

# FOD

Support

fatty oxidation disorder communication network



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We hope all of you are enjoying the summer months and staying as cool as possible. Now that the FOD Group is an official 501c3 non-profit, we'll need to get 'HOT' as far as raising funds for our 2008 National Metabolic Conference! It would be terrific if everyone could look over our homepage and read how each of us can help raise the money needed to offer an exciting opportunity to all our Families for learning about FODs, as well as for networking with others from around the world. Kathy Stagni, Director of the Organic Acidemia Association (OAA) and I are exploring whether to have our own conference as we did in Texas in 2006, or join forces with the United Mitochondrial Disease Foundation (UMDF) ~ we will keep everyone posted on what we decide. A lot of it depends on whether we have enough funds to even offer a conference ~ so **PLEASE share the '2007 Annual FOD Letter of Giving' with your Families and Friends** ~ all donations are tax deductible and a receipt will be mailed acknowledging their donation. Please make checks (US currency) out to the 'FOD Family Support Group' and mail to our Michigan address above. Because we don't have the funds and we are waiting to be registered in various states, we cannot 'snail mail' the Letter, so please pass it along to anyone you can! You can also **donate directly from our homepage ~ we have a PayPal link** on the right sidebar. You can donate to the General FOD Fund (which will cover day-to-day costs and conferences), a Clinical Trust

Fund (to raise funds to help clinically train new Drs/professionals in the field of metabolism and genetics), a Research Trust Fund (to raise funds for FOD research), and an option for grief consulting donations if you choose to 'work' on a grief issue with me (more info is on our homepage) ~ all of these options can be given in honor or memory of a loved one.

Along with the Letter of Giving and other 'fundraisers' on our homepage (cafepress, iGive, etc), you can add our Group's name to your local **United Way contribution program**. Using their form, go to the space where you can designate an agency and write in 'FOD Family Support Group.' Include our address, Tax ID # and other needed info (see letterhead above). There are some chapters that only designate to local groups but you can try writing in our Group. Every donation counts ~ small or large!

And if you're thinking there is nothing YOU can do to 'fundraise' be sure and read the articles in this issue about how the Archetti and Shannon Families raised funds for a hospital, as well as for our Group ~ what a terrific job they did! Be aware, however, that if you're planning a Family FOD Awareness 'Fundraiser' to benefit the FOD Group, please be sure to print off and mail me the '**Family Project for FOD Awareness Form**' on our homepage. **This form just states that you acknowledge that the FOD Group is not sponsoring your project.**

With nonprofit status, comes extra bills for insurance, accounting help, conference costs, etc ~ **we hope you will be as generous as possible ~ since we are an ALL volunteer Group no one has a salary ~ every cent raised helps our Families!**

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, 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.

We also would like to hear from our Professionals ~ we always welcome new Medical, Research, Nutritional, Counseling, etc articles. Whether you're a Family or a Professional, we are all striving to create awareness, education, screening and diagnosis, long-term clinical treatment, and research ~ by sharing your story or your expertise...

**'We Are All in This Together!'**

Take care... DLG





# Editorial

## FOD Concerns Need to be Addressed

Over the course of the last 17 years, I've had the opportunity to talk with MANY new FOD Families. It's a great way to share what I've learned over 22 years of living with the death of an undiagnosed daughter and with a surviving son with MCAD, as well as try to answer the many questions they might have and also to listen to their many fears. From those conversations, I've noticed several themes that keep recurring that I feel all of us (Families and Professionals) need to be aware of. Many of these issues are addressed on our site and in past newsletters, as well as over our Email List (with 450 members), but I thought if they were posted here they might be a way to create a focused dialogue between ourselves and those that we come in contact with in regard to our child's/our disorders. Even though EACH disorder has differences and EACH affected person responds differently to their disorder, the following points impact us all ~ affected and unaffected. This is not a complete list of concerns (others include being accused of munchausen and making your child sick, not being able to get coverage for formula or other supplements, school and therapy issues, etc), but it's a starting point for all of us to discuss with our Families and medical professionals. Feel free to cite any of these points in your discussions with others or when you might be presenting your own story in front of medical students ~ the more that are aware of these concerns the better ~ **COMMUNICATION and PROACTIVE BEHAVIOR are KEY when living with a rare disorder and ineffective communication and passive behavior can be fatal in some circumstances!** It would be great if you would create your own list of concerns and share it with us too ~ I can post more concerns/issues and your own positive or not-so-positive comments in future newsletters.

- With **expanded newborn screening (enbs)** spreading across the country (albeit too slowly for me!), there are several issues: 1) states/countries/hospitals that do NOT yet screen for the 'latest' full panel (that's another whole issue in itself – what they consider a full panel!) **NEED to tell parents that it's still possible to get the expanded nbs and where they can send the filtered paper!** [refer to our **Med Info page, Diagnostic Labs**] 2) be aware that the enbs is just that - a screening tool – it is not the Diagnostic test that will be done when the 'redflag' screen shows some abnormalities ~ even though both use the tandem mass spectrometry instrument, there are different ranges for the enbs and the diagnostic tests. 3) also be aware that there **may be false positives** in regard to the screening ~ this is where I hear the most disturbing comments from parents [note that this doesn't happen every time but even ONCE is too much!]

For example, and I've heard this several times, parents will receive a call on a Friday and the message is left on their answering machine that their infant "has a fatal disease and we need to see him/her immediately!" – but by the time they call the office back the staff has left for the weekend – so the parents are FRANTIC and DEVASTATED about this call and not sure what to do or whom to call! First of all, stating that the infant HAS the disorder is not fully accurate UNTIL the follow-up DIAGNOSTIC test is performed (and sometimes even a further skin biopsy may be needed as well to confirm a diagnosis), and it's HOW the positive screen information is conveyed (by the staff/genetic counselor or other professional) – I do NOT feel it's appropriate to leave a frightening message on an answering machine and then not have a number the parents can call (even over a weekend) to get the FULL information.

Some parents were told NOTHING about the FODs and what they might be able to proactively do while waiting for a follow-up DIAGNOSTIC test and the results (i.e. make sure to feed their infant often, avoid fasting, watch for vomiting, what might be done in a possible crisis, etc). Families have also had to endure weeks of waiting for results – only to find out that the staff knew the results 2 weeks before but failed to convey that to the Families! Of course they were relieved to find out their child DIDN'T have an FOD but the PROCESS to get to that point was HORRENDOUS for them!

Professionals --- if you see yourself in this scenario PLEASE be aware of HOW and WHEN you communicate a positive screen to a Family- it's important to convey the information without frightening them to death or putting them into panic mode by not giving them enough info. Again - Effective Communication is KEY!

4) Additionally, too often moms are made to feel guilty for supplementing with formula (versus just breastfeeding) after giving birth – we need to get the information out to medical professionals that **that formula COULD be the difference between LIFE AND DEATH for some infants while waiting for the results of the enbs!** Encouraging breastfeeding is important BUT being so adamant about it and telling moms to ONLY breastfeed can be very dangerous to those yet-to-be diagnosed infants with an FOD.

- Another set of issues that many Families have discussed as I speak with them over the phone and also over our Email List, is the **use of the terms 'mild' and 'non-disorder' in regard to various FODs.** Although this topic will have further comments written in our Jan issue by one of our leading FOD experts, I felt it necessary to briefly address them here. Those infants being picked up via enbs most likely will NOT have the symptoms that many of our kids had (especially due to NOT being diagnosed at birth, some diagnosed after years of episodes or after their death) because they are being DIAGNOSED and TREATED from birth BEFORE ANY SYMPTOMS CAN OCCUR! And the parents KNOW what to do in an emergency BEFORE a serious problem can occur!

To be told their child has a 'mild' case ONLY based on the mutation they might carry makes new parents complacent – they think they don't have to follow any type of treatment protocol because the Dr told them it's a 'mild' case and the child shouldn't have any problems – they are being told it's not serious enough to be concerned about! That's a simplistic explanation (of the comments I have heard from many parents) on my part, but you get the point! **How in the world does someone KNOW FOR SURE AT BIRTH how a child will respond to their disorder or to a future illness or stress??? THAT – is what I am objecting to – NO ONE KNOWS FOR SURE!**

As for the **'your disorder is a NONdisorder and doesn't need to be treated'** issue: then why are the affected individuals so symptomatic??? We are hearing from SCAD Families that are being told SCAD is one of those NONdisorders – these comments really concern me. As Mary, our webmaster, commented to me, if someone was diagnosed with skin cancer, one that might be considered 'mild,' do you think they'd just let it go without treatment? Most likely NOT – they would treat it so it would not become more complicated and cause more medical problems and possibly a fatal outcome! **In my opinion, if one has a metabolic disorder they either HAVE IT or they don't** (just like being pregnant – you either are or you aren't) **so TREAT TO PREVENT COMPLICATIONS**—why get stuck on whether it MIGHT be a so-called 'mild' case or a so-called NONdisorder??

If the professionals don't think it's a REAL disorder then why don't they RENAME it to reflect what it truly IS just like they did with LCAD years ago – renaming and re-diagnosing many initially diagnosed with LCAD with VLCAD. The same applies to some initially diagnosed with LCHAD – only to be re-diagnosed with Trifunctional Protein Deficiency (TFP) a few years ago. Yes, some of our Families have a dual diagnosis of an FOD and a mito disorder – so why don't they do further testing on these NONdisorder kids and find out if there's something else going on – instead of just calling the SCAD a NONdisorder and telling the parents no further treatment is needed!?

(cont'd on page 3)

## Editorial...cont'd

So what's next – if SCAD is a so-called NONdisorder, are we going to have NBS Labs making the decision to NOT tell Families that their baby screened positive for it because it's NOT a REAL or SERIOUS disorder?? I would think that would be pretty dangerous to NOT tell parents. Also what impact will being labeled a NONdisorder have on the entire enbs process – will states be the ones to just drop the disorder because SOME THINK it's a NONdisorder!? Again, I think that would be setting a very dangerous precedent.

