some solid foods and was enjoying this new sensation. As most infants do she especially enjoyed razzing when she had food in her mouth. At 7 months she was crawling all over the place trying to keep up to her older siblings. She especially loved to crawl towards the sunniest place in the room to bask in its warmth. Almost like she remembered where she had come from. At 8 months she was able to crawl upstairs. By nine months she had started cruising around the furniture. We were certain that she would be walking very soon. Jenna was a wonderful little girl who loved to babble dada and melt her daddy's heart. She would giggle with delight when you gave her raspberries on her tummy. Developmentally she seemed to be reaching her milestones and more.

The week of November 11, 2002, Justin was very sick with a virus. Jenna seemed fine except for a bit of nasal congestion.

On Wednesday November 20, 2002 Justin seemed better. He and Jenna seemed to have an average day. They played, and ate just like normal. I was relieved. I thought that we were finally free of the virus.

Later that evening, I went upstairs to put Justin in his pj's. Jasmine was holding her baby sister for me. Just as we were making our way down stairs, Jasmine yelled at me that the baby had just vomited all over the place. I cleaned everybody and everything up. Jenna seemed fine. I thought that she had vomited from gas or maybe she was coming down with the flu. I offered her a bottle and she drank the whole thing and kept it down. She seemed really tired so I put her to bed.

Thursday morning she woke up around 7:45 am, which was late for her. She didn't seem herself. She was very lethargic. I called the pediatricians office right when they opened at 9:00 am.

When I spoke with the pediatricians nurse I told her Jenna was not herself and that Justin had been sick the previous week with what I suspected was the flu. She asked me if Jenna had wet her diapers. I told her that she hadn't and that since Jenna had woken up she was very lethargic. She asked if Jenna had a bowel movement recently, I replied that she hadn't had one as of yet. The nurse advised me to come to the office for 1:30 and to keep pushing fluids in the mean time. I asked her if it was ok to give a nine-month-old pedialite. She said yes it was fine.

At 1:30 Thursday November 21, 2003 I brought all three children to the pediatrician's office. When the doctor entered the examining room I was holding Jenna in my arms and she was quite sleepy. He commented that while she was quiet he would look in her ears. I then placed her on the examining table and she suddenly seemed more alert. I guess it was the change of environment or being out of the warmth of mommy's arms. The doctor listened to her heart. He informed me that Jenna was running a fever and to give her Tylenol or Tempra but that he didn't think anything serious was going on. Her diagnosis was the flu. I was told to keep pushing fluids. Once again I asked if it was all right to give Jenna pedialite and he replied that yes it was fine but that I should try gastrolite as it was a less expensive alternative to pedialite. The doctor then switched his attentions to my other two children. After reassuring me that they were fine he left the room. The whole appointment lasted no longer than 5 minutes. I was not happy with the diagnosis but I said nothing. Something I deeply regret.

We arrived home from the doctor's office around 3:30. I made dinner for my two older children. After making dinner I sat on the sofa with Jenna. Her condition had not changed from when I had brought her to the doctor's office. She was still very lethargic so getting her to drink was a challenge, but she was drinking so I took that as a good sign. When my husband came home the baby seemed to perk up a bit at the sight of her daddy.

We were having some flooring installed in our house the next day so my husband was busily preparing the house for the next morning. Around 11 pm my husband was getting ready to go to bed. I asked his advice regarding the baby. She had just finished about two oz of Similac and wasn't vomiting any longer. I wasn't feeling the best about putting her to bed. My husband reassured me that Jenna was in need of a good nights rest as I had been handling her all day long. I agreed and put her to bed. About half an hour later I heard her cry out. I checked on her and she seemed fine.

On Friday November 22, at 4:00 am Jenna cried out. I bolted from our bed to check her. She was lying in her bed with her eyes closed. I decided to change her diaper and try to get her drinking again.

I tried giving her a bottle but she would not drink. She just kept moving her head side to side as if to say no. When I finally did get her to take a sip of pedialite she seemed content. Awhile later Jenna started making noises as though she were going to vomit. At that moment my gut instinct started telling me that something more wasn't right.

I heard my husband in the shower. I went into the bathroom to look at the baby where there was more light. She didn't look good to me. I mentioned this to my husband. We agreed that if she weren't any better in a few hours we would take her back to the doctor.

Continued on Page 6

Jenna's Story, Canadian MCAD… cont'd

Unfortunately, we didn't get that opportunity at round 6:30 a.m. all hell broke loose in our home. The baby stopped breathing I tried to perform CPR while my husband was on the phone with the 911 operator. After what felt like an eternity, the paramedics finally arrived at our home. They whisked her off in the ambulance and told us to meet them at the children's hospital.

