We hope everyone is staying cool this summer! One thing that is staying ‘hot’ is our Awareness Bracelets ~ the response has been tremendous! It’s a great way to discuss Expanded Newborn Screening and explain what FODs are and how they impact our Families. If you’d like to either be a Seller or a Buyer, please visit our site and click the right sidebar where it says ‘Bracelets are here.’ Please read the entire page before you order ~ you can pay using a credit card via the FOD PayPal link or you can write a check out to FOD (not tax-deductible) or to OAA (tax-deductible ~ we are doing this project with the Organic Acidemia Association www.oaanews.org).

Our next HOT topic ~ planning for our joint National Metabolic Conference which will be held on June 23-24, 2006 in Dallas, Texas. We are once again joining forces with the Organic Acidemia Association to provide a variety of speakers and discussions for our Families. Families and Professionals from around the world are welcome! We are pleased to announce that The Institute of Metabolic Disease, Baylor Healthcare System will be our host, Dr Charles Roe is Medical Director of the Institute, as well as Medical Advisor for our Group (since 1991). As we get closer to the Conference, we will post Hotel information and Registration Forms on our website. There will also be a special mailing in February or March with not only the Conference Registration Form, but also updated Family and Professional Lists. It’s been a few years since we updated those lists. The next several newsletters will ALL be online only due to using much of our funding for the Conference.

We have had several suggestions for topics (effective communication with professionals; neurological, GI, sleep, adult presentation, feeding, and sibling issues; new treatments; and an update on NBS, just to name a few) and we will post all of our Speakers once we confirm the agenda. Plan on coming in on the 22nd, because we hope to have a casual get-together at the Hotel before we start our busy 2 days of meetings. Because of various reasons (ie. cost, liability), there will be NO Childcare available. Our sessions will be open to mature pre-teens, teens and adults.

A great way to stay updated on Conference news as well as for networking with other Families, is to sign up for our Email List on our site. Once you are signed up, be sure to save the email that explains how to send messages, read the archives, how to change your password and set your options. It is important for you to check your spam filters because if they are set too high you will not receive list type messages or they will immediately go to your junk mail/spam folder and you may get ‘bounced’ from the List.

Please note that at times (usually 5-7 days a month) I am away from Greensboro ~ Dan works at Michigan State, so we are commuting until next August when Brian graduates from high school. If I don’t get back with you right away, that’s why ~ and I will email/call when I return to NC. In any case, if it’s very important you can contact us in MI at 517-381-1940.

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

We also have a variety of Professional Medical and Nutritional information in this issue ~ Thank you to all our contributors. Some of those articles will be posted on our website ~ we always welcome new Medical articles (in pdf form).

Whether you’re a Family or a Professional, we are all striving to create awareness, education, screening and diagnosis, clinical treatment, and research ~ by sharing your story or your expertise...

‘We Are All in This Together!’

Take care… DLG
Letter from the Editor

I have been meaning to write these comments for several issues now ~ but now after yet another Family’s tragic situation I am compelled to do it now! In the field of Metabolic and Mitochondrial Disorders, Families are not immune to having to withstand FALSE ACCUSATIONS of hurting their own child (aka Munchausen’s syndrome by proxy) ~ especially when Drs have not been able to determine a diagnosis ~ they sometimes give the MOM the ‘diagnosis’ of child abuser due to an ‘insatiable need for attention from medical practitioners!’

It’s quite ironic that on one hand we have some professionals/states/companies trying to DELAY the expanded nbs process (thus more babies die and go undiagnosed and experience possible CHRONIC medical complications AND Drs’ visits) and on another hand we have some professionals BLAMING PARENTS for MAKING THEIR OWN CHILDREN SICK and constantly taking them in for medical care. Children that should have been DIAGNOSED AT BIRTH instead of after YEARS of going from Dr to Dr with MANY not knowing what these disorders are, let alone what diagnostic tests to order (Refer to http://www.fodsupport.org/dx_fod.htm) ! These kids ARE SICK ~ because they AREN’T DIAGNOSED EARLY BEFORE CRISIES OCCUR ~ so why wouldn’t the parents take them into the Drs? RESPONSIBLE PARENTS DO!!

And THEN all because these Families have a child’s medical record 4+ inches thick with past Drs’ visits and lab tests etc, they are ACCUSED (many times by someone that hasn’t even SEEN the child clinically and based on some ridiculous ‘perpetrator profile’ of perceived psychological and behavioral characteristics!) of deliberately making them sick to get the attention. Some professionals have made the jump from so-called observations to psychiatric diagnosis without even having a ‘well-defined criteria for determining what IS and IS NOT a case of MSBP’ as well as that ‘profile’ not being ‘subjected to controlled empirical studies.’

There probably ARE real cases of this syndrome ~ and there ARE WAYS to TRULY investigate the CRIMINAL ACTS (ie., cameras in the hospital room, etc) if this is going on. Yet, ALL of the Families I have spoken with are THRUST into the ‘YOU ARE GUILTY’ realm before any TRUE investigation may occur! And before you know it family services is in there threatening to take the child ALL BASED on the constant Dr visits and the child chronically being sick!

Metabolic and mitochondrial disorders are relatively ‘young’ disorders and they ARE COMPLICATED ~ not only in the diagnosis but in some cases the treatment. It has taken some of our Families YEARS to FINALLY get a correct diagnosis and I would take a guess that many have medical professionals’ ‘suspicions’ written up in their child’s medical files ~ some of our Families have SEEN them! And fighting to clear those records is ANOTHER NIGHTMARE for Families!

There was an August 9, 2004 EXCELLENT article in The New Yorker, by Margaret Talbot, that addressed this very issue (the above quotes are taken from that article) ~ how FALSE ALLEGATIONS can SHAMEFULLY destroy a Family and also place the children in even more danger by delaying what they REALLY NEED ~ a CORRECT DIAGNOSIS and TREATMENT!