As many have learned, these disorders can 'wax and wane' over time -- who's to say some trigger won't put an affected individual in a crisis? **Individuals have DIFFERING THRESHOLDS for when they may present with symptoms.** Then the parents will happen to mention to the ER Drs that they were told their child/self had a 'mild' case or that their disorder was a NONdisorder and the staff may dismiss the seriousness of immediate treatment – **what are you going to say to that parent when that child/adult is medically damaged or died because it wasn't taken serious enough?!**

Lastly, one of those 'symptoms' that I hear a lot about is what many consider '**low blood sugar' or hypoglycemia** – I really think medical professionals **NEED to rethink that term in regard to FODs.** We've had many Families get to the ER only to be told to go home because their child's (or your) blood sugar was in the 60s or 70s and with fluids should be okay! As stated earlier – EACH affected person will respond DIFFERENTLY to DIFFERENT TRIGGERS and **blood sugar CANNOT BE TOTALLY RELIED ON to determine whether one is in crisis or not!** If you read my Jan 2006 newsletter Editorial you would know what I am referring to – when my son, Kevin (19 at the time, MCAD) was in crisis after oral surgery where the anesthesiologist TOTALLY DISREGARDED the 10% dextrose IV protocol and only gave him 5% – even though his BS was 76 (which most consider close to normal range of 80-110) it was NOT at the NECESSARY 100-120 level to keep him from going into crisis from such a stress – and that ARROGANCE put Kevin in the ICU!! NEW Parents – you will need to learn that waiting for the blood sugar to drop to 50 or below to go to the hospital (which is often what medical professionals call hypoglycemia) could be VERY DANGEROUS for an FOD individual. **We have to create an awareness about how 'normal' blood sugar levels don't always relate to FODs!**

I'll stop there for now – but it really bothers me that some are being so 'casual' about SCAD and some other FODs, as well as these other FOD concerns. I hope our FOD expert will be able to further expand on some of my comments (most likely in a more medically professional way – since I am not a medical person!) for the Jan newsletter.

Deb Lee Gould, MEd  
Director, FOD Family Support Group



### **VLCAD, LCHAD and GA2/MADD Email Networks**

Gina R. (Brett, VLCAD) has started an FOD subgroup Email List for VLCAD families. If you are interested in networking with other VLCAD families around the world please email Gina at [ginamjb@optonline.net](mailto:ginamjb@optonline.net) or call her at (845) 928-9574.

Valerie Fulton (Adam, LCHAD, <http://adamslchad.com> ) is also networking many of our LCHAD Families. If you'd like to become a part of her email network contact Valerie at [vallchadmom@yahoo.com](mailto:vallchadmom@yahoo.com).

Terilyn Peterson (Jackson, GA2/MADD) has formed an online GA2/MADD list. If you are interested in networking with other GA2/MADD families around the world please email Terilyn at [irishgemin@gmail.com](mailto:irishgemin@gmail.com) or call her at (715) 299-0435.



# Family Stories

## Rebecca's Story ~ possible SCAD

Almost 14 years ago, I was finishing my pediatric residency and expecting my third child. I thought being a pregnant mother and doctor-in-training was hard...until Rebecca was born. I already knew something was wrong in my eighth month of pregnancy, when I had pre-term contractions. The early labor stopped with some bed rest and fluids, but I learned that Rebecca had only one kidney (pelvic kidney, which is a combination of both kidneys that stay joined in her lower abdomen).

Even though I was told that this is a common and harmless condition, I KNEW something was really wrong with my daughter. She had an abnormal umbilical cord, which we did not know about until it torn as she was born. Fortunately, she only needed to take iron. She did not need any transfusions. She went home in the usual 2 days and seemed to be fine for the next few weeks. Then, she had a slight fever, and was never the same. She stopped gaining weight, and was floppy.

When she was 2 months old, I screamed at her doctor, "There is SOMETHING wrong with my daughter!" Fortunately, this very wonderful doctor really heard me. She did a complete work-up, even including a screen for organic acids. Everything was normal, except the urine organic acid test. Off we went to a metabolic specialist, who did a ton of tests, including more urine organic acid tests. Sure enough, all the urine organic acid tests were clearly abnormal, and suggested SCAD (short-chain acyl CoA dehydrogenase deficiency). However, her skin biopsy did not show a specific abnormality, and, supposedly, SCAD was ruled out.

Rebecca continued to gain weight painfully slowly, and vomited frequently. Since we did not have a metabolic diagnosis, we did not know how or what to feed her. Different doctors had different opinions - change her diet, nasogastric tube, gastrostomy tube. Meanwhile, she needed calories ASAP! I found a way to get them in, in a very unorthodox way. I gave her Ensure in a bottle, adding rice cereal to up the calories and lower the fat content. We gave it to her at night, too - she vomited so much during the day. She did start to gain, and even got a little chubby!

Meanwhile, she got a lot of therapy for her low muscle tone and developmental delays at a wonderful early intervention program. Fortunately, she rarely was ill, and did not require hospitalizations. I did notice that whenever she had a fever, she began to vomit uncontrollably. If I gave her Tylenol suppositories to bring down the fever right away, she would stop vomiting. She never went more than 8 hours without eating, because we fed her while she slept. Finally, at age 4½ years old, she started to eat, and refused the bottle. She wasn't as chubby any more, but she continued to grow and gain. She still drank a lot of Ensure, which was good, because she did not eat a wide range of foods. Her first food she asked to eat was chocolate cake, and we were so happy that she was eating, we didn't care whether it was nutritious or not!

Just when we thought that she was catching up, when she was 5, she had her first seizure. It was long and scary, and she needed to be hospitalized. She had 2 more seizures before the medication controlled them. A nice "side effect" of the medication, Tegretol, was to control her difficult moods.

Rebecca did nicely for the next 3 or 4 years, in her special education private school. But, when she was 9, she began falling asleep a lot in class. I started to look for a new school, did psychological testing, and did online research into metabolic diseases. I found out that there was new knowledge about SCAD, and that Rebecca could have a "variant" form of this disorder, even though we thought that it had been ruled out by her "essentially normal" skin biopsy.

I tried to find a new metabolic doctor to do further testing, but got bogged down in finding a really appropriate school for Rebecca. She got a late but helpful diagnosis for autism spectrum disorder. We found a wonderful psychiatrist to medicate her for severe anxiety and mood swings. (We stopped the seizure medication during this period, because she had not had a seizure in years, and it can cause sleepiness). This psychiatrist sent us to the NYU Epilepsy Center, where another truly terrific doctor recommended not just a video EEG to look for missed seizures, but also a sleep apnea test. The video EEG did not show any seizure activity, but the sleep apnea test showed that Rebecca would stop breathing every 13 seconds, and wake up. No wonder why she was so sleepy and irritable during the day!

She did not have very big tonsils or adenoids, and we knew that her low muscle tone and narrow mouth was at least part of the problem. So, we did not know whether removing her tonsils and adenoids would help. The ENT doctor predicted at least some improvement, so it was worth a try. She had the surgery last fall. We were delighted to see Rebecca feeling much more well-rested and cheerful within days of the operation.

This tonsillectomy was Rebecca's first surgery. I realized, when speaking to the anesthesiologist before the surgery, that it would be so much better if we could get a definitive diagnosis for Rebecca. I was so scared that she might have a bad reaction to the anesthesia. They actually changed the protocol, based on my worries, and she tolerated the surgery really well. I realized, however, that we are just guessing what is safest. We don't really know.

So, now, after a long struggle with her day-to-day issues, I am trying again to get THE diagnosis. It has been so frustrating and lonely, knowing something is wrong, but not knowing what it is exactly, and what to do about it. We are going to a new doctor next week, thanks to a recommendation by another parent in the FOD Support Group. I am very grateful to this mom, to Deb, and to all the FODsupport parents, for being there for all of us. This story is obviously still a work in progress, but the main things I learned are to always listen to your own instincts and to NEVER give up!



Alisa, mom to Rebecca, almost 14, "variant SCAD"?



# Family Stories

## Theodore and Katarina's Story ~ MCAD (UK)

Our story starts on 10th August 2005 when our much loved daughter Katerina, known as "Kitty" was born. She was a healthy weight of 8lb 6oz but would not feed well. When her BM went down to 3.6 at one day old, I more or less abandoned breast feeding and went over to bottles. She lost about 15% of her birth weight in the first week which was worrying but we weren't unduly concerned. She also seemed to be very cold all the time, even though it was middle of August. Kitty had the heel prick test at 5 days old. At this point we had no knowledge of what conditions were tested for at newborn screening. Only that it was recommended all children were tested.



On 19th August we got a phone call from Professor Clayton at Great Ormond Street Hospital (GOSH) in London. To say this call frightened us is an understatement. We were terrified. He told us that we had to immediately come to the hospital. Of course we dropped everything to rush up there. Once there he tried to explain the results of the heel prick test, although I don't think much of what he said really sunk in. He told us that Kitty had MCADD, a rare metabolic disorder. He asked me and my husband if we were cousins because of the rare nature of the disorder!

We made several visits to GOSH over the next few weeks. We met a dietitian and a metabolic nurse specialist. We also had to take our son Theodore, known as "Teddy" to be tested for MCADD. He was 2yrs old at the time and although he had been ill before with gastric bugs, we were sure that he didn't have the same condition. Imagine our shock when we received a phone call from GOSH to say that Teddy also had MCADD. We were devastated to say the least.

After the initial visits started to tail off we felt a bit alone. As though we were now left to deal with the condition on our own. It was then that I started to trawl the Internet for information. When I tried to explain the condition to friends and family they just stared blankly. Most would reply, "But they look perfectly normal!"

It was at this time that I discovered a life-line..... FODSupport.org!

I read all the messages from the Email List and responded to a couple. It was amazing to hear other peoples' stories and a comfort to know I wasn't alone. I was also very impressed with the amount of knowledge the parents had regarding their children's condition.

I eventually met up with 2 other UK MCADD families via FODSupport.org. This was very special as we were all going through similar difficulties. Here in the UK they treat MCADD quite conservatively, e.g. don't prescribe Carnitine, Creatine etc. When I queried this with GOSH, they dismissed it as unnecessary, which was frustrating as I understandably wanted the best treatment for my children.