My husband sped off after the ambulance in his car while I waited for someone to come and stay with our other children. Finally, a police officer arrived and agreed to take me to the hospital . It was the longest ride of my life. I kept my composure throughout the ride. I envisioned episodes of the television series ER. I thought that I would get to the hospital and they would tell me that my baby was going to be alright after a brief stay in hospital.

When we arrived at the emergency my husband was waiting outside for me. I figured that this meant one of two things.

1. That they were working on Jenna and he didn't know what was happening.

Or

2. That Jenna had died.

Unfortunately it was number two.

To say the least we were in shock. How could this have happened to our baby who only two days previous seemed so healthy?? This was the first time she had ever been ill. Surely there had to be some mistake. **Initially we were told that the cause of death was Reye's syndrome.**

About a month later we received a call from the coroner's office stating that **the cause of death had been changed to MCAD.**

In my grief I decided to seek out as much information as I could regarding MCAD, and that is when I came upon this wonderful FOD Family Support Group. We would like to express a special thank you to Deb and to all of you who have reached out to our family.

Because of all of you we are not alone!!!!!!!!!

Sincerely,
Tammy and Roger Clark
rclark@storm.ca
Jenna 9 months ~ deceased MCAD
Justin carrier of MCAD
Jasmine unaffected
Please visit Jenna's story at www3.sympatico.ca/tammy_roger/Jennastory.htm

Morgan's Update, GA2

This is an update on Morgan Lynnae Jones. We wrote her story about 3½ years ago. At the time she was only 7-months-old. **Now she is almost 4-years-old. Morgan has gone through a lot of changes in the last 3 years.** After a diagnosis (not finalized until July of 2002) of GA2 in February of 2000, Morgan had had another brain hemorrhage. In May of 2000, Morgan had what was called a "washout." This is where they drill holes in her skull and drain the fluid and blood off of the brain. She did so well that she got to come home after 2 days. **She started making remarkable progress** after that, and after 6 months to a year, her brain filled up the space inside the skull. **No more hemorrhages!!**

Morgan had a lot of help to get where she is today. She saw 5 therapists a week: PT, OT, DI, vision, and speech, and still does to this day. But, we noticed a problem at 1-year-old. I noticed she wouldn't respond to her name. The speech therapist told us we should have another hearing test done. So we did. It showed that her hearing was on the low-normal range, and they told us to come back in 6 months for another hearing test. But after a few months, I noticed her putting toys to her ears to hear them. We immediately went to Cincinnati Children's Hospital (CHMC) where we had an ABR test done. It showed that she had a moderate hearing loss in the left ear and a mild hearing loss in the right. I felt so bad knowing that all of this time she couldn't hear things we were saying and that's why her speech was so bad. **Morgan was almost 2 before she got the hearing aids to help her hear.** She finally started to babble with more intent. She could babble using sentence inflection, but not real words. She mostly used vowels.

At 1-year-old, Morgan wasn't walking yet. She crawled everywhere and would put some weight down on her legs, but she wouldn't try to let go of things to walk. Her doctors and therapists attributed this to her vision, because her leg muscles were strong. Morgan is legally blind in both eyes and has a cataract over her left eye that right now is not operable. She couldn't see exactly where she was going and had a depth perception problem. **Two weeks after her 2nd birthday, Morgan finally walked!** We bribed her with food (she loves to eat). **Morgan proved the doctors wrong again. If they say it can't be done, she does it anyway!**

Morgan was doing great! She was learning how to cope with her vision. No one can tell now that she is legally blind. Her vision therapist even said, "What do you need me for?" because she learned to cope all on her own. It wasn't something we taught her, but we let her explore and find her way. She still has a little problem with depth perception on curbs and stairs and texture changes on the floor, but if I know her, she'll get it!

Continued on Page 7

Morgan's Update, GA2… cont'd

We now had discovered a problem that covered a lot of areas in her life, sensory integration. First of all, she couldn't be in large crowds or noisy places. She would scream and kick, and at first we couldn't figure out why. The therapists said that it is like us going to a crowded mall at Christmas time, but 10 times worse for her. So, we have to adjust our schedules around less crowded times if we go out, or if she gets upset, we have to take her out of the situation immediately to calm her anxiety. Another problem was she couldn't feel what are called the "smooth muscles" very well in her body. The smooth muscles are in the mouth, stomach and bladder. We noticed she would stuff her mouth with food and then eat, and eat and eat. They said she does this because she can't feel things very well and doesn't know when to stop. We have tried to help this by using spicier foods. She also isn't potty trained yet at 3 ½, because she can't feel that she needs to go to the bathroom. I'm sure it will come in time. The sensory integration also has to do with her body in space. She doesn't like to be in a wagon, on a bike or on a swing, because she can't figure out where her body is in that space. We have worked on this by just putting her on these things, and letting her be the judge of what she wants to do.