Without going into a lot of detail, the article describes several Families that sought medical care for their children yet some professionals felt they were seeking TOO MUCH care, ‘almost addictively’ (despite some of the children’s Drs telling them to see these other Drs for tests etc!). It also discusses results from recent systematic studies, that the ‘profile’ being used IS FLAWED yet still being used ~ and SHAMEFULLY still carries weight in the judicial and more recently in the educational system!

I strongly suggest you read this article ~ I have it in word document if anyone would like to read it. It will make you BOIL as it did me!

Our Families have enough to be concerned about ~ they don’t need some person ‘diagnosing’ how they are trying to help (although to them, it’s HURT) their child/children. What they need, as I stated at the beginning of this piece, is a CORRECT DIAGNOSIS and TREATMENT ~ not medical and legal authorities and family services wrongfully breathing down their necks!!!

Deb Lee Gould, Director
deb@fodsupport.org
In 1997 my husband and I were stationed on Marine Corps Base, Camp Lejeune, NC. Our last year in the Marines, we decided to have a baby. That same year we had our first son. We brought him home and three days later he was very cranky. He didn't sleep all day. Finally he fell asleep and it was time to feed him. I told my husband maybe we should let him sleep for at least a half an hour. For some reason my husband kept saying we have to feed him. So I went to pick him up and he was so limp. We thought he was just very tired. Eric put the bottle in Anthony’s mouth and he began to eat. A couple of days later we thought his belly button looked infected, so we took him to the Naval Hospital and they told us that he looked very jaundice. They took some blood and told us he needed to be admitted and put under lights. After a few days we were able to go home, but they told us that we needed to feed him every two hours so he would go to the bathroom a lot.

A month later I received a phone call from a doctor at Chapel Hill, and he started telling me that our son may have a problem and we needed to come up there, and get retested. I thought someone was playing a cruel joke on me. I never remembered being told, that my son’s blood was going to be sent to another hospital for NBS. I called my husband and he came home, and we both cried as we watched our son sleep. Eric called the doctor, and he explained everything to him. Two days later we traveled three hours and they took his blood. The results came back positive for MCAD. Anthony was the seventh one found that year. We were told that they were testing that year to see if it was worth testing for. The next couple of months we went for monitoring and we were taught so much. At that moment we realized how blessed we were when Anthony ate that bottle that night…that he was born in North Carolina, and that because of his jaundice he had to eat so often. At this point we were both getting out of the Marins and decided to move up North. We were told of a real good doctor in the area, and to make an appointment with him. When we called he had told us he didn't want to see us unless our son was sick. We didn't know what to do. I began reaching hospitals in the surrounding areas.

We made the move up North and found a hospital three hours from where we lived. We started seeing them and when our second son was born, we told them that we wanted everything done through Neogen Screening (bought by Pediatrix), like North Carolina did. We decided to take the more expensive test, even though we were told it was not needed. It ended up paying off. We found out Tyler had MCAD and G-6-PD. We tested Anthony and he also had G-6-PD. [Glucose-6-Phosphate Dehydrogenase, an enzyme found throughout the body which is necessary to change sugar to energy. Anemia can happen when certain Meds or foods are taken.] At that hospital we met many different doctors, but they would always end up leaving after so many months. Our insurance company wouldn't cover us to follow the doctors so we would stay and meet another doctor.

We had our third child. When Michael was born, we did the same thing as our second son. We gave the protocols and told them to take his levels. Michael’s glucose level was 40. We told the nurse what needed to be done. The nurse called downstairs to their Genetic Department to tell them what was going on, as we were calling the on call doctor at our hospital. The Genetic Department said a level of 40 was fine, and the on call doctor did not call us back. Eric and I knew this wasn't right, but no one would listen. Instead of arguing and wasting more time as his levels maybe dropping, we decided to feed Michael a bottle, and take his levels twenty minutes later. His levels were now 70. We took them again thirty minutes later and they were good. Mean while the nurse called the on call doctor again, and no one ever called us back. We decided to feed him and test him, never going very long. We did this through the night. Our whole stay in the hospital we did this. I could not believe how we were abandoned, by both hospitals. Eric and I had blood taken and sent it to Neogen ourselves. When we got home we called our doctor, asking why didn't they call us back? She told us she didn't know, but would find out and call us back. We never received a call back. We ended up leaving that hospital. A few days later our pediatrician called and told us Michael had both MCAD and G-6-PD.

The new hospital that we changed to was terrible from the start. It would take weeks for them to call us back and months to get a letter for my son’s school. I always told my husband, "If they can't return a simple phone call, how will they be in an emergency?” My worst fears came true. My son Michael got an ear infection. When he gets ear infections, he sometimes throws up. When that happens he take his levels and watch him very closely. Michael had thrown up a couple of times but still kept his regular amount of food down, his levels were fine, and he was acting happy. We decided to try to change ear medications because it wasn't working. That didn't work. Michael had thrown up again, but he was still acting fine, levels were good, and still was keeping most of his food down, but now he had gotten diarrhea. So we called the on call doctor and told him the
situation. The man laughed at me. He said obviously there isn’t a problem with MCAD, but he may be getting dehydrated. We
decided to take Michael to the hospital; on the way Michael ate a bottle. When we got there they made us wait. I told them that
doctors were expecting us. My sister had called me and wanted to know where we were. I had told her we are in the waiting room.
She said the doctors were calling my house looking for me. I kept telling them over and over they are calling my house. Finally
they took us back. Michael wanted to eat again, so we gladly fed him. We gave them our protocol and all our papers. The ER
doctor told us they were going to start an IV and follow the protocol. The whole time the resident on call was there. The doctor
and him decided to change the plans and give D5 instead of D10. No one told us of this change.