By the end of 2005, we were finding out lots of information about newborn screening. Kitty had been so lucky to have been included in a pilot study here in Essex. Our health authority was only one of 6 in the UK that was screening for MCADD. The pilot had started in October 2004, so our son had missed out as he was born in July 2003. The pilot study was continuing until 2008. Then a decision would be made whether to screen for MCADD on a national basis. I regularly completed questionnaires regarding the pilot study and joined the UK charity called "CLIMB" - Children Living With Inherited Metabolic Disorders. We were determined that national screening should start as soon as possible. *[CLIMB has an Oct 20, 2007 conference on FODs, <http://www.climb.org.uk/>]*

In March 2006, Kitty had her first hospital admission. The local hospital where she was admitted did not know how to treat her, even though we'd been there previously on visits to see one of the paediatric consultants. Unfortunately he was on holiday at the time of admission and no one else seemed to know about MCADD. I had various meetings with doctors, dietitians and nurses and between us we worked out the best treatment. However, the day we went home, she became very ill - she had caught RotaVirus whilst in the hospital! So we went back for another week's stay. This was very frightening as she was slipping into unconsciousness. Eventually Kitty recovered. However, the experience had left me slightly traumatized and angry. I met with the ward matron and explained my worries concerning the ward's lack of knowledge, the lack of infection control etc. etc. She was so impressed with my feedback that she told me I'd make a good nurse! She also arranged with GOSH for the metabolic specialist to come and give a presentation on MCADD.

During the rest of the year, both children were healthy, until we reached Christmas when Kitty had her second admission. This time I was telling the doctors what treatment she needed as I was now more knowledgeable. In early 2007 Kitty and Teddy were both admitted with a stomach bug. Both only stayed a couple of days and recovered quickly with IV glucose.

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## Theodore and Katarina...*cont'd*

At this time we became concerned that Kitty may have some learning developmental delays as her speech was not forming yet. We still have to wait and see if this improves.

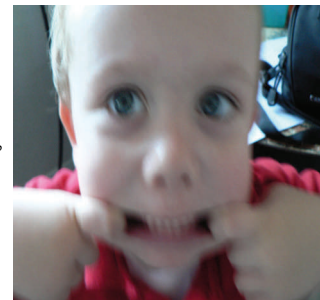
In February 2007 I was delighted to read a press release by UK Health Minister that the interim results of the UK pilot study had been so overwhelming that national screening would take place. It will be gradually rolled out and in full implementation by March 2009.

We've all come a long way since August 2005. Both children are happy and doing well. Teddy is particularly bright and I think our comparisons to him may have us worrying unnecessarily regarding Kitty's development - time will tell.

On a positive note, due to the last 2 years' experiences, I decided to become a paediatric nurse. I'm just about to finish my year at college doing combined sciences, and will be starting University in September to do a three year degree. I hope one day to specialise in metabolics. After all, with the new screening programme, I will be sure to meet many more FOD children.

I also wanted to add that I feel very privileged to be the mummy of two very special children. They have had to cope with blood tests, painful IVs, hospital visits etc. at such a young age and have shown great courage. It makes me proud to be part of such a special family.

Best wishes to all.



Susan Wood, Essex UK

[the\\_willow@btopenworld.com](mailto:the_willow@btopenworld.com)

mum to Theodore, 3yrs (MCAD) and Katerina, 15months (MCAD)



## A Family Affair ~ Shawna (MCAD), Zachary (Mito), and mom, Melissa (GA2/MADD)

My name is Melissa Cummings and I have four children. Their ages range from 15 years old to 1½ years old. On April 24<sup>th</sup> in 2003 my daughter Shawna was born. I had a wonderfully normal pregnancy. On the day she was born I had a non-stress test which showed contractions 3 minutes apart and that her heartbeat was dipping after each contraction. She came into the world perfectly healthy, or so we thought. We had chosen to bottle feed so due to this she spent the first night in the nursery with the nurses. The next morning they brought her in and told me she had turned blue when she was eating, but they said it was because she was so hungry. Now we know why this happened.

We left the hospital with no delays or extra instructions, as we were "seasoned parents." On the Friday when she was only 2 weeks old we received a phone call from our pediatrician. My only thought was "how many pediatricians call a parent just to see how things are going?" The doctor said one of Shawna's tests came back abnormal in the newborn screening. I look back and realize I don't remember being informed about the screening that was done. The doctor could only tell us one test came back abnormal and a specialist from Albany Children's Hospital would be calling us. I sat in shock and fear of what this all meant and just what was wrong with my daughter.

Within an hour we received a call from an angel. That angel's name is Cheryl Clow; I now know that she must have been specially chosen to work with my family and families like mine. She explained the possibility of our daughter having MCAD (Medium Chain Acyl Co-A Dehydrogenase deficiency) and asked that we go to Albany, which is four hours away, that Monday. Cheryl also stressed to us the importance of feeding our daughter every three hours whether Shawna wakes on her own or not.

We drove the four hours to Albany. Cheryl explained in detail about MCAD, the importance of not fasting and what would occur if Shawna did fast. Cheryl explained that 80-90% of people have the MCAD gene on a specific gene site. However, Shawna did not have this "common" mutation. Cheryl also stated the new testing was giving some false positives so more testing was done. We were so sure our little angel would be one of those false positives so we did not do any research on MCAD.

During the "waiting" period we prayed and were optimistic that nothing bad would come of this. Unfortunately we were wrong. Our daughter has MCAD. We had to take extra precautions about her being exposed to illnesses. I am a teacher and my husband is a New York State Trooper so we knew we would be exposing her to many illnesses. I took the rest of the school year

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## Family Affair...cont'd

off with my administrator's blessing and wishes of luck. Fortunately I would not have to go without my insurance - only my pay. The wonderful group of people that I work with approached and asked the teachers to help our family due to all of the expenses we were incurring. Since then some of these people have been a support group I can turn to for help, a shoulder or an ear to lean on and listen to.

Shawna was 8 months old when she had her first episode. She was hospitalized for two days on IV dextrose and on monitors. I still can see her little body lying there hooked up to machines and IVs. She was in the hospital again for 3½ days when she was 3½ years old for a stomach virus. Since then she has been relatively healthy. Most people see a healthy, happy, "normal" little girl with big blue eyes and beautiful blonde hair. It is hard when medical staff look at her and say "she looks fine" when I know that inside her body is screaming for help.

After Shawna was diagnosed we had the other children tested and all came back well. Two years later our specialist ordered test for me and my husband. My husband's tests came back normal however I was diagnosed as having a rare metabolic disorder called GA2/MADD (Glutaric Acidemia type 2, a different FOD). I remember asking Cheryl, "Is that even possible?" Obviously that answer is yes. I had a normal and healthy childhood. I do remember at a young age that if I ate fatty food I would feel very sick to my stomach. My mom was always by my side when I was ill and always made sure to keep me hydrated. Little did she know it was keeping me alive. In my early twenties I began having bouts of hypoglycemia and dizziness. I was able to control it by eating many small meals throughout the day. In my late twenties I had my gallbladder removed yet I got through that very well. I think it was due to the IV I was hooked up to the entire stay. I had four pregnancies and with only two did I have gestational diabetes.

On September 22, 2005 we had our last blessing, a son we named Zachary Richard. I had a very rough pregnancy consisting of placental previa, anemia, gestational diabetes and urinary tract infections. The last three months I was taking Insulin and Iron twice a day, and an antibiotic 3 times a day. We feared that Zach would have MCAD or MADD. We thought we were blessed when the tests came back negative. At 3 months old we noticed that he could not turn his head in a particular direction. He was diagnosed with Torticollis and underwent physical therapy. All seemed to be going well until he was 6 months old and we noticed a problem with his left arm. He would drag it behind him in his saucer and he still could not sit up even with help. He started physical therapy again this time under the diagnosis of Erb's Palsy.

Everything went well from there until he was 14 months old. It is a night that will be forever etched in my mind. He was playing with blocks on the floor and began crying in pain. When I sat beside him to see what was wrong his body stiffened and he fell backwards. I grabbed him up and his body went limp and he stopped breathing. He had had a seizure. I rushed him to the ER and they could not find anything wrong but kept him for observations. Our pediatrician at the time said that we were overreacting and that we should just let him be because there was nothing wrong with him. We found a new pediatrician who takes our children's medical issues seriously. I contacted Cheryl Clow because I knew there was something wrong and was hoping that maybe it would be a simple metabolic issue. She ordered a series of blood tests. We also followed the ER doctor's instructions of meeting with a neurologist. The neurologist had an EEG performed which found nothing wrong. Between the time of the first seizure and March, Zach had approximately 5 seizures that we are aware of.

On March 14<sup>th</sup> Cheryl called to let us know that his blood tests indicated a Mitochondrial disorder. They repeated his tests and again it strongly indicated a Mitochondrial disorder. On April 10<sup>th</sup> Zachary underwent a skin and muscle biopsy. We are now waiting for the test results to give us a detailed diagnosis. Zach has started on Carnitor and Co-Enzyme Q10 along with Ross Carbohydrate Free Formula and a special diet.

We know we are blessed to have all four children. Newborn screening in our house is a blessing that is worth any expense. I know that without it we could have lost our daughter. MCAD opened up our eyes to metabolic and in turn mitochondrial disorders which saved our son. There are times when looking at the bright side of the dark cloud is difficult, that is when my husband pushes the darkness away to help me to see the bright side. I used to question why three of us have three different disorders. The only answer I have been able to determine is that God knew we could handle it and blessed us with the knowledge of how precious life truly is and what truly matters in life. I have discovered a strength I never knew existed. Our lives have taught us to cherish every day, every step, every word and especially every "I Love You." My mom has always been there for me and she has taught me acceptance, patience, prayer, and given me my inner strength. She drops whatever she is doing whenever my family needs her. She has driven 30 minutes at 3am to watch the children when I have had to take one to the hospital. We feel blessed to have our friends and family here to help us. We are blessed to have Cheryl Clow working with our family. She is so dedicated and always helps to calm my fears. She also makes me feel like my concerns are genuine and that no matter what, I know my child the best, this is not something that all medical staff have done. I have tried to turn all of this into something positive. I take the time to explain the disorders to anyone and everyone because we are all in this together. The more people who know the less likely people with these disorders will be misdiagnosed or not understood.