At almost the age of 3, we discovered another problem. When we would go outside in 70, 80 and 90-degree weather, Morgan's whole body would turn really red. She would get a little puffy, too. When we would bring her in the air conditioning, she would start to sweat. Then, it would take her hours to turn back to a normal color. After speaking with her neurologist, he believes that **the hypothalamus may have been damaged during her brain hemorrhages earlier in her life. The hypothalamus controls the temperature in your body.** He thinks that Morgan's can't control the heat in her body and make it sweat properly, and this can cause a heat stoke. So as a result, Morgan can't be out in the summer heat above 75 to 80 (depending on other conditions).

Other than that, she had a great year. She wasn't sick or in the hospital for a whole year! But then **around Christmas of 2002, Morgan started having 20-second seizures.** We weren't sure why because she had been seizure free for 3 years. Then, we noticed Morgan, who was normally bouncing around the house, was lethargic and didn't want to do anything. We made an appointment to see her neurologist, and he put her back on her seizure medication that she had been off of for 2 years. The seizures increased from one a day to 4 or 5 a day. And then, on December 28, she was in the bathtub, and started staring. She wouldn't come out of it. We called 911 for an ambulance, and then she started jerking her arm and head. We realized she was having a seizure. The rescue team gave her oxygen, but wasn't allowed to do anything else to stop the seizure. She ended up seizing for 45 minutes. From the hospital, they flew her to the hospital in Cincinnati, where they could-n't determine what was wrong. They put her on more medication and sent us home. The medication made Morgan have erratic and irrational behavior. **But after a tumultuous 2 months, the medication stopped the seizures completely, and she returned to her normal playful self.**

Again things are great! She is going to preschool two half-days a week to get all of her therapy, and she loves it. Her behavior is great (for a three-year-old). She is still slow on her speech, though. Her doctor said the other day that he was really worried that her speech wouldn't come, but **she has proved him wrong before! I'm sure she will do it again!!**

We thank God for her. She is our little miracle child who has the determination to overcome any obstacle.

April and Chad Jones
Grayson, KY
ajones@carter.k12.ky.us

Pharmaceutical Update

Sigma-Tau Pharmaceuticals, Inc., makers of Carnitor®, can be reached at 1-800-447-0169 or on their website http://www.sigmataupharma.com/. Their Carnitor® website at **www.carnitor.com** helps to Educate and Empower Providers and Consumers by providing important information about carnitine deficiency and supplementation with Carnitor®.

Professional Questionnaire for Referral Purposes

All Medical/Health Professionals: *Please complete the Questionnaire on our website www.fodsupport.org under 'Online Forms' (even if you are already listed on our printed Professional List ~ this is an Update)* ***if you would like Families to know that you diagnose, clinically treat and/or do research with Children and/or Adults with Fatty Oxidation Disorders.***

Please return to Deb Lee Gould via email or regular mail.
deb@fodsupport.org or 805 Montrose Drive Greensboro, NC 27410

Medical Update

LCHAD…*or is it*… TFP? : An Update

Jamal A. Ibdah, MD, PhD
Associate Professor in Internal Medicine/Gastroenterology,
Pediatrics and Biochemistry
Wake Forest University School of Medicine
Winston-Salem, NC

In a late evening few months ago, I opened my e-mail to find a message from a caring mother that read in part **"My daughter…is diagnosed with TFP…She was diagnosed in 1993...as LCHAD and several months ago was diagnosed as having TFP. I have done some research on the computer about TFP, but find it keeps referring me to LCHAD. Do you have a good description of TFP, for me, for anyone in the medical field whom I come in contact with (who has never heard of TFP...which is everyone), and for our FOD support group and for a grass roots group called Save Babies…?"** In this article, I will attempt to first clarify the difference between LCHAD and TFP, which, I admit, can be confusing. Second, I will provide an update of what we have learned about these disorders in the past few years.

LCHAD and TFP: What are they?