We were taken up to a room. Eric went downstairs to move the car and I stayed with Michael. A nurse came in and
checked his levels. She started yelling we have a low blood sugar. That is when I found out that they didn’t follow the protocol. I
started yelling why didn’t they follow the protocol? The doctor in a panic said I don’t know and ran out of the room. I paced
the floor in fear for a couple of seconds and shoved a bottle in Michael’s mouth. All I could do is rock him and pray. He was so
quiet. Finally after a few minutes they hooked him up to D10. My husband came in the room and knew something was wrong by
the look on my face. He started demanding to know who made the change that almost killed our son. We found out it was the on
Call doctor, he never even saw my son. After a few days we were discharged. We finally got to talk with our own doctor and it
was so depressing. The whole time she made excuses, and never apologized. All I can say is Thank God for the nurse that checked
his levels, I was watching my son slip away, and didn’t even know it. He was dropping a point a minute, any longer and we would
have lost him, due to their ignorance. It was a disgusting experience. We started going to another hospital an hour away, but they
didn’t have a lot of experience or the facilities to test my boys’ blood.

We ended up having to go again to the hospital to admit Michael. He couldn’t keep one thing down. This time we baby-
sat every move they made, we checked the IV bag to make sure they gave him the right stuff, we made sure they followed every-
thing on the protocol and we brought our own glucometer so we could check levels ourselves. I could honestly say our last experi-
ence was terrible, but it made us very knowledgeable.

We decided it would be best to try and go back to our first hospital to get the care from them. That was where we felt
most comfortable. I felt because of all the knowledge they had given us; that is how we had made it through all these years. I
hope by us, telling our story it helps let other families know that if you have had problems with care for your children, your not
alone. We can never give up. Our children count on us to be strong. The last few things I have to say is If it doesn’t kill you, it makes you stronger
and the other is Semper Fi (always faithful). We will always fight for our boys, and we will always be faithful to them, no matter
what happens. Thank You God for protecting our family, and Thank You Anthony, Tyler, and Michael. We are so proud of you.
Mommy and Daddy love you. You are…our three little heroes.

Eric and Theresa Leibengood
Anthony MCAD, G-6-PD
Tyler MCAD, G-6-PD
Michael MCAD, G-6-PD

Family Stories - Tylah’s Story ~ VLCAD, Australia

Our much longed for second child, Tylah, arrived in May, 2004. My husband and I
were ecstatic when we heard the words, “It’s a girl!” What a blessing ~ a beautiful baby
girl to go with our handsome 3 year-old-boy.

My labour was long and difficult, complicated by Tylah being in a posterior posi-
tion. Eventually after the use of stirrups and forceps Tylah entered the world. She had AP-
GARs of 9 and 10 respectively. I was so relieved to have such a healthy baby. I intended
to breastfeed Tylah so we had out first attempt shortly after she was born. Unfortunately she
didn’t latch on well and her ability to suck seemed weak. I was told not to worry and that
we’d try again later.

Tylah was taken to the nursery as I was paralysed from the waist down due to a
strong epidural block, and throughout the day (and then night) the nurses would bring her to
me to be fed. Each time she failed to latch on, so I would express a few drops of colostrum
and it would be fed to her through a syringe. Each time her body temperature would lower
**Tylah’s Story... cont’d**

to the point where she would feel cold to touch. Each time the nurses would take her back to the nursery to be placed under a heater. *(I will always wonder if I would have noticed Tylah’s symptoms sooner had I been able to spend that first day with her).*

The next day the anaesthetic had worn off so I hurried to the nursery to see my little angel. A nurse was standing by her cot writing on her chart. She explained that Tylah had had a ‘mucus’ vomit, had turned blue and had needed some oxygen. I was reassured that this was quite normal and it was fine to take her to my room. I was concerned but also excited about being able to bond with my new baby.

**Throughout the morning Tylah would not latch on to the breast and fed poorly. She would feel cold** when I unwrapped her to change her nappy (diaper). **She hardly cried;** when she did it was soft and feeble. **She rarely opened her eyes;** when she did they stared vacantly, not responding to me at all. **At noon I sat down and flipped through her chart.** Tylah had had three mucous vomits since being born, requiring oxygen each time.

I went to the nursery to voice my concerns. I was told not to worry — some babies are tired after the trauma of birth, some swallow a lot of mucous in the womb, some have trouble maintaining their body temperature... although not convinced, I returned to my room.

I watched Tylah sleep. She just didn’t look right. Her breathing was shallow, her skin colour was grey, and there was a complete lack of movement. I returned to the nursery. Once again, I was reassured she was fine, given a pat on the back and told to go and rest. **I walked out of the nursery only to turn around and walk back in again.** This time I was given the “here she comes again” look. With what I now think was divine intervention, my paediatrician walked in shortly after (he was only visiting the nursery because a meeting he was scheduled to attend at the hospital had been cancelled).

**Peter spent two minutes examining Tylah, and then she was gone:** then he was sitting beside me telling me “Your baby is very, very sick. I have to work on her now but I will speak to you later.” A nurse led me back to my room - it all seemed so surreal. I couldn’t stay in the room without my baby, so I walked into the NICU with nurses telling me it might be best not to watch what was happening. **But I had to watch** (I saw the distress on their faces when they couldn’t find a vein), I had to listen (I heard a nurse say Tylah’s blood glucose level was 1.9mmol/L), I had to know what was happening, even though it was obvious they (the nurses and doctors) had no idea why she was so ill. Eventually when they moved her from her I could see tubes protruding from her and leads attached (to many different monitors) to her tiny body while she lay lifeless in a humidity crib/incubator.