Melissa Cummings  
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## Family Stories

# Pipsa & Peppi's Story ~ LCHAD (Finland)

Our so dear daughters Pipsa and Peppi were born too early. My double pregnancy had gone well. Exactly on the 30th week of pregnancy, I left to the childbearing clinic because of a small blood turn. Less than six hours after I left home two big brothers had both two little sisters. Pipsa weighed at birth 880g, Peppi at 1183g. I always remember our girls' tiny-winy tips of the noses which peeped from huge swaddling cloths. I saw my girls only briefly before they were rushed into intensive care unit.

Our dear daughters were really small; they were certainly not any models of "baby magazines." The girls grew a little while in the children's clinic of Helsinki and the growth still continued in the hospital of Espoo (Jorvi) for about ten weeks.

After anticipating for two months, during the mother's day (here in Finland always on the second Sunday of May) we got our loved ones home. We lived for the summer of our life ~ we enjoyed every moment despite the small size of our girls! The girls were still so tiny after the growing periods at the hospitals that one unknown lady inquired us at the mall "which department the sweet radio-controlled dolls can be bought" ~ our wonderful girls.

After the summer of our life autumn came and with autumn the big black sorrow. Our dear Peppimme got a common cold. "Don't worry" was the message from our doctor even though I had phoned him before our departure. Our girl had meant to die in my arms before we left....

We left the hospital about at 18 (6pm). Peppi got the medication doctor's described at ten a clock at the evening and again at night about 4 am. At the time of the following medication Peppi was not in this world any more...our dear child was dead. I collapsed! Immediately after the death of our Peppimme, Pipsa was taken to department care. There was no indication of illness on Pipsa's behalf.

### To PEPPI:

When writing this 5years 8months and 8days have past from your death. Not even one day passes so that I don't think about you. After your death we got many words of comfort from our close ones and our friends. Of course, they consoled and gave strength to go on for those of us that were left behind. Anyway, the phrase "Hardships makes you stronger" I hate most of all...if your death makes me grow somehow bigger man I'd rather be small.

Every single night I send my wishes of "good night" for you where ever you are. At night I dream, that you run around in a green summery meadow with your light curly hair fluttering and you smile and wave to me in my sleep. To your double sister to Pipsa you appear more often...I know that everything is all right with you. You know, my sweet girl that you always live in our heart. So much you were loved and are loved. With your own death you saved your sister's life. Take care of yourself in the invisible world. We will meet again.

### To PIPSA:

When I am watching you now, it is hard to believe that physiotherapist taught you how to move; you are an excellent football player and gymnast. If this were not the case you would not play soccer with your older boys and wouldn't be asked to be part in the pre-competitive gymnastic group.

When you were taken to hospital care during your premature infant checkup following Peppi's death, we thought (with your father) that this was overreacting. We were wrong! They found out that your head was slightly over sized. The skillful cardiologist also stated an oversized liver. The same was diagnosed and stated in Peppi's post mortem report -- child's fatty liver.

I remember the morning after this finding when I was returning to the children clinic of Helsinki. At night I had tried desperately to find information from the Internet. I managed even to connect the illness to LCHAD as the result of my detective job. Your father had a shift - he only was able to take me to the children clinic. I ran inside, took you into my lap, showed you pictures of Finland's winter landscapes and asked that you would not leave us. I also replied to a feedback form of the children clinic part where they asked best and worst experience during the sickness of your child's "diagnosis." There have been times that your death was imminent and had been almost as close as your twin sisters. You survived. You my darling, Pipsa "pepper" are a survivor. In spite of your basic illness you are able to lead a normal life. An excess amount of fat (in one's diet) is not good to anybody. We love you so much. You will begin

pre-school next autumn: it also will go well!

To my FAMILY and my FRIENDS:

Our beloved Peppi died Sept 30, 2001. In 2004 Pipsa made it so our faces were known to the public here in Finland through TV so that these hereditary illnesses would be made aware of to all. When Peppi died and Pipsa was diagnosed, our dear sons also began to have symptoms. If then somebody had dared to say to me at the beginning of my double pregnancy

(cont'd on page 9)



## *Pipsa & Peppi...cont'd*

that after a year one of my children would be dead, one would have a serious illness, and a psychologist would visit twice, I would have considered the comment as a joke. However, this happened. I thank for these years my family and for my good friends!

During this somewhat bumpy ride from time to time our strength has been low but you would stand on your head for 24 hours for your children if necessary. Without forgetting friends - gigantic thanks for my family for these time to time very rough years. Jammo, Nooa, Miio, Pipsa and our guardian angel Peppi.

And at last to TIINA & TERO...!

I do not know if this story goes automatically to the specialists of the field. I want, however, to give a big thanks especially to Tiina Tyni which has made its doctoral thesis here in Finland about the illness called LCHAD. Thanks also to Tero Kivelä and all doctors who take care of Pipsa.

You are doing valuable work with a big heart!!!

Kati Ronkonen from Espoo, Finland

[tuplat2001@yahoo.com](mailto:tuplat2001@yahoo.com)

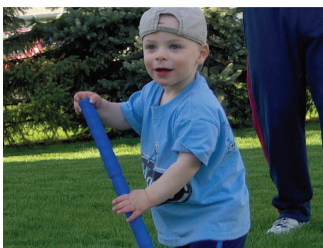
8/6/07



## Luke's Story ~ LCHAD/TFP

August 20, 2005 was a day that will forever be a complete muddle of emotions for me. How will I one day tell my child that the day of his birth was the most joyful yet horrific day of my life? His story actually begins much before his birth.

My husband, Mike, and I did everything "right." We met in college, fell in love, had good professions, got married, bought a house, and then decided to have children, in that order. In April 2003, I was diagnosed with multiple sclerosis. This event had been the worst day of my life. I was devastated, yet as time went on, I healed emotionally. Physically I took such excellent care of myself that the doctors gave me the go-ahead to start my family. Mike and I took the laid-back approach of "let's just see what happens" to getting pregnant. Well, we only had one month to "let's just see what happens" because Luke was conceived almost immediately. I had a wonderful early pregnancy...no sickness, healthy weight gain...I was a glowing mom-to-be. The first real bump in the road came when the prenatal Ultra screen was done. It came back with markers for both Down's Syndrome and Trisomy 18. Scared yet undiscouraged, we decided to handle whatever was given to us and denied any further testing.



Once the initial shock was over, and later ultrasounds proved that our baby was healthy, things went back to normal. I did everything by the book to make sure that this baby was the healthiest baby in the world! Why wouldn't he/she be? No one else in my immediate family had anything less. Plus, I already suffered from MS. God surely wouldn't do anything bad to me again!

Several days before Luke was born, I just didn't feel myself. I had a urinary tract infection, cervical bleeding (which sent me to triage, but was completely "normal," a fever, slept non-stop, and was nauseous. (Note: I ate hot wings & Mexican food through my pregnancy, so nausea was not normal for me). I also had a tremendous weight gain and swelling over my entire body. I had gained 5 pounds in just 3 days, had seen about 3 different doctors that week, and nothing was wrong with me. It was "just pregnant and uncomfortable in August."

On Friday, August 19<sup>th</sup>, I felt sicker than I ever had before. My body hurt so badly from all of the water pooling in my legs and arms and I did not even recognize my own face. I kept drinking water to flush it all out, but I couldn't urinate. I remember just sitting on the toilet trying to go while reading "What to Expect When You Are Expecting" and searching for all of the information I could find on pre-eclampsia. I had a lot of those same symptoms, but I had just been to the doctor 4 hours before. Surely, I would feel better after a goodnight's sleep.

A good night's sleep, however, was not to be had. The next morning, I felt worse, had swollen more, was nauseous, and still could not urinate. I called my mom to tell her that I would not be able to come over to her house, as planned. Using her motherly instincts, she told me to call my doctor immediately! I did, and was told to come to triage...nothing new for me. I was really terrified of the idea of being put on bed rest for the remainder of my pregnancy (6 weeks). I had no idea about the horrors that were about to ensue over the next several days.

When I saw my own doctor come into triage, I knew something was wrong, as usually they just treat you using their own resident. Dr. Sallash came into my room and explained that I had severe pre-eclampsia and needed to be delivered within 24 hours.

(cont'd on page 10)

## *Luke...cont'd*

Not really knowing what that was, I protested and told him he couldn't take my baby. He then explained everything that could happen to both of us if I didn't have that baby. I agreed and asked if I could have a C-section. Crazy, right?! Dr. Sallash told me that I was not able to have a C-section unless it became more dangerous, as it is a risky procedure. I had not even prepared myself for giving birth that day and then things very quickly went from bad to worse. I told them that the baby had been breech, so they might want to check that out before inducing me. Sure enough, he was feet first. The doctor then told me my "wish" had been granted and I would be given my C-section. Then the anesthesiologist came in and quickly burst my bubble. Because of my high blood pressure and my pre-existing condition of MS, he was not comfortable putting a needle in my spine for the epidural. I was to have an emergency C-section with general anesthesia! Because the baby was a preemie, they explained that to get him out as quickly as possible, they would need to do a long incision, from belly button to pubic bone. I had not even gotten over the idea of having a baby within 24 hours, and now they were telling my husband to make all the calls to family because I would be going in 1 hour!!!!

I was so terrified! I was sobbing that I was not going to hear my baby's first cry or the announcement of his sex (we still didn't know through all of this)! I was scared that I may not wake up and ever even see my baby. Mostly, I was afraid he would be too premature to make it! I instructed Mike to not call anyone with the baby's sex until I knew what it was, said good-bye to my parents, who had rushed to the hospital to see me, and was off to the ER, shaking and sobbing! At 4:44 pm on Saturday, August 20<sup>th</sup>, my baby boy was born! I awoke to Mike feeding me ice chips in the recovery and Dr. Sallash sitting by my side. I asked what we had and Mike told me a boy, and I said, "His name will be Luke Jameson Folcher. Spell the middle name right, with an 'E'!" I got my boy! The nightmare was over! Or so I thought!