Both LCHAD and TFP are proteins (enzymes) that are necessary for fatty acid breakdown in the cell. However, LCHAD is actually part of TFP (which is an abbreviation for trifunctional protein). TFP is simply a complex protein that is composed of LCHAD and other 2 enzymes that are, like LCHAD, necessary to break down fatty acids. This knowledge of the relation between TFP and LCHAD was not known in 1989, when the first case of LCHAD deficiency was described. The fact that LCHAD is part of TFP became known in 1992. After that, it became clear that some of the children who were initially diagnosed as LCHAD, in fact, had TFP deficiency. In MTP deficiency, all the 3 enzymes, including LCHAD, are deficient. So, there are 2 groups of LCHAD children: The first with deficiency of LCHAD alone, and the second with deficiency of LCHAD and the other 2 enzymes. The second group is labeled with TFP deficiency since all the 3 enzymes in TFP are deficient. The majority of LCHAD children belong to the first group (only LCHAD affected). Over the past few years we learned several lessons about LCHAD and TFP disorders.

Lessons from Kids

In 1999, we reported our findings in 24 children with LCHAD and TFP disorders. By enzymatic measurement, we found that 19 of these children had LCHAD deficiency and 5 had TFP deficiency. Although there was some overlap in the clinical features, we noted that, in general, children with LCHAD deficiency presented at few months of age (5 months on average) with liver abnormalities due to accumulation of fat in the liver. A severe form of TFP deficiency affected 3 children who presented primarily with heart problems in the first 2 weeks of life. We found a mild form of TFP deficiency in the remaining two children who presented at older age (more than 2 years old) primarily with problems in the muscles of the extremities and some neurological abnormalities. Our molecular analysis in these children revealed some important findings. A common mutation for LCHAD was found in all children with LCHAD deficiency either in both copies of the gene or in one copy (the one from the mother or the father). This mutation was not found in any of the 5 children with TFP deficiency. Our research in additional families confirmed these findings. It should also be noted here that few of these children had sudden unexpected death and initially labeled as SIDS.

Lessons from Women

Our research in the past few years has provided an important understanding to an association between LCHAD deficiency in the child and development of liver disease in the pregnant mother while carrying the affected child. The liver disease late in pregnancy is called acute fatty liver of pregnancy "abbreviated as AFLP" which is difficult to distinguish, sometimes, from another condition called HELLP syndrome, which also causes liver disease late in pregnancy and reported to occur in association with carrying a child with LCHAD deficiency. In our families, we found that approximately 70% of the women who carry children with LCHAD deficiency develop AFLP or, to a less extent, HELLP syndrome. In all of these cases, the unborn child had the common mutation in one or both copies of the LCHAD gene. None of the mothers in the families who had children with TFP deficiency had AFLP or HELLP syndrome. This suggested to us that this association between liver disease in pregnancy and fatty acid oxidation disorders in children is primarily unique to LCHAD deficiency. However, we should caution that there are few reports of an association between these pregnancy diseases and other fatty acid oxidation disorders including TFP.

In a recent study of 27 cases of AFLP and 81 cases of HELLP syndrome, we screened the women and the newborns for the LCHAD common mutation based on the mother's history. In 5 of the 27 pregnancies complicated by the AFLP, the child was LCHAD deficient with either one or two copies of the LCHAD common mutation. This finding led us to an important recommendation: newborns should be screened for the LCHAD common mutation in all pregnancies complicated by AFLP. This is critical because screening the newborn can be life saving, such that a diagnosis will be established before development of the disease, which is potentially treatable. These disorders are treated by dietary modifications including frequent feedings, high carbohydrate and low fat diet, and replacement of fat with "medium chain" fatty acids that do not require LCHAD and MTP for their breakdown. In addition, screening the newborn in these complicated pregnancies and establishing a diagnosis of LCHAD deficiency can be important for genetic counseling including prenatal diagnosis in future pregnancies.

Acknowledgments

Drs. Arnold Strauss (Vanderbilt University), Michael Bennett (University of Texas-Southwestern), and Piero Rinaldo (Mayo Clinic) contributed to the work summarized in this article.

Continued on Page 9

Medical Update

Health Insurance Portability and Accountability Act (HIPAA)

Lynne A. Wolfe, MS, APN, BC
LAWPNP@aol.com

In 1996 the Health Insurance Portability and Accountability Act, commonly known as HIPAA, was passed, but it did not go into effect until late 2001. Its most recent amendments went into effect on April 14, 2003. It is considered the most significant piece of Healthcare Legislation passed since Medicare was approved in1965. **HIPAA sets a national minimum standard of basic privacy protections, but does not supersede stricter, already existing State Laws regarding privacy and Public Health.** Likewise, HIPAA does not specify specific organizational policies or security practices. Each Organization has some flexibility in implementing the HIPAA regulations according to their corporate culture, size, and available resources, so long as the HIPAA requirements are met. Additionally, not complying with HIPAA costs $100 for each violation to a maximum of $25,000 per year/per specific provision. So there is plenty of motivation to comply with this new law.