Peter ordered a chest x-ray, a head ultrasound and various blood and urine tests. **He suspected Tylah was suffering from an infection, possibly picked up in the birth canal such as Strep B.** I knew I had been tested for this whilst I was pregnant so I ruled it out. When the test results returned negative, he performed a lumbar puncture as he suspected a viral infection such as meningitis. Again the results showed nothing. A meeting was held with my obstetrician. Although difficult, the labour and birth (especially the use of forceps) could not have caused her poor health.

**For days Tylah lay lifeless,** with no positive signs coming until she was a week old. I know now that she was using all her strength to fight her way out of a coma. **Tylah’s Newborn Screening Test was delayed until she was 7 days old because of her poor condition.** Initial results showed a rare metabolic disorder, so the test was administered again, along with a urine metabolic screen and more blood work. **At 2 weeks of age Tylah was finally well enough to leave hospital and we had our answer** to her puzzling ill health — a fatty oxidation disorder called VLCAD (Very Long Chain Acyl Co-A Dehydrogenase Deficiency).

We were given strict instructions to feed her every 3 hours (not a problem as I had been expressing every 3 hours at home whilst she was still in hospital) and to NEVER let her fast (the cause of her initial episode). Peter was to be contacted at any sign of illness. An MRI was taken of her brain - it showed Tylah had suffered haemorrhaging in four different parts of her brain, with the largest and of most concern being at the base of the brain stem. **Later scans have shown that the bleeding has stopped, but at this stage it’s unknown if any long-term brain damage has resulted** (this is one of the reasons I’m so keen for Tylah to reach all her milestones).

At 3 weeks of age we met with a team of people at The Children’s Hospital at Westmead - most notably a clinical geneticist, a clinical nurse consultant and a metabolic dietitian. **A skin biopsy was taken and has confirmed the diagnosis.** This “team” gave us information about VLCAD and how to treat Tylah. We continue to see them regularly along with our wonderful paediatrician and Tylah’s dedicated GP who is our partner in keeping her well.

**Tylah has been hospitalised 5 times due to illness but has not suffered another crisis (touch wood!).** She is responding well to a low-fat/high carbohydrate diet, although her poor weight gain is concerning. At time of writing, Tylah is not taking any supplements but is given Polyjoule (a glucose syrup) when she is unwell. Tylah interacts well with others; she especially loves watching and playing with her big brother. **Currently, at 12 months old, she is very small for her age and has a slight delay in the area of gross motor skills.**

After searching on the internet for further information I came across the FOD Family Support Group and decided to join. **The support and knowledge I have received online have been invaluable.** Deb, Dan and family are truly inspirational and giving people. I have great respect and admiration for so many parents whom have had to deal (and are still dealing) with misdiagnosis/non-diagnosis, especially when their children have suffered devastating episodes as a result. My heart goes out to all who have lost their precious angels to these “silent killers.” I’m in awe of those who then have the strength to campaign for legislation for Newborn Screening in their states and/or countries.

After reading all the newsletters Deb sent I wanted to tell Tylah’s story to encourage parents to trust their instincts. **Have faith in your gut instincts** — Tylah could have suffered a fate so much worse if I had continued to just sit in my room. In fact she might have been one of the 25% of infants who don’t survive their first crisis. **Although expanded Newborn Screening is routine in my state, the results from it would have arrived too late in Tylah’s case.** Of course I strongly advocate NBS for all babies, regardless of circumstance.

**Twelve months on, I am thankful for so many things — for being blessed with 2 beautiful children and family and friends who helped us cope through such a terrible time.** I now appreciate many simple things that I used to take for granted (whoever thought a mother would love to hear her baby cry!).

God bless to all those affected by these disorders, directly or indirectly.

Teressa Cuthbert
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Our metabolic story started on August 12, 2001 (my father's birthday and what was to have been Emily's christening day) in Exeter in SW England, where we were on holiday in a camper van, staying near my father's home in Devon. Emily was born on September 23, 2000 and spent the first ten and a half months of her short but happy life completely healthy, never a line of fever or a meal missed, despite having an older brother at nursery school who brought home all the usual kids' illnesses. They were very happy months as we had hoped for a daughter and Ben (Emily's brother, now 7 years old) was so happy to have a little sister.

We left for England in the camper at the end of July 2001 and spent a wonderful few days introducing Emily to the rest of my family and enjoying the company of Ben and Emily's little cousins. Emily's christening was to be a joint one together with my brother's son (Henry, then 13 months old). The day before the christening Emily had her normal breakfast and we set off to visit a country park and children's play centre with my sister and family for the day. All was fine until lunch time, when Emily didn't want any food, so I left her thinking she'd catch up later, but after not even half an hour she vomited all her breakfast milk (a great surprise as she had never vomited before). As we had been in my father's garden all the day before in the sun and Emily had not had a hat (she had white blonde hair and fair skin: a typical English baby), I imagined she must have got a bit of sunstroke which had upset her stomach, so I thought it best to not force her to eat, we just offered her water and dry crackers for the rest of the day. She was very sleepy (in hindsight we can assume now that that was due to a low sugar level, but at the time had no reason to take it as an alarm signal). In the evening she was quite happy and we went to bed thinking, "Tomorrow she'll probably be back to normal."

During the night she was quite restless (we were sharing a bunk in the camper) and at one point she must have bumped her head on the wall of the camper but as she didn't cry or appear bothered by it, I let her be (the doctors later said she had probably had a kind of fit or seizure due to her body being by now well on the road to disaster).