After all of the morphine and other drugs started clearing out of my system, I got used to the idea of my preemie being in the NICU. "There is no better place for him to be," I recall telling other parents and myself. I honestly believed it, too. That is, until, the Wednesday after I gave birth. I was cleared to go home, under strict order of bed rest and I thought it would be really nice that I was able to recover while my baby was being taken care of, that is, until I was being wheeled out of the hospital. I remember just starting to sob that I was leaving without my baby, on what was supposed to be the happiest day of my life. Once I started crying, there was no stopping. While Mike went to get the car, a nurse on her way into the hospital noticed how distraught I was and gave me tissues and just talked to me. I remember saying that I was leaving my heart on the third floor of the NICU.

Once home, Mike got me my favorite take-out, and yet I still sobbed. I cried until the next morning. I finally felt calm enough to fall asleep and the phone woke me up. It was the doc from our NICU, telling me that our baby tested positively for LCHAD on the NBS and would have to be further evaluated at Children's Hospital of Philadelphia (CHOP). He then hung up. I was HYSTERICAL! I called my husband, who had tried to get a day of work in at the office, who raced over to the hospital to get the full story.

The next day, at only 6 days old, Luke Jameson Folcher was ambulated to CHOP, with Mike and me following. He was officially diagnosed with LCHAD and we met with the metabolic team there. After spending a total of 19 days in the hospital, he was released to us on September 6<sup>th</sup>, 2005 to begin his life as a happy, wonderful miracle.

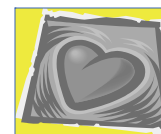
Since then, Luke has suffered several childhood infections, one severe metabolic crisis, where he was almost lost, and a g-tube placement. He has suffered no long-term disabilities, to date. Today, he is an extremely smart, happy, precocious, curious, and impish 22-month-old, who keeps Mommy and Daddy constantly on their toes!

Bethann Folcher

[folcherb@eastonsd.org](mailto:folcherb@eastonsd.org)

[Please note: When Luke was born the NBS showed red flags for LCHAD. About a year ago, Mike and I had our blood sent to Dr. Strauss, then at Vanderbilt, to be studied. He was the one who determined that I had the TFP mutation, and Michael had the regular LCHAD mutation. He behaves like and LCHADer, but the doctors at CHOP list him as LCHAD/TFP on the reports.]

## 'Fundraising' Information



Now that we have obtained full 501c3 tax-exempt status all of your personal donations, as well as those from your individual project efforts at raising FOD/Newborn Screening awareness are tax deductible! If you would like to plan a project for FOD Awareness please fill out the **Family Awareness Project Form** on our homepage to help guide you in planning for the project.

Some of the projects, in addition to the ones mentioned on page 1, have included a percentage of profits from selling products from Pampered Chef, Tupperware, iGive, and PartyLite Gifts. Keep up the GREAT work ~ all for the benefit of our Families!



### ACAD9 Deficiency: A New Inborn Error of Fatty Acid Oxidation Presenting as Liver Failure and Sudden Death

*Jerry Vockley, M.D., Ph.D.  
Children's Hospital of Pittsburgh  
University of Pittsburgh*

The acyl-CoA dehydrogenases (ACADs) are a group of enzymes that function in the mitochondria in fatty acid oxidation. There are at least nine known members of the ACAD gene family, some of which are also active in amino acid metabolism or fatty acid oxidation. Deficiencies of the ACADs are among the most common inborn errors of metabolism. Seven different ACAD genetic defects have been described in the past. The most frequent signs of fatty acid oxidation defects are low muscle tone and recurrent hypoglycemia with fasting or stress. Clinical symptoms can, however, range from neonatal death to adult onset recurrent muscle break down (rhabdomyolysis). Affected individuals can also remain asymptomatic for life. The variability is great enough to preclude a definitive diagnosis on the basis of clinical symptoms alone.

Of the three long chain specific ACADs, inherited defects have only been identified in very long chain acyl-CoA dehydrogenase (VLCAD). Patients present with recurrent episodes of hypoglycemia with or without rhabdomyolysis, sudden unexplained death, and cardiomyopathy. Recently, tandem mass spectrometry has been used to detect characteristic metabolites in blood spots collected from newborn infants allowing diagnosis of VLCAD deficiency before symptoms develop. Long chain acyl-CoA dehydrogenase (LCAD) was characterized about a decade before VLCAD was discovered, however, its function remains a mystery. Acyl-CoA dehydrogenase 9 (ACAD9) has only recently been recognized and is very similar to VLCAD in many ways. My lab has recently reported the first cases of ACAD9 deficiency presenting with episodic liver dysfunction during otherwise mild illnesses or cardiomyopathy, along with chronic neurologic problems.

The first patient was a 14-year-old male who presented with a Reye-like episode (low blood sugar and high blood ammonia) triggered by ingestion of aspirin during a minor viral illness. He progressed to coma and liver failure, and died of brain swelling in spite of intensive care. On autopsy he had evidence of a stroke in his brain. The second patient is a girl who initially presented with hypoglycemia and liver failure at 4 months of age. Evidence of a mild stroke was present on a brain scan. She has continued to have recurrent episodes of liver dysfunction with hypoglycemia, usually triggered by viral infections though these have improved with age. She is now 10 years old. Another girl, now 2 ½ years old has had similar symptoms. Hypoglycemia in both only developed when they were sick. The final patients were sisters. One died at 4½ year of age of cardiomyopathy (weakness of the heart muscle) first diagnosed at 18 months of age. Her sister died of cardiomyopathy at 22 months of age. Liver abnormalities and hypoglycemia with illness were present in both girls.

Diagnosis of ACAD9 deficiency in these children has been difficult. Abnormalities in the blood acylcarnitine profile and urine organic acids suggested a problem with fatty acid oxidation when they were ill but could not identify the specific defect. These abnormalities disappeared when the patients were well. Acylcarnitine profiles in liver tissue were abnormal in the two patients tested and may prove to be the most reliable test. DNA testing and special staining of cultured skin cells were useful in all of the cases. The development of rapid liver failure in three of our patients suggests that ACAD9 deficiency should be considered in any child with unexplained liver failure, especially when hypoglycemia or cardiomyopathy are present.

In spite of the high degree of similarity between ACAD9 and VLCAD, it is clear that their functions do not compensate for each other when either is deficient. Rather, our studies of the function of these two enzymes shows that each has a distinct role in metabolism and identifies new functions for fatty acid oxidation in brain metabolism.



### Research Abstract: Fetal Fatty Acid Oxidation Defects and Maternal Liver Disease in Pregnancy

*Marsha F. Browning, MD, MPH, Harvey L. Levy, MD, Louise E. Wilkins-Haug, MD, PhD,  
Cecilia Larson, MD, and Vivian E. Shih, MD*

*[Please note: full citations and article on FOD website – Medical Info page, FOD Research articles]*

**OBJECTIVE:** The objective was to evaluate the relationships between all types of fetal fatty acid oxidation defects and maternal liver disease, including acute fatty liver of pregnancy and hemolysis, elevated liver enzymes, and low platelets (HELLP) syndrome.

**METHODS:** This was a case-control study comparing fetal fatty acid oxidation defects to the outcome of maternal liver disease. Fifty case infants with fatty acid oxidation defects were identified, with 25 matched controls collected per case. This generated a total of 50 case infants and 1,250 control infants. Pregnancies were evaluated for the presence of maternal liver disease (comprised of acute fatty liver of pregnancy, HELLP syndrome, and preeclampsia evolving into HELLP syndrome) using a conditional logistic regression model. Subgroup analysis compared long chain to short and medium chain fatty acid defects.

(cont'd on page 12)

## Medical Update...cont'd

**RESULTS:** Maternal liver disease was noted in 16.00% of all fatty acid oxidation defect pregnancies compared with 0.88% in the general population (odds ratio 20.4, 95% confidence interval 7.82–53.2). These pregnancies demonstrated an 18.1-fold increase in maternal liver disease when compared with our matched population controls with unaffected fetuses. All classifications of fatty acid oxidation defects were at high risk of developing maternal liver disease. Long chain defects were 50 times more likely than controls to develop maternal liver disease and short and medium chain defects were 12 times more likely to develop maternal liver disease.

**CONCLUSION:** Maternal liver disease is significantly higher across the entire spectrum of fatty acid oxidation defects pregnancies compared with the matched control population. Notably, there is significant risk to the pregnancies with fetuses affected with short and medium chain defects, not just those with fetal long chain fatty acid oxidation defects as previously reported. Future studies should examine the pathophysiology of all infant fatty acid oxidation defects and its implications for maternal liver disease for improved future health outcomes. (*Obstet Gynecol 2006;107: 115–20*)

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## Pharmaceutical Update

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

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### ***FDA Approves Carnitor® SF (Levocarnitine) Sugar-Free Oral Solution For Patients With Carnitine Deficiency***

**GAITHERSBURG, MD, June 22, 2007** – The Food and Drug Administration has announced the approval of Carnitor® SF (levocarnitine) Sugar-Free Oral Solution for the same indication approved for the current Carnitor® Oral Solution containing sugar. Carnitor® SF and Carnitor® Oral Solution are indicated to treat primary systemic carnitine deficiency and for acute and chronic treatment of patients with an inborn error of metabolism which results in a secondary carnitine deficiency.

Adverse events reported with Carnitor® use include nausea, vomiting, body odor, gastritis, and seizures. There were no contraindications or warnings. Please see full prescribing information.

Primary systemic carnitine deficiency is a very rare genetic disorder that typically presents in infants. Secondary carnitine deficiency may present in infants, children, and adults. Factors contributing to carnitine deficiency may include inborn errors of metabolism, fanconi syndrome, chronic renal dialysis, carnitine-deficient diet, extreme prematurity, malabsorption, HIV infection or antiretroviral therapy, valproic acid (VPA) and a Ketogenic Diet. Symptoms of secondary carnitine deficiency may include cardiomyopathy, encephalopathy, muscle weakness, anemia, and fatigue.