It has been generally accepted that **medical information is among the most sensitive and personal information collected and shared, and that privacy and protection of that information is central to the Provider-Patient relationship.** And, yet, there is no comprehensive Federal Law protecting those beliefs, and existing State Laws are inconsistent and piece meal, usually focusing on specific medical conditions (i.e. Communicable diseases), populations (i.e., Abused children), and/or organizations (i.e., Planned Parenthood).

New urgency exists for privacy protection with the rapid increase in electronic information and communications technologies, rise in managed care, concerns raised by the soon to be completed Human Genome project, the increased demand for health data, and the commercial use of health data for marketing purposes. Secondary users of healthcare information include: Drug and Medical device marketers, Public Assistance Programs, Law Enforcement Agencies, Courts, and Private Database Companies, such as the Medical Information Bureau. **The overriding concern by many is the potential that increased access will create increased risk of misuse by those who are not bound by ethical or legal standards, those motivated by profit and/or those motivated by curiosity.**

So who is affected by HIPAA? The legislation uses the collective name "Covered Entities" to specify those directly affected by this act. Covered entities include: Healthcare providers (Hospitals, Clinics, and individual Doctors), Health Plans (Medicare, Medicaid and Veteran's Healthcare Programs), Laboratories, Pharmacies, Healthcare Information Clearinghouses, Billing Agencies, and their business associates - such as contracted Accountants, Lawyers etc…who transmit any health information electronically. Although protected health information transmitted in any form (electronic, written, or oral) is covered. HIPAA also covers: how and what information can be used for health and medical research, limits the use of health information for marketing and fundraising activities, and describes how health information can be obtained and used for Public Health purposes.

Who is not affected by HIPAA? Employers, Auto and Life Insurers, Worker's Compensation programs, Social Security and Welfare Benefit programs, disclosures required by Law which may include Domestic Abuse, Drug Abuse, and/or Communicable public health diseases.

What is Protected Health Information (PHI)? Protected Health Information (PHI) relates to past, present, or future health conditions of an individual, except those related to treatment, payment, or healthcare operations, or those specifically authorized by the patient.

Protected Health Information (PHI) includes18 identifiers: Name, Address (other than State), Telephone/Fax numbers, Certificate/License #'s, Web URL's / Internet Protocols (IP), Email Addresses, Any dates within Medical Record (excluding year of birth, death etc…), Health Plan #'s, Vehicle Identifiers, Any unique ID #, Medical Record #, Social Security #, Account #'s, Medical Device Identifiers, Biometric Identifiers (Voice, Fingerprints, Retinal scans), Full Face Photos.

What is meant by Treatment? It is the provision, coordination, or management of health care, and related services, by one or more healthcare Providers, including consultation or referral. So **for families caring for children with special health needs who see multiple Providers and need information shared, nothing should change.**

What is meant by Payment? Collection of premiums, reimbursement, billing, claims management, medical necessity determination, utilization reviews, and pre-authorizations. **Again, you should not see any change in these services.**

What is meant by Healthcare Operations? Internal provider programs such as: Quality Improvement, Peer Review, Training and Credentialing of Providers, Business Planning and management activities can utilize Protected Health Information **so long as this use is disclosed up front to all individuals seeking healthcare services from a certain Provider.**

HIPAA does require that a covered entity (Healthcare Provider) must make reasonable efforts to limit uses, disclosures, and requests for protected health information to the minimum necessary to accomplish the intended purpose. This means that **internal uses of protected health information must be classified on a "need-to-know" basis with appropriate controls over who has access to the protected health information.** There is an allowment for developing standard protocols to be used for routine and recurring disclosures such as billing. For non-routine disclosures, a covered entity must develop and apply criteria for determining the minimum necessary amount required, and a mechanism to notify the patient when such a disclosure has occurred.