Next morning she appeared to be sleeping normally when I got up to shower, but at the noise of the water pump starting she cried out and when I came out of the shower Mauro (my husband) was trying to feed her a bottle but she was very obviously not right. Her limbs were limp, her eyes didn't focus and she appeared to be drugged and unable to respond. Fortunately my sister was camping with us, so we got her up and called our father to get an ambulance to his house and we went there in my sister's car. I tried to keep Emily conscious all the way there, as she seemed to be falling asleep. We explained all of this to the ambulance men as they took us into hospital (fortunately only a 10 minute ride) and they reassured us that she was probably only dehydrated and a drip would revive her completely. We didn't know what to think. By now I was very worried and numb with the shock of it all, but willing them to be right.

In hospital we were received immediately but lost valuable time (although who knows if they could really have saved her, by now she had been without food since Friday evening, a total of about 36 hours) re-explaining everything. The nurses measured her blood pressure and examined her but did not consider it necessary to take a blood test - again, probably it was too late anyway, but at least they would have seen a fatally low blood sugar level and maybe would have acted quicker - instead they put the anesthetic cream on her hands and arms to numb the places where they intended to attach the drip and said we had to wait for it to take effect (feeling reassured at this point, we sat down to wait. I don't know how long we waited, but it was time enough for my sister to come and fetch Mauro to go and get his mother who was also over in GB for the christening (to bring her to hospital so she could see all was ok) and tell everyone else to go on with the day's arrangements and take Ben to the christening with them. I was carrying Emily to the bed assigned to her for the drip to be attached when she arched her back, made that awful noise in her throat that I will never forget and we lost her. Mauro and my mother-in-law arrived right then to see the doctors desperately trying to get her heart to beat again, but it was no use. At that point shock and automatic pilot took over. I remember certain things and am a bit hazy about others... the hardest thing was having Ben come home to my father's and asking straight away "Where's Emily?" How do you explain to a 3-year-old boy that his little sister has gone to heaven and from now on he can send her his kisses and smiles by
**Emily & Grace’s Story... cont’d**

looking up at the sky? Ben was the sole reason Mauro and I managed to go on, I think. His need of our love and support meant we had to react and get on with things somehow. Our families were wonderful. Of course they too were mourning the loss of a granddaughter or niece, but I think that Mauro and I had to find our own way to come to terms with it.

The doctors in Exeter were very thorough and when finding that the autopsy could not give a clear cause for her death, they sent skin grafts to a specialist centre in Sheffield and the DNA test came back with the MCAD diagnosis. Ben was tested straight away to confirm he did **not** have the same disorder (almost certainly so as he had already been in lots of situations where a crisis would have presented itself), although we do not know if he is a healthy carrier of MCAD.

Once back in Italy, we were put in contact with Doctor Alberto Burlina in Padua, who is one of the few doctors in Italy dealing with metabolic disorders. We are lucky that Padua is only an hour away from here by car. Speaking to him, we began to understand more about Emily's disorder, face the possibility of having another child, and all the implications that would have. **When Grace was born (December 30, 2002) and diagnosed with MCAD when only 8 days old;** we immediately joined the Association Asmme (a family association in Italy for patients with all types of metabolic disorders) that has given us a lot of support and most importantly allowed us to know other families with children diagnosed with MCAD.

These first 2 and a half years of Grace's life have not been easy, she was in hospital several times in her first year for various illnesses, which necessitated her being fed by drip until she was self-sufficient again. The second year was easier, although she had salmonella and a stomach virus at the same time in October 2004 and was in hospital for 2 weeks. It's up and down really, it seems, isn't it? **Grace is a happy, very active child and appears to be growing well as any healthy child.** She speaks both English and Italian and is very mischievous – her greatest trick so far was cooking the telephone in the microwave!!

When Grace is well, she eats well and all is well, as soon as she gets a bad throat or teething trouble or worse, things go a bit astray. In December 2004 she had a month where she refused to take her night bottle, and we had to give it to her with a syringe: no fun for her but the only way to make sure she took it. Generally she does well. She's about 92 cm tall and weighs 12 kgs 500 gms. Her diet is low fat of course, she doesn't drink milk, but a rice-based drink, and we avoid giving her butter, cream cheeses and all fatty products. Being in Italy, home of pasta, she has a pasta- and rice-based diet, with plenty of fruit and vegetables, fish and lean meat, cereals etc. Fortunately she does not have any food intolerances and is keen to taste new things. She has taken 1 gram of carnitine a day since she was diagnosed and I imagine the dose will increase as she gets older.

We know a family near Padua who have a 5-year-old son with MCAD (they too lost another daughter when this son was 2 months old and then he was diagnosed, and they have a healthy 7-year-old boy too – it's amazing how the same pattern seems to repeat itself again and again!), but as far as we know there are very few other cases in Italy. Naturally there would be far more cases in the USA, but it's incredible to know you have 450 MCAD members in your association (+ 400 of various other FODs), when we are about 500 metabolic patients of all kinds in Italy, although Doctor Burlina is convinced there are many more who do not yet know they have a metabolic disorder.

Mauro and I tried to make contact in GB with some form of association through one of the doctors in Exeter a while back, but didn’t get very far. I intend to spend some time in the summer while on holiday contacting groups in GB. We are very pleased to have met you and to have the chance to exchange information and ideas and stories with you and other American families. The more people you can talk to and listen to, the greater chance you have of giving your child a happy and normal life. Having come into contact also with children with far more complex disorders than MCAD we also realize how lucky we are that our daughter can lead a normal life as long as we are careful and attentive to her diet and eating times.

I realize that I have been writing this to you for about an hour and it's time to make Grace's dinner, so I better go now. **Telling you Emily's story has been hard but I am pleased I have done so.** There's a lot more to say about these last 2 years, but that will come later.