Carnitor® SF, the sugar-free version of levocarnitine, is appropriate for patients with carnitine deficiency for whom a sugar-free option is desirable. This may include patients with diabetes or those who are on a Ketogenic Diet who need to limit sugar and carbohydrates.

“We are pleased to be able to offer a sugar-free version of Carnitor® for patients who are diabetic or those who are on a Ketogenic Diet, those who are intolerant or have sensitivities to sugar and develop carnitine deficiency,” *said Gregg Lapointe, Sigma-Tau Chief Operating Officer. “In keeping with Sigma-Tau’s commitment to rare diseases, we are pleased to provide this new option for this important group of patients.*

Carnitine functions in the body as a carrier of fatty acids to the energy centers in muscles (mitochondria). A deficiency of carnitine, normally produced by the liver and kidneys, can result in extreme muscle weakness and other related symptoms.

“Previously there was nothing available for patients with carnitine deficiency who needed to limit sugar intake, especially children with diabetes or those on a Ketogenic Diet,” said Dr. Darryl De Vivo, Associate Chairman (Neurology) for Pediatric Neurosciences at Columbia University Medical Center. “Carnitine deficiency is a debilitating illness, so it is reassuring to know that these patients can still treat their symptoms without complicating their condition with undue sugar intake.”

Carnitor® SF (levocarnitine) Sugar-Free Oral Solution is the only U.S. FDA approved prescription sugar-free oral solution of levocarnitine available. The product is expected to be available June 22, 2007.

(cont'd on page 13)

## Pharmaceutical Update cont'd

### About Sigma-Tau Pharmaceuticals, Inc.

Sigma-Tau Pharmaceuticals, Inc. is a U.S. based, wholly owned subsidiary of the Sigma-Tau Group, and is dedicated solely to the global development and commercialization of medicines for patients with rare diseases. Sigma-Tau Pharmaceuticals, Inc. is based in Gaithersburg, Maryland.

Since 1989, the company's products have been focused on rare diseases, kidney disease, and cancer. With more than 6,000 identified rare diseases that affect approximately 25 million patients in the United States, Sigma-Tau places its considerable scientific resources behind the discovery of compounds that benefit the few. The company has a substantial development program focused on transplant, cancer, inherited genetic disorders, malaria, as well as other areas of unmet medical need. For more information about the company, visit [www.sigmatatau.com](http://www.sigmatatau.com).

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A new L-carnitine (generic form) supplement was approved for distribution by the FDA a year or two ago. This **generic form (as well as the brand name Carnitor®) needs a Prescription from the Dr.** Please also note that Carnitor® and the generic form of L-carnitine are **NOT** the same as the over-the-counter carnitine supplements often bought at healthfood stores ~ those products are **NOT** regulated or approved by the FDA to be used for metabolic disorders (read the article on <http://www.fodsupport.org/pharmaceuticals.htm>). The term 'generic form of a drug' should **NOT** be used interchangeably with the term 'over-the-counter supplement.'



## A Special FOD Child's Story ~ Feeling Grumpy

By Austen (with help from Joe-Joe)

*To all the children who feel grumpy in the morning*

*[From mom: Austen thought that the FOD kids might want to read his story that we read (over and over) at breakfast.]*

Do you feel grumpy in the morning? If you do...remember, try to have a snack before you act mean. A snack will help you to feel a little better until you eat your breakfast. Breakfast is very important for everyone. The best breakfast for Austen is warm oatmeal. It is yummy in your tummy and it helps you to feel much better. Every morning after eating oatmeal, Austen says "Thank you, I feel much better now. I don't feel sick anymore." and Austen is on his way to school to enjoy a good day.

Austen knows what it means to feel grumpy. He also understands why he feels grumpy. Sometimes he needs help re-directing his thoughts and reading this story always seems to work.

So, all you children who feel this way...before you scream, scratch, bang your head or bite, remember, try to have a snack.

The End

Gwen, mom to Austen, age 10, GA2/MADD

## NBS Update



**FOD website:** Be sure to visit our website (*In the News* page) for the current articles on NBS efforts across the US and Canada. More states are getting on board (albeit slowly!) so check <http://genes-r-us.uthscsa.edu/> every now and then to update yourselves on what your state is adding to their NBS panel of tests. Keep up the great work!

### **FOD and Expanded Newborn Screening Awareness Projects**

*[Both of the following projects were a GREAT way to raise FOD awareness and funds for both the FOD Group and a Medical Center! Thanks to the Shannon and Archetti Families for sharing your successful projects! DLG]*

**“TOKENS FOR THOMAS”  
MCAD AWARENESS  
In Memory of Thomas Wood Shannon  
Mar 14, 2006 - Mar 16, 2006**

As I started approaching what would have been the first birthday of Thomas, a couple of ladies I worked with at the high school asked how they could help me deal with the sadness of the weeks ahead. The school secretary mentioned doing something to raise money and send it to a charity of my choice in memory of Thomas. That day, I began thinking what we could do to raise awareness while memorializing Thomas. I approached these two ladies with my “Tokens for Thomas” idea. My original idea was that we would have an awareness walk and at the end of the walk, students would have the opportunity to donate their spare change and we would send the money into the FOD Family Support Group. From that idea, the project grew and grew.

To introduce the two week event, each student, teacher, and staff member received a flyer summarizing MCAD and emphasizing the importance of newborn screening. The Save Babies Website and the FOD Family Website was listed on the flyer for further research opportunities. Each person was encouraged to read the information and pass it on to someone with children.

Each 4<sup>th</sup> period class at the high school was challenged to bring in their coins and the class raising the most money would be treated to a pizza party. The students not only brought in their spare change; they asked their parents, grandparents, neighbors, and fellow church members for donations also. When businesses in town heard of the students’ efforts, they sent in contributions as well. The Pre-K class where Tucker Shannon (Thomas’ big brother) attends also participated in the fundraising efforts and the memorial walk.

After the two week token drive was over, a memorial walk was held. The purpose of the walk was to announce the amount of money raised, present a check to the family of Thomas Shannon, and walk to raise awareness for MCAD. News media were present to cover the awareness walk and to interview Thomas’ mother, Melanie Shannon, about MCAD. The event was covered in the local newspaper and was shown on three news segments that evening. Through the flyers and the media coverage, thousands of people learned about MCAD and the importance of newborn screening that week. AND over \$4,000 was raised and sent to the FOD Family Support Group in memory of Thomas Wood Shannon (divided evenly between the FOD General, Clinical and Research Trust Funds).

Thanks to New Albany High School and the community for their generosity in contributing to the token drive. Special thanks to New Albany High School’s Anchor Club and Mu Alpha Theta Club for sponsoring the event and to organizers Sandra Pannell, Mary Margaret King, and Heather Ferrell for their efforts in planning the program and walk and counting the money.

Submitted by:

Melanie Shannon

[mshannon@newalbany.k12.ms.us](mailto:mshannon@newalbany.k12.ms.us)

New Albany, Mississippi

Mom to Thomas (MCAD 3/14/06-3/16/06)

Tucker 5 years old (unaffected)

## *NBS Update...cont'd*

### **3-YEAR-OLD MONROE BOY NAMED GRAND MARSHAL OF THE 'GO THE DISTANCE' WALK AT WESTCHESTER MEDICAL CENTER**

*Contact: Lisa Archetti*

MONROE---A 3-year-old Monroe boy with a rare metabolic disease has been selected as grand marshal of the third annual 'Go the Distance Walk' for the Maria Fareri Children's Hospital at Westchester Medical Center in Valhalla.

Marcello Archetti, a patient at the hospital's Inherited Metabolic Disease Center, will receive his sash at a kickoff breakfast on March 3 and have, among other things, the honor of leading the walk on May 6.

The walk, in its third year, has raised almost \$700,000 for the children's hospital. A team named to honor Marcello, 'Marcello's Miracle,' raised \$17,000 toward that amount.

Marcello, the son of Lisa and David Archetti, has LCHAD (Long-chain 3 hydroxyacyl CoA Dehydrogenase) deficiency. LCHAD is caused because the body is missing this enzyme that is needed to break down long chain fats. This results in the body's inability to break down fatty acids into a usable energy source. When Marcello's body doesn't have the proper calories or energy resources attained from foods, his body will then break down muscle to get that energy. Unknown to them until Marcello's eventual diagnosis, Lisa and David are carriers of the recessive gene that causes this disease.

At approximately six weeks of age, Marcello experienced his first virus. "From that point forward it seemed as though his overall health never got better," said his mother Lisa. "He had a virus after virus. Weeks later he was diagnosed as 'failure to thrive' because he was no longer putting on weight like a normal infant would. He had no appetite. I just knew something was wrong and no one thought anything of it."

Marcello soon became sick again with another virus, but this time it was different. "He was so lethargic and his temperature was 95 degrees," she said. "We took him to his pediatrician, and he made the right call to send him to Westchester because he knew his symptoms were so out of the 'norm.' "He looked as though he was going to die."

All kinds of tests were done, but the results were normal. After three days as an inpatient, Marcello was discharged with what doctors thought was a virus. "A few days later I got a call back from doctors saying that they discovered what was wrong with Marcello," Archetti said. "They did a metabolic workup and it came back that he had LCHAD. And when we brought him back, we then found out that as a result of this metabolic condition, he also had cardiomyopathy. We were totally devastated."

Marcello survived, and his mother describes him as "a happy, adorable little boy who has an amazing personality and thoroughly enjoys life regardless of the obstacles he faces on a regular basis with his disorder." He has weekly speech, occupational and physical therapy sessions and is under the daily care of a nurse.

Because of their experiences, the Archettis are committed to raising funds for the Inherited Metabolic Disease Center. "We're so grateful to the medical center and their physicians for their expertise and for their quick thinking to test Marcello for this disorder," she said. "Because of them, Marcello is alive and thriving today. If it wasn't for them, it's a very good possibility that Marcello could have eventually went into a metabolic crisis and died." Currently, the only treatment for this disorder is an intravenous drip of glucose, but the Archettis remain hopeful a cure will be found.