What are the Individual Patient Rights under HIPAA? **You have the right to written notification of privacy practices by all "covered entities," including a full accounting of their disclosures of your protected health information.** This is creating a lot of extra paperwork, as we have been required to sign that we have received these notices at our Doctors Offices, Pharmacies etc…but **I do encourage all of you to read them. You have the right to access, copy, and request amendments to your Medical record.** However, **you may be charged copy fees, and requests for amendments do not have to be honored by your Healthcare provider.** Although, any refused amendments have to be explained to you in writing, and **there are appeals processes in place if you feel strongly that information in your Medical Record is incorrect.** You can request restrictions in use and/or disclosure of your health information, although again your Healthcare provider does not have to comply with your wishes. Again any denial of requested restrictions or disclosures must be given to you in writing and you do have the right to appeal the decision. You have the right to request communication means and **the right to file complaints if you feel the HIPAA protections have not been followed.**

Continued on Page 10

As part of the HIPAA legislation **there is a Security Rule that requires covered entities to establish administrative, physical and technical safeguards, to control access to health information in order to ensure: 1) Confidentiality – No accidental or intentional disclosure to unauthorized recipients, 2) Integrity – Data has not been altered or destroyed in an unauthorized manner, and 3) Availability – Accessible and useable upon demand by an authorized entity.**

What are your likely to experience? Many hospital information personnel will give out only room number and general condition of a certain patient if you ask for them by full name. For other patients, specific information will not be given over the phone. Postcards with scheduled appointment reminders and lab results will now come in envelopes. There will be more use of first initials and full last names on Clinic Sign-In sheets, as well as when being called for Clinic appointments in a Waiting Room, "White" Boards and Patient Rooms on the Hospital units will also be limited to first initials, last names, and perhaps the Doctor's name. **You should see more strict controls on where conversations occur, especially in the hospital setting.** For example, if you or your child are in a semi-private room, your Doctor should draw the curtain and speak quietly, or take you somewhere private versus potentially allowing someone to over hear protected health information. Hallway conversations and rounds will be limited. **Stricter control over who accesses your Medical Record will occur.** Limitations will be placed on leaving detailed messages on answering machines. Any protected health information that is to be discarded will now be shredded.
Stricter security access and monitoring will occur related to computer access of Patient Medical records, as well as the faxing and emailing of health related information. Hence the new disclaimers at the bottom of many fax and email messages.

INFORMATION SOURCES: www.healthprivacy.org and www.hhs.gov/orc/hippa

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NBS Update

April 07, 2003 Contact: HRSA Press Office, 301-443-3376 ~
HRSA Awards $1.5 Million to Expand Newborn Screening HRSA has awarded $1.5 million in first-year funding for five projects to expand and strengthen newborn screening systems and promote ongoing screening of children with special health care needs:
•**Georgia Department of Human Resources, Atlanta,** received $295,522 to design and implement a statewide Newborn Surveillance and Tracking Integrated Information System to help ensure that children with or at risk for inheritable disorders are identified and monitored.
•**Colorado Department of Public Health and Environment, Denver**, was awarded $300,000 to implement a state genetics plan to improve and coordinate services for children with special health care needs, in particular those children identified by newborn metabolic and hearing screening and infants coming out of hospital neonatal intensive care units.
•**Minnesota Department of Health, St. Paul,** received a grant of $300,000 to develop a coordinated newborn screening and follow-up system through program and data integration.
•**Oregon Department of Human Services, Portland,** was awarded $300,000 to help hospitals, medical homes, and public health programs identify newborns that need follow-up and to coordinate and monitor service delivery.
•**The University of Tennessee Graduate School of Medicine, Knoxville**, received a grant of $300,000 to integrate and improve systems of care for newborn screening by developing ways to track and evaluate system effectiveness.
Each of these four-year grants is awarded through Special Projects of Regional and National Significance – a program of HRSA's Maternal and Child Health Bureau that supports a broad range of innovative maternal and child health improvement strategies.

• • •

Families in Canada are also becoming aware of the **importance of expanding their Newborn Screening**. One of our newest Families is trying to **network with other Canadians affected by an FOD (or one of the other metabolic disorders that can be detected through tandem mass spectrometry).** Tammy Clark (rclark@storm.ca) recently experienced the **tragic death of her 9-month old daughter, Jenna (**Please visit Jenna's story at www3.sympatico.ca/tammy_roger/Jennastory.htm**), because her MCAD was NOT diagnosed at birth.** She, like many of our Families that have gone through a similar experience, feels and KNOWS that **NO FAMILY should have to have a child die!** And if she lived in Saskatchewan (a province that currently screens using tandem mass spec), **Jenna most likely would still be alive!** Unfortunately it's similar in some states in the US ~ **it should NOT matter where you live ~ ALL babies should be screened AT BIRTH ~ and NOT through an autopsy!** Medical Professionals also NEED to become aware of disorders like FODs in order to **broaden their Differential Diagnoses skills** (i.e. when a parent brings a child to the ER) so that they can **move beyond the 'It's ONLY the flu' type of diagnosis!** Too many of us have heard that before!
If you would like to **join forces with Tammy and Dorene Bellaire (Canadanana49@aol.com), whose granddaughter, Alyssa, also died from Undiagnosed MCAD, and get the word out about expanded NBS in Canada,** please contact them directly ~ **change IS possible because 'We Are All in This Together!'**