I wish you all the very best and look forward to keeping in contact with you in the future. Best wishes to you and your family and all the members of the FOD Group,

Sue Udina
sue.mauro.mizz@katamail.com

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**URGENT NEED for Medical Professionals**

With more Families being identified with an inborn error of metabolism (through expanded newborn screening), our Families will need ongoing Clinical Care from knowledgeable and caring professionals. In addition to our Newborn Screening Advocacy, our Group is hoping to bring awareness to medical schools and other medical organizations and facilities the need for educating and training new Professionals (physicians, metabolic nutritionists etc) in the field of Medical Genetics and Metabolism to treat our children, as well as our FOD adults. If you are a medical student or in a related health profession, and you are interested in gaining more information about this exciting and challenging field, please email me (deb@fodsupport.org) and I can possibly get you in contact with some of the clinical physicians listed in the Diagnostic Lab information. We NEED your help NOW and in the FUTURE so our children will thrive and grow into adulthood with the best of ongoing care!
During the many years that I have practiced clinical social work, I worked with the developmentally disabled population. I have helped individuals and family members better cope with the challenges with which they were faced. Helping them accept “why me” and gain an acceptance for these life-long challenges was a large part of what my job entailed. Ironically, I now find myself in a parallel situation. This time, I am not the therapist rather I am the parent.

Having a healthy child after a long endeavor to get pregnant, I was thrilled the second time around to have far less challenges with conceiving. While pregnant with Evan, my second child, it was recommended to me by a family I know to have an expanded newborn screening done simultaneously as the state mandated testing. The test was about $70, why not have it done? After all, better be safe then sorry. I read the packet, filled out the forms and felt confident that I was doing the right thing. I also felt strongly that I would never be dealing with any of these rare disorders beyond reading about them on these forms.

Evan arrived and all was well, or so we thought. He was six days old when the pediatrician called and instructed us that we had to repeat the newborn screening. We did not even rush back to the hospital to repeat the blood work until the next day when the doctor firmly instructed me to get there ASAP. Five days after that, we found ourselves in a geneticist’s office feeling very confused. We were learning that Evan has SCAD, short chain acyl-CoA dehydrogenase deficiency, a rare metabolic disorder for which there was little information and research. Basically, we were told that all we had to do was feed Evan every 12 hours and he would be fine. We have since learned through other doctors and our own research that we needed to feed Evan every four hours around the clock for several months, at which point we would be able to increase slowly the span in which he could sleep at night while maintaining the four hour daytime regimen.

My husband and I found ourselves faced with a disorder that to this day I am not clear how to say it or what it means. I just know that we are so very lucky to have the knowledge that Evan has SCAD. It’s been a long 16 months. From waking up every few hours to the other medical issues, I am tired and exhausted. At two and a half weeks, Evan was not nursing so we found ourselves in the emergency room frantic that his sugar levels had dropped. After blood tests, an IV of D10, lots of tears and much anxiety, we were thrilled to learn that Evan’s sugar levels were within the normal range. At this time, Evan was put on formula so that we could better monitor his intake and he was prescribed the first of many reflux medications due to continually spitting up since birth. By seven weeks, Evan’s spitting up evolved into projectile vomiting and he was diagnosed with Pyloric Stenosis which required immediate surgery for correction. Of all the kids to have a problem with keeping food inside him, why must it be the one child who has to monitor every ounce of intake? At present, Evan has had ten ear infections in the past six months so he underwent surgery to put tubes in his ears yesterday. This common out-patient procedure for most has become, for Evan, an overnight stay at the hospital for preventative reasons. He had an IV of D10 starting the night before so as to ensure that his blood sugar never dropped below the normal range.

We are a very lucky family - we know about Evan’s disorder. So for all of our sleepless nights and frustrated days, we are truly thankful for them because we know that for Evan and us, without the knowledge of SCAD, could be so much worse.

The irony of all of this is that we learned via DNA testing in Denmark that Michael, my husband, has SCAD. For him, he never had an underlying medical issue that went unexplained until now therefore SCAD has minimally changed his life. I, myself am a carrier of SCAD but do not have it. Other family members have undefined versions of SCAD for which there is little to no concern. Alexis, my three-year-old daughter, though a carrier a carrier of SCAD but do not have it. Other family members have undefined versions of SCAD for which there is little to no concern.

In all the years that I’ve worked with families confronted with an entirely different problem, I find myself now faced with an enlightened level of support for which I can apply both professionally and personally.

My friends think it’s been so tough for us because we are so tired and cannot allow Evan to sleep through the night. I think some of them think of Evan as a “sick” child but what they may not realize is everything we do for Evan is to keep him as he is, a healthy 16-month-old little boy. Our friends also know about the unrelated medical things that Evan has endured. That said, many of our friends do not realize the appreciation we have for our endless fatigue and tireless efforts to always do the best for him.

We feel so lucky to have learned of Evan’s SCAD so that we can work hard towards having Evan grow-up to be another “normal” little boy.

Lauren Hammer
Westfield, New Jersey
415hammer@comcast.net
We followed 14 children with LCHAD or TFP deficiency for 2-5 years. Physical, biochemical and ophthalmological evaluations, including electroretinogram (ERG) and visual acuity by evoked potential (VEP), were performed at baseline and every year following the beginning of docosahexaenoic acid (DHA) supplementation and continued treatment with a low-fat diet. 65 mg DHA per day was provided for children under 50 pounds and 120 mg DHA per day was provided for children over 50 pounds in a concentrated gel cap that provided little other long-chain fat (Neuromins, Martek Biosciences Inc.).