"We're involved in this walk for several reasons," said Archetti. "First, we want to increase awareness of LCHAD. This disease has the ability to be quite debilitating and it can be life threatening, especially when a 'crisis' happens. Children have died from this." New York State just recently began mandatory newborn screening for LCHAD. "If we had known about his disease at birth, we could have started early intervention immediately," she added.

Secondly, Archetti also wants to increase awareness about the hospital's services. "We want people to know about the outstanding pediatric care available at Westchester," she said. "We need so many pediatric specialists, and we feel we have the best care we could get anywhere."

Lastly, she is hopeful people will consider supporting the 'Marcello's Miracle' team she and her husband are forming for the third straight year. "While the walk benefits all the pediatric services at the hospital, teams can opt to earmark their funds raised to special areas. Ours is the Inherited Metabolic Disease Center. We're hopeful that as newborn screening reveals more cases, researchers will take an even greater interest in developing new treatments and hopefully find a cure."

At the age of eight months, Marcello had a feeding tube inserted into his stomach that helps him to ward off a metabolic crisis and treat him when one happens. He happily runs around with a special backpack that automatically provides his nutritional needs through the tube. Shortly, he will have a port surgically implanted so he can have easier access to IV therapy when needed. During a metabolic crisis Marcello experiences severe leg pain because his body is breaking down muscle for energy, and most times he can't walk. He has low muscle tone due to his muscular issues, doesn't have the stamina that most children his age do and wears supports for his feet. Long term, Marcello may face peripheral vision loss, a problem facing many LCHAD patients.

"Marcello has had to face many health issues and will continue to do so as he gets older," said Archetti. "He's had many so hospitalizations or visits to hospitals for tests in his short little life due to metabolic crises. But he's a real trooper and such a joy to us.

(cont'd on page 16)

## *NBS Update...cont'd*

that he's been selected to be the grand marshal. It's going to be a really special day for him and our family."

For information on how to be a part of the 'Marcello's Miracle' team, contact Archetti at 497-1103; or the Maria Fareri Children's Hospital Foundation 914-493-2575.



*Marcello Archetti flirts with Magee Hickey of WCBS-TV Channel 2 News. Marcello will lead the 'Go the Distance Walk' for the Maria Fareri Children's Hospital at Westchester Medical Center in Valhalla on May 6. Hickey was host of the kickoff breakfast on March 3. (Photo courtesy of Lisa Archetti)*

• • •



## Nutritional Update

### **Monogen<sup>®</sup>** **A low fat, high MCT formula for the** **dietary management of long chain fatty acid oxidation disorders** **now available from Nutricia North America**

Ulrike Reichert, MS  
 Nutricia North America

#### ***Fatty acid oxidation disorders recommended for newborn screening***

Fatty acid oxidation disorders (FAO) are increasingly better-known disorders within the medical community. They have also attracted more attention recently due to the expansion of newborn screening and efforts to make screening more uniform throughout the United States. Medium and long chain fatty acid oxidation disorders, trifunctional protein and carnitine uptake deficiency are all part of the recommended core panel of disorders for which every newborn in the US should be screened.

Nevertheless, even with earlier detection, these disorders pose a challenge to families and medical teams alike. Nutritional therapy based on a restricted diet can be challenging to create and manage. Nutricia North America is committed to partnering with families and healthcare professionals in finding solutions to these dietary challenges via innovative products, programs and tools. Since our very beginning, we have continuously and diligently worked with families and the medical community to improve the quality of life and care for metabolic patients. From these efforts, **Monogen** was developed, and it is now available to families in the United States and Canada.

(cont'd on page 17)

## *Nutritional Update cont'd*



### *Monogen - a new formula available for patients with long chain FAO disorders*

**Monogen** is a medical food formula for the dietary management of long chain fatty acid oxidation disorders. These disorders are rare—long chain 3-OH acyl-CoA dehydrogenase deficiency, for example, is estimated to have an incidence of 1:50,000 to 1:200,000. Individuals with these disorders cannot metabolize long chain fatty acids (LCFA), a certain group of fats commonly found in fat-containing foods. These disorders result from defects in enzymes needed to convert LCFAs to smaller fatty acids—a process necessary to produce energy in the body.

The main goals of dietary management are to reduce dietary long chain fatty acids and avoid fasting. Tolerance of dietary LCFA is individual and depends on the remaining activity of the blocked enzymes. The actual recommended amounts of restricted dietary LCFA and total fat may differ from clinic to clinic. Typically, 10% or less of the daily required calories is needed and tolerated from LCFA. Recommendation of total fat intake may be reduced to as low as 25% of total energy intake.

In order to reach the goal of reduced total fat intake with limited LCFA and to provide adequate calories, medium chain fats (MCT) are used in formulas or directly added to the diet. Medium chain fats bypass the enzymes that are blocked in long chain fatty acid oxidation disorders, and they can be used as alternative sources of fat.

However, MCTs do not contain any of the long chain essential fatty acids needed by the body. Consequently, the amount and type of long chain fatty acids in the diet must be carefully considered to provide sufficient but not excessive amounts of these important fatty acids.

### **Monogen – optimized for use in long chain fatty acid oxidation disorders**

**Monogen** is an ideal formula for use in long chain fatty acid oxidation disorders. Here are some of the unique features and benefits of the product:

**Monogen** provides 25% of calories from fat as recommended for a low fat diet

**Monogen** has a high content of MCT (90% of fat is MCT), which bypasses the blocked enzymes. In contrast to other formulas, **Monogen** offers a balanced ratio of essential fatty acids, which is important in avoiding essential fatty acid deficiencies

**Monogen** also provides a complete nutrient profile, including trace elements, to minimize the risk of nutrient deficiencies.

In order to make **Monogen** suitable for most patients, the essential fatty acid content is 1.2% of total energy. This allows for a tailored approach to the individual needs of patients. Dietary essential fatty acids can be easily increased by adding a prescribed amount of walnut or canola oil to **Monogen**. In long chain FAO disorders, monitoring of essential fatty acids and micronutrients is generally recommended.

### **Contact Nutricia North America for samples of Monogen**

Families, please have your metabolic dietitian or physician contact Nutricia North America. We can send a sample of **Monogen** conveniently to your home.

Healthcare professionals, please contact our Nutrition Services Department or your regional Nutricia representative.

**Monogen** is an excellent choice when considering a formula for a child with a long chain fatty acid oxidation disorder.

For more information, please call our Nutrition Services Department at 1-800-365-7354. Monday – Friday 8:30 am – 5:00 pm EST or visit one of our Web sites, [www.myspecialdiet.com](http://www.myspecialdiet.com) and [www.nutricia-na.com](http://www.nutricia-na.com). We are here for you.

**Nutricia North America - Improving lives through specialized nutrition solutions**

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(cont'd on page 18)

## Nutritional Update cont'd

### Research Abstract: Metabolic control during exercise with and without medium-chain triglycerides (MCT) in children with long-chain 3-hydroxy acyl-CoA dehydrogenase (LCHAD) or trifunctional protein (TFP) deficiency

Melanie B. Gillingham, Bradley Scott, Diane Elliott, Cary O. Harding

[Please note: full citations and article on FOD website – Medical Info page, FOD Research articles]

#### Abstract

Exercise induced rhabdomyolysis is a complication of long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) and mitochondrial trifunctional protein (TFP) deficiency that frequently leads to exercise avoidance. Dietary therapy for most subjects includes medium-chain triglyceride (MCT) supplementation but analysis of diet records indicates that the majority of patients consume oral MCT only with breakfast and at bedtime. We hypothesized that MCT immediately prior to exercise would provide an alternative fuel source during that bout of exercise and improve exercise tolerance in children with LCHAD deficiency. Nine subjects completed two 45 min moderate intensity (60–70% predicted maximum heart rate (HR)) treadmill exercise tests. Subjects were given 4 oz of orange juice alone or orange juice and 0.5 g MCT per kg lean body mass, 20 min prior to exercise in a randomized cross-over design. ECG and respiratory gas exchange including respiratory quotient (RQ) were monitored. Blood levels of acylcarnitines, creatine kinase, lactate, and *B*-hydroxybutyrate were measured prior to and immediately after exercise, and again following 20min rest. Creatine kinase and lactate levels did not change with exercise. There was no significant difference in RQ between the two exercise tests but there was a decrease in steady-state HR following MCT supplementation. Cumulative long-chain 3-hydroxyacylcarnitines were 30% lower and *B*-hydroxybutyrate was three-fold higher after the MCT-pretreated exercise test compared to the test with orange juice alone. Coordinating MCT supplementation with periods of increased activity may improve the metabolic control of children with LCHAD and TFP deficiency following exercise.

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**Keywords:** Long-chain 3-hydroxyacyl-coA dehydrogenase; Long-chain 3-hydroxyacyl-coA dehydrogenase deficiency; Trifunctional protein; Trifunctional protein deficiency; Fatty-acid oxidation; Exercise; Rhabdomyolysis; Medium-chain triglyceride; *B*-Oxidation



## Recipes



### Tomato Sauce

*I learned to make a "full fat" and full flavor version of this from a wonderful little, old Italian lady from Central Italy. I simply tweaked it to make it fat free. My Irish husband, who used to only eat jarred sauce, now turns his nose up at it in favor of this. Hope you enjoy as much as we do.*

- 1 onion, peeled
- 10 or so whole cloves garlic (to taste, again- I love them!)
- 4 cloves garlic, minced
- 1 pint (or so) dry red wine (whatever you like to drink)
- 1 bay leaf
- 2 28-oz. cans tomato sauce
- 2 28-oz. cans crushed tomatoes (or pureed if you like it smoother)
- 2 6-oz. cans tomato paste
- ½ cup of sugar or so (I like things a bit tart)
- Pam cooking spray

Spray bottom and sides of big pot thoroughly with cooking spray. Make a pincushion with the cloves and peeled onion. (It's actually quite pretty). Sauté the cloved onion on medium heat until it starts to sweat a little. Be careful not to burn it... Pam has a really low smoking point compared to olive oil. Add garlic, wine, and bayleaf and simmer until it smells like you've died and gone to heaven. Add all tomato products and let simmer on very low, stirring often. Add sugar and allow to simmer even further, until you are happy with the taste. This is great alone or with meat added. This is completely fat free, but if you want a full fat version, use oil, not Pam. Also use marrow bones, beef roast, pork roast, and maybe a little veal before you add the tomato products. Magnifico for our non-LCHAD friends.