Love Messages

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

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

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

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

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

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

Jennifer and Bill Boucher
Alyssa - Birth Nov 18, 1999 Death July 22, 2000

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

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

Barry and Julie Bryson
Skyler - Born November 25, 1989 Death Aug 25, 1990

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

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

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

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

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

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

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

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

Sandy and Jon Cooper
Noah - Birth Oct 5, 2001 Death June 30, 1998

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

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

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

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

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

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

Shelly and William Grabow
Caleb - Birth Sept 14, 2001 Death Sept 27, 2001

Brandis Greichunos
Madison Burchette - Birth March 8, 2001 Death March 24, 2002

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

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

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

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

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

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

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

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

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

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

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

Stephanie and Doug Huber
Jace - Birth March 8, 2000 Death Feb 14, 2001

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

Karen and Steve Imhoff
Michael - Birth July 25, 1991 Death July 8, 2002

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

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

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

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

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

Sondra Koehn
Darcy - Birth Aug 10, 2000 Death March 19, 2002

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

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

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

Darlene and Larry Lopez
Marissa - Death Feb, 1999

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

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

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

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

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

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

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

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

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

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

Diana and Kevin Patterson
Trevor - Birth Aug 30, 1997 Death Mar 4, 1998

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

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

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

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

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

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

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

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

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

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

Lisa and Scott Sleezer
Emily - Birth March 5, 1998 Death June 18, 2001

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

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

Anne and Gary Stitt
Sydney - Birth Oct 13, 1995 Death May 20, 1996
Lisa and Doug Tennyson
Sammy - Birth Aug 23, 2000 Death Dec 21, 2001

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

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

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

Sirpa and Jay Waananen
Nora - Birth Mar 29, 2001 Death Aug 9, 2001

Jenni Wagoner
Lauren - Birth Oct 26, 1993 Death Nov 13, 1999

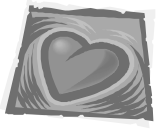
Richard and Amy Warner
Andrew - Birth May 1978 Death Nov 18, 1979
Scott - Birth May 1983 Death April 25, 1985

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

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

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

Christi and Ronnie Williams
Preston - Birth Mar 11, 2000 Death Mar 15, 2000



'Intense love does not measure, it just gives'

~ Mother Teresa

Successful Pampered Chef 'Fundraiser'

THANKS so much to Sheri Merrill (Kristen 5, MCAD and Jamie 3, Carrier) for coordinating the Pampered Chef fundraiser! Along with Maureen Pienta, Michelle Bray, Cindy Wilt, and Lori Michaud, **a total of $744.23 was raised for our Group!** What a WONDERFUL effort to help our Families ~ all of the funds will go toward providing new Family Packets, as well as for this year's only printed newsletter issue. **THANK YOU for thinking of us!**

Deb's Updated Email Address
Please update your address book ~
Main address is deb@fodsupport.org
Backup address at fodgroup@triad.rr.com

Welcome to new babies!

Shelly and William Grabow welcomed Caden (brother to Caleb, undiagnosed TFP) on December 31, 2002. He weighed in at 6lbs 9.1 oz.

Carsen Richards (brother to Caden, GA2) was born June 9, 2003. *Krystena and Mike*, and half-siblings, Warren Prater and Jaime Richards are excited to have Carsen home! He weighed 6lbs 9oz and 19" long.

We have a brand new baby brother!! His name is Jakob Garyrobert. His first name is from the bible and his middle name after our Grandpa Stahr and our Grandpa Fisher. Jakob was born on January 22, 2003 at 12:22pm while we were all in Minnesota at the Mayo clinic. Daddy was there when it happened and even video taped it, of course Mommy won't let anyone watch it. Jakob weighed 6lbs and 2oz and he was 18in long. He is a pretty good little brother even though he does bite your nose if you get to close to him when he's hungry!