Three children with TFP b-subunit mutations had normal appearance of retina at enrollment and no changes over the course of the study. The other eleven subjects were homogyzous or heterogyzous for the common LCHAD mutation, G1528C. They had no change to severe progression of pigmentary retinopathy with time. Four children had marked to severe chorioretinopathy associated with high levels of plasma hydroxyacylcarnitines and decreased color, night and/or central vision during the study. The plasma level of long-chain 3-hydroxyacylcarnitines, metabolites that accumulate as a result of LCHAD and TFP deficiency, was found to be negatively correlated with the electroretinogram. Children with sustained low plasma long-chain 3-hydroxyacylcarnitines maintained higher ERG amplitudes with time compared to subjects with chronically high 3-hydroxyacylcarnitines.

VEP appeared to increase with time on DHA supplementation and there was a trend for a positive correlation with plasma DHA concentrations. Thus, optimal dietary therapy as indicated by low plasma 3-hydroxyacylcarnitine and high plasma DHA concentrations was associated with retention of retinal function and visual acuity in children with LCHAD or TFP deficiency. Optimal dietary therapy was defined as maintaining low plasma hydroxyacylcarnitine levels (the sum of the long-chain hydroxyacylcarnitine species under 2 mmol/L) with a low-fat, MCT supplemented diet and moderate DHA supplementation.

“Effect of optimal dietary therapy upon visual function in children with long-chain 3-hydroxyacyl CoA dehydrogenase (LCHAD) and trifunctional protein (TFP) deficiency” (in press: Molecular Genetics and Metabolism)

Melanie B. Gillingham PhD1,2, Richard G. Weleber MD2,3,5, Martha Neuringer PhD3,4,5,6, William E. Connor MD4, Monte Mills MD7, Sandy van Calcar MS8, James Verhoeve PhD7, Jon Wolff MD8,9 and Cary O. Harding MD1,2

From the Departments of Pediatrics1, Molecular and Medical Genetics 2, Ophthalmology3 and Medicine4, the Casey Eye Institute5, and the Oregon National Primate Research Center6 at Oregon Health & Science University, Portland, Oregon 97239 and from the Departments of Ophthalomology and Visual Sciences7, The Waisman Center8, and the Department of Pediatrics9, University of Wisconsin Medical School, Madison, Wisconsin, 53705

Corresponding author and reprint requests to:
Nutritional Update

Nutrition and Mitochondrial Fatty Acid Oxidation Defects
Phyllis B. Acosta, DrPH, RD

INTRODUCTION

Three forms of energy (fuel) are used by the body: glucose, fatty acids and ketone bodies. Glucose, after eating and until its storage form (glycogen) is used up, is the major source of fuel for all of the body except heart muscle. Breads, cereals, fruits, milk, pastas, sugar and root vegetables furnish most of the carbohydrate used to make glucose. Carbohydrate supplies from 45% to 60% of fuel (measured in units called Calories) in the American diet. Some 6% of liver and 1% of muscle are glycogen, the storage form of glucose. Liver glycogen helps keep blood glucose concentration in the normal range while muscle glycogen supplies fuel to muscle. The liver of an adult has 12 to 18 hours of glycogen while an infant or child’s liver may become glycogen depleted in much less time.

Some amino acids (building blocks of protein) obtained from food protein and body protein may be used by the body to make glucose after glycogen stores are depleted. This may occur after fasting; during illness with fever, vomiting, diarrhea or poor appetite or following injury. Protein sources are cheese, eggs, meat, milk, poultry, seafood and dried beans and peas. Normally 8% to 15% of Calories are provided by protein.

Fatty acids, obtained from food fat, supply about 38% of Calories in the American diet. The breast-fed baby gets about 55% of its energy as fat. Milk, butter, cheese, cream, margarines, meat, oils, poultry, and seafoods supply fat. Fatty acids can be short, medium, long and very long chain. Only two of the long chain fatty acids are essential -linoleic and linolenic acids. The remaining fatty acids help supply energy to infants and children whose stomachs are not large enough to hold all the protein and carbohydrate required to fill all their energy needs. Fatty acids supply over two times the amount of fuel as the same weight of carbohydrate and protein. Fatty acids are the main source of energy for the heart at all times. During fasting and any time liver and muscle glycogen is depleted, fatty acids become the main fuel for the entire body.

Fatty acids are also used to make ketone bodies, an energy source for the brain when glycogen stores are depleted. Other body tissues and organs also use ketone bodies for fuel, as needed. About 25 enzymes and carrier proteins help in this process. Any of these enzymes or carriers may be deficient and cause a mitochondrial fatty acid oxidation defect (FAOD). Mitochondria are compartments within cells that contain the enzymes that change fatty acids to the form of fuel required by the body. Thus, they are called the powerhouses of the cells.

MITOCHONDRIAL FATTY ACID OXIDATION DEFECTS

In order for fatty acids to be used for fuel, they must first enter cells from the blood. This is accomplished by carriers. Although now in the cell, the fatty acid must enter the mitochondria. This step requires that the long chain fatty acids be attached to a hitch called coenzyme A (CoA) by enzymes named synthetases (sin-the-tases). The fatty acid is now renamed acyl-CoA (a-sil-CoA). Pantothenic acid is part of CoA.

Several uses may be made of the acyl-CoAs but we will only discuss what happens to them when all the body’s glycogen has been used. The acyl groups must pass through the walls of the mitochondria and to do so must be fastened to carnitine. Before attachment to carnitine can occur, the CoA is removed. The removal of CoA and connection of carnitine are carried out by the enzyme CPT I (carnitine-palmitoyl transferase) to form acyl-carnitine. The CoA is now free to be used for other tasks as well as to be fastened to other fatty acids. The acyl-carnitine is carried across the mitochondrial wall by carriers (translocases).

Now in the powerhouse of the cells, the carnitine is removed from the acyl group and a new CoA is fastened. The carnitine is moved out of the mitochondria to be reused. The enzymes that carry out these steps are called CPT II. Four different enzymes (dehydrogenases) now act on acyl-CoA groups of different chain lengths (short, medium, long, and very long) to form compounds (electrons). Enzymes carry these electrons to a system of other enzymes that changes them to the special form of fuel used by the body. One group of enzymes requires riboflavin (vitamin B2) and others require niacin to help them work. Defects have been found in all the carriers and enzymes needed to help the body use fatty acids for fuel. One disorder resulting from defects in this system is called glutaricaciduria type II.

CLINICAL SYMPTOM

Inability to use fatty acids for fuel requires changes in the diet to prevent symptoms that may lead to death. Low or very low blood glucose concentrations brought about by fasting, injury, infection or other stress may occur in patients with an FAOD. Some of the other most common clinical features of the mitochondrial FAODs are given below:

<table>
<thead>
<tr>
<th>FEATURE</th>
<th>DEFECT/DISORDER</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acidosis, metabolic</td>
<td>Glutaricaciduria type II</td>
</tr>
<tr>
<td>Acute liver failure during pregnancy</td>
<td>CPT I; translocases; very long, long, medium, short chain FAODs; glutaricaciduria type II</td>
</tr>
</tbody>
</table>
Disorders of heart and muscle function | CPT II; translocases; very long chain FAODs: glutaricaciduria type II
---|---
Elevated blood ammonia concentrations | Very long, medium and short chain FAODs; CPT I
Muscle loss with excretion of a compound similar to hemoglobin, brought on by exercise (rhabdomyolysis)(rab-doe-my-o-ly-sis) | CPT II; very long, long and short chain FAODs
Failure to thrive | Medium and short chain FAODs
Fat stores in muscle including heart | Glutaricaciduria type II
Malformations of brain and kidney | CPT II, glutaricaciduria type II
Severe liver disease | CPT I, CPT II; translocases; very long and medium chain FAODs

**TREATMENT**
Several causes for the symptoms seen in patients with FAODs have been suggested. Some physicians suggest that the acyl-Co A groups are toxic when made in large amounts as during fasting and other stresses. Others theorize that free CoA and carnitine may not be present in adequate amounts during stress to carry out their other functions. Increased blood ammonia concentrations, low blood glucose concentrations, and decreased ketone body formation have also been suggested as causes for the symptoms. Because of the differences of opinions as to the causes of symptoms, treatment varies. Also, therapy differs somewhat during an acute illness from that given during long term care. A metabolic physician should examine the ill patient and prescribe the appropriate therapy. If the patient does not live near a metabolic clinical center, the metabolic physician and dietitian will prepare “sick-day” directions for the family/patient to carry with them at all times.

The approaches or strategies used in long-term treatment are given in the table below:

<table>
<thead>
<tr>
<th>STRATEGY OR NUTRIENT</th>
<th>ENZYME DEFECT</th>
</tr>
</thead>
<tbody>
<tr>
<td>Avoid fasting</td>
<td>All FAODs. All physicians agree on this strategy.</td>
</tr>
<tr>
<td>L-carnitine. Administer in amounts to maintain normal plasma free carnitine.</td>
<td>All FAODs. Not all physicians agree on this strategy.</td>
</tr>
<tr>
<td>Restrict fat to about 40% of Calories if one-half the fat is supplied by MCTs (medium chain triglycerides)</td>
<td>Very long and long chain FAODs. Not all physicians restrict fat.</td>
</tr>
<tr>
<td>Restrict fat to 30% or less of Calories</td>
<td>Medium and short chain FAODs. MCTs MUST NOT be used with these enzyme defects. Not all physicians restrict fat.</td>
</tr>
<tr>
<td>Supply required linoleic acid-3% of total Caloric intake.</td>
<td>All FAODs. Deficiency has been reported with severe fat restriction in FAODs.</td>
</tr>
<tr>
<td>Supply 1% of total Calories as linolenic acid</td>
<td>All FAODs.</td>
</tr>
<tr>
<td>Protein to supply 10% to 15% of Calories</td>
<td>All FAODs except glutaricaciduria type II in which protein may require restriction.</td>
</tr>
<tr>
<td>Carbohydrate to supply energy (Calories) not provided by fat and protein. Uncooked cornstarch, after 8-9 months of age, may be used to give some of the carbohydrate Calories</td>
<td>All FAODs.</td>
</tr>
</tbody>
</table>
Riboflavin (vitamin B2), 120-200 mg daily, by mouth, with food. Administer in 30 mg doses for best absorption. Try with glutaric aciduria type II. May or may not help. Try with multiple acyl-CoA dehydrogenase deficiency. Improvement in some patients with short chain FAODs.

Triheptanoic acid
Undergoing clinical trials in very long, long and short chain FAODs, CPT I, CPT II and translocases. Not yet approved by Food and Drug Administration for use.

Creatine monohydrate
Very long and long chain FAODs. Improved muscle function in a few patients.

Sources of linoleic and linolenic acids include canola, walnut, soybean and wheat germ oils. Once a container of these oils is opened, it should be recapped and refrigerated between use to prevent spoilage.

OUTCOMES OF TREATED PATIENTS
Outcomes of patients with FAODs is variable since newborn screening and early therapy have only recently begun. No large-scale clinical trials have been conducted to determine the best approaches to therapy. However, a few case studies and anecdotal reports suggest that outcomes may be excellent with newborn screening and therapy begun in the newborn period.

[Please note (from DLG): With new research, there are some alternative nutritional approaches available so be sure to discuss the various options with your FOD specialist and metabolic nutritionist.]

Recipe Books for really low-fat, healthy recipes:

Recipes from the Weimar Kitchen
Weimar Institute
PO Box 486
Weimar, CA 95736
This one was developed to go along with the NEWSTART Lifestyle Center in CA, and it’s low-fat, no cholesterol, vegetarian