Bethann Folcher  
Northampton, PA  
[folcherb@eastonsd.org](mailto:folcherb@eastonsd.org)

(cont'd on page 19)

## Recipes cont'd

### Slow-cooker Chicken Stew

*This dish was originally made with beef, but we just simply added chicken to make it Lukey-approved, the gravy is brownish and hearty, but still pretty good. I didn't make up this recipe, so you'll have to check your soups & chicken for individual fat content.*

1 lb. chicken breast cutlets  
 5 red or white potatoes  
 1 lb. mushrooms (your choice)  
 ½ to 1 lb. carrots  
 1 can 98% fat free cream of chicken soup (cream of celery works well too)  
 1 can 98% fat free cream of mushroom soup  
 1 6-oz. can tomato sauce  
 1 package of dried onion soup  
 Salt (to taste)  
 Pepper (to taste)

Mix together all of these soups & tomato sauce in a medium bowl and set aside. Throw all other ingredients in crock pot. Pour soup mixture over everything and mix until well-incorporated. Cover and cook on low for several hours, until chicken is done and falls apart.

Serves 4-6 people, depending on the size of servings.  
 Bethann Folcher (mom to Luke, LCHAD/TFP)  
 Northampton, PA  
[folcherb@eastonsd.org](mailto:folcherb@eastonsd.org)

### No-fat Cheesecake

*(also no-gluten, if no-gluten brands are chosen for ingredients.) I adapted this from a Kraft cheesecake recipe. I use a spring-form pan and a food processor, but you can use a mixer)*

#### *Bottom Crust:*

2 cups of corn flakes  
 3 tbs sugar

Grind up corn flakes and sugar in a food processor (or crush in a bag) and spread evenly over the bottom of the pan. Preheat the oven to 325F. Bake the crust in the pan for ~5 minutes.

#### *Filling:*

4, 8oz. packages of fat-free cream cheese (I like Kraft Philadelphia fat-free best)  
 1 cup sugar  
 1 tsp vanilla  
 1 cup fat-free sour cream  
 6 egg whites

Put ingredients in food processor (or use a mixer) and blend until smooth. Pour filling slowly over corn-flake crumbs in the pan starting from the center and spiraling outward, (trying not to displace the cornflake crumbs too much). Smooth the filling with spatula. Bake at 325F for 45 minutes or until the center is almost set. (My oven always seems to take longer) Let cool.

#### *Berry topping:*

1 can of berries or cherries in heavy syrup  
 Cornstarch for thickening

Heat berries & syrup in a small pan until boiling. Reduce the heat. Stir a tablespoon of cornstarch into a small amount of cold water until smooth. Stir into hot berry mixture and stir until it thickens. You can add more cornstarch/cold water to thicken it more, or some extra sugar to make it sweeter. When the cheesecake is cool to the touch, spread the berry topping on top. Refrigerate for ~2 hours.

#### *Other flavoring options:*

Add a teaspoon of lemon or orange zest to the filling  
 Add ¼ cup of cocoa to the filling

Makes 12 servings

Nutrients per serving: Protein 14g, Carbohydrates 36g, Fats 0g, Calcium 44% RDA, Vitamin A 29% RDA

Christyne Bliton



## Welcome to New Babies!

Kristen and Ken Mitchell are proud to announce that **Logan Andrew Mitchell** (LCHAD), brother to Nolan (LCHAD, birth 8/8/04, death 5/16/05), was born Mar 31, 2007 at 10:10 am weighing 3lbs exactly. "We are ecstatically in love already and are the happiest that we have been in 2 years!!"

Welcome to **Madison Young** (LCHAD) born to Jessie and Bryan Young on April 23, 2007. Madison was 15 weeks early and weighed in at 1 lb 15 oz and 13 $\frac{3}{8}$ ". You can follow Madison's journey at [www.youngshomeimprovement.com](http://www.youngshomeimprovement.com).

Karen and Ian Underwood and big sister, Laura (GA2/MADD) of Auckland, New Zealand are proud to announce the birth of **Hannah** (GA2/MADD) on May 31, 2007. Hannah weighed 7 lbs 4 oz and was 19 $\frac{1}{2}$ " long. Karen wanted to thank the Group for all the thoughts and prayers ~ they appreciated them so much.

Eileen and Matthew Shank along with Big Brothers Graham and Spencer (MCAD) announce the birth of **Elizabeth Theresa Shank** on June 7, 2007. Elizabeth is unaffected and weighed in at 9 lbs 8 oz.



- Dr. Jerome Groopman, a staff writer at *The New Yorker*, has written a book about how doctors make decisions regarding their patients. It's called *How Doctors Think*." Groopman is chief of experimental medicine at Beth Israel Deaconess Medical Center in Boston and teaches at Harvard Medical School.

- I found some excellent videos for kids with autism from Model Kids. Very soothing and calming. *The Model Me Kids*® video series demonstrates social skills by modeling peer behavior at school, on a play date, at a birthday party, on the playground, at a library, restaurant, and more. Children narrate and explain each skill.

Designed as a teaching tool for children, adolescents, and teenagers with Autism, Aspergers Syndrome, PDD-NOS, and Nonverbal Learning Disorder (NVLD or [NLD](http://www.modelmekids.com/index.html)). <http://www.modelmekids.com/index.html>

Gwen

[gwenabele@hotmail.com](mailto:gwenabele@hotmail.com)

mom to Austen, age 10, MADD, aspergers



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

**Please refer to our Jan 2007 issue for the complete Love Messages section ~**

*'Without a sense of caring, there can be no sense of community.'*

~ Anthony D'Angelo



**Deb's New Address and Grief Consultation Services**

**Deb Lee Gould, MEd  
Director, FOD Family Support Group  
2041 Tomahawk  
Okemos, MI 48864**

**Home Office Phone: (517) 381-1940  
US/Canada Fax: (866) 290-5206  
Overseas Fax: (313) 432-5928  
[deb@fodsupport.org](mailto:deb@fodsupport.org)**



I am offering (**at no charge**) additional grief consultation services to our Families, as well as to the public, that have experienced the death of a child or other loved ones and are having a difficult time living with this reality. These services will be offered via our website. In place of a fee, a donation to our FOD General Fund will be requested, but not required. All emails or phone contact will be confidential.

Specific Grief Intake forms are on our homepage. For information on my educational background and grief training experience, I have posted some links on our homepage ([Grief Support and Consultation Services for the Death of a Child or Loved one](#)), and on the Coping and Healing page.



# Kids Korner



Anna Hardy  
14 months  
MCAD



Jordis Serafin  
16 months  
VLCAD



Maren Harwood  
20 months  
MCAD



Kadin Johnson  
MCAD

Molly Caplette  
MCAD





# Family & Professional

**Family Donations:** Mr & Mrs Larry Wood and Mr & Mrs Joey Wood in memory of Thomas Wood Shannon (undiag MCAD). Mary Thorson, in memory of her daughter, Wendy (TFP). Lisa and Bob Curtis in memory of Kayla Goodman's Grandpa. Arleen Phang, Leanne Pence, Angela Riley, Debbie of Lynne's Charms, Dori Bischof, Erica Serafin, Julie hagen, Ryan Harrell, Jennifer Cox, Kristin McFetters and Terilyn Scholze - T-shirts and/or Bracelets. The Widmann Family in honor of McKenna Widmann (MCAD), as well as an anonymous donor.

Thank you to all that have bought products from companies on the Internet that support the iGive and Cafepress.com program of donating a certain percentage to Groups like ours. All of those links are on our homepage, right sidebar boxes. **All donations are tax-deductible ~ we have 501c3 tax-exempt status.**

**Professional Donations:** Sigma-Tau Pharmaceuticals, Inc. (makers of Carnitor®); New Albany High School and John Stroud Agency, Inc of New Albany, MS, in memory of Thomas Wood Shannon. JM Family Enterprises, Inc., (Dave Bastian - FOD Family, and Tara Desmet) in honor of their 'Dollars for Doers' volunteer hours. Brenda Goodman (Usborne Online books) in honor of her daughter, Kayla (SCAD and mito).

We greatly appreciate donations to help with postage and copying costs, website fees, conference costs, phone calls, and raising funds for FOD Clinical Training and FOD Research. **US Checks can be made payable to 'FOD FAMILY SUPPORT GROUP' and mailed to Deb.** We also have a PayPal link on our homepage. **ALL donations are tax-deductible. Our Tax ID # is 83-0471342.**



Thank you to Erika Wallace - erikawallacepa@yahoo.com (Mailing Lists), Mary Lingle - Mcartwrite@aol.com (Web Page) and Brian Gould - gouldbr1@msu.edu (newsletter) for all your hard work. Special thanks to Sigma-Tau Pharmaceuticals, Inc. for their continued financial support.

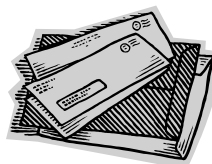
*The views expressed in the FOD Communication Network Newsletter do not necessarily represent the views of our Advisors or all of our members. Before trying anything new with your child or yourself in regard to treatment, please discuss matters with your doctor or specialist.*



## Reminders

**Families** - Please send **TYPED (preferably in word document)** stories by **Dec 10, 2007**. To be listed on the printed FAMILY LIST (refer to our website, Online Forms), please return the SIGNED Family Questionnaire to Deb. 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 10, 2007**. Also, please return to Deb the **Professional Questionnaire** even if you are already listed on the printed Professional List. Refer to our website, Online Forms.



## Communicate With Us

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

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

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Please include ideas for future issues or your questions

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**'Our souls are not hungry for fame, comfort, wealth or power. Those rewards create almost as many problems as they solve. Our souls are hungry for meaning, for the sense that we have figured out how to live so that our lives matter, so that the world will be at least a little bit different for our having passed through it.'**

~ Rabbi Harold Kushner