Isabelle and Rebekah Fisher, mom and dad Sharon and Tyler

•• ***Signing with your baby before they learn to talk*** by Victor Garcia, 2001. $12.00
•• ***Inclusion Research Institute*** of Washington, DC publishes a weekly report ~ you can subscribe for the free report at http://www.inclusionresearch.org/index.htm. They bring to you the events of importance to parents, children, and individuals with disabilities. Please send your comments to mmwdc@inclusionresearch.org. "We are committed to working for full inclusion in communities of all people with disabilities, including the most significant disabilities, according to principles of self-determination."
•• ***The U.S. Department of Transportation*** is calling on all disability organizations to promote public education about its **Toll Free Hotline for air travelers with disabilities.** The Toll Free Hotline for disabled air travelers has been in operation since August 2002 and is available for callers from 7 am - 11 pm Eastern Time, seven days a week. **The Hotline serves two main purposes: (1) education and (2) assistance in resolving disability-related air travel problems.**
Call the Toll Free Hotline 7 am - 11 pm EST
1-800-778-4838 (Voice)
1-800-455-9880 (TTY)
*Many disabled air travelers are not aware of their rights and the Hotline, in part, exists as an educational service to inform air travelers with disabilities about their rights under the Air Carrier Access Act and the Department's implementing regulations 14 CFR Part 382 (Part 382). Hotline operators are well versed in the ACAA and Part 382 and can provide callers with on the spot general information about the rights of air travelers with disabilities. The Hotline operators also respond to requests for printed consumer information about air travel rights of the disabled.
*The Hotline can also assist air travelers with disabilities in resolving real time or upcoming issues with air carriers. The purpose of "real-time" assistance is to facilitate airline compliance with DOT's rules by suggesting to the passenger and the airline involved alternative customer-service solutions to the problem. The airline remains responsible for deciding what action will be taken to resolve the issue in accordance with the ACAA and Part 382. Generally, if a caller has a real time problem or an upcoming issue with an air carrier, a Hotline Duty Officer will contact that air carrier and attempt to resolve the issue. For example, there have been a number of incidents in which Hotline Duty Officers have contacted air carriers and convinced them to accept service animals and electric wheelchairs on board flights, to stow folding wheelchairs in the cabin, and to provide requested wheelchair assistance. Air travelers who want information about the rights of persons with disabilities in air travel or who experience disability-related air travel service problems may call the Hotline to obtain assistance. Air travelers who want DOT to investigate a complaint about a disability-related issue still must submit their complaint in writing via e-mail at aircon-sumer@ost.dot.gov or postal mail to:
Aviation Consumer Protection Division
U.S. Department of Transportation
400 7th Street, S.W.
Washington, D.C. 20590

Recipes

TEX MEX SOUP

1 pound lean ground turkey
1 package (1 ounce) reduced sodium taco seasoning mix
1 package (1 ounce) ranch salad dressing mix
1 can (15 ounces) black beans
1 cup frozen corn
1 can (15.5 ounces) northern beans
1 can (15.5 ounces) pinto beans
16oz broth or water (or 1can lite beer for the adults!)
1 can (14 ½ ounces) tomatoes with chilies
1 can (14 ½ounces) stewed tomatoes

1.Brown turkey in a large stockpot. Drain excess fat. Add taco and ranch seasoning mix to turkey
2.Drain and rinse canned beans. Add to turkey in stockpot. Add all remaining ingredients. Bring to a boil. Serve hot. 8 servings. Serving Size: 1½ Cups. Calories: 321, protein 21g, Carbohydrate 43g, Total Fat 6g, Sodium 800mg.
Sheri Merrill (*Kristen 5, MCAD and Jamie 3, Carrier)*

Very Low fat Hot Dog Roll-Ups

1 roll of refrigerated low fat crescent rolls
1 package of fat free hot dogs
Fat free cheese slices

Divide crescent roll dough into triangles. Place a hot dog and ½ slice of cheese on the dough and roll up as directed on the package. Cook according to the directions on the crescent roll package - usually about 12 minutes at 350°.
Shelly Garbo

Kids Korner

Mary Flahive
(MCAD)

Nicholas Cywinski
(MCAD)

Thomas Joshua Calk
(GA2)

Family & Professional Donations

Family Donations: Sara and Steven Cywinski in honor of Nicholas (MCAD). Pampered Chef donations (THANKS Sheri Merrill for coordinating this fundraiser!) ~ Maureen Pienta, Michelle Bray, Cindy Wilt, and Lori Michaud ~ totaling $744.23! Mary Thorson in honor of Wendy (TFP) ~ several AT&T calling cards that I (Deb) use to contact new Families.

Professional Donations: Sigma-Tau Pharmaceuticals, Inc. (makers of Carnitor®)

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

Thank You

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

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

Reminders

Families - Please send **TYPED** stories by **DEC 1, 2003.** 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 **DEC 1, 2003**. Also, please return to Deb the **Professional Questionnaire** even if you are already listed on the printed Professional List.

"Take the first step in faith. You don't have to see the whole staircase, just take the first step."

--Martin Luther King, Jr.

Communicate With Us

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

_____

_____

_____

_____

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

_____

_____

_____

_____

Please include ideas for future issues or your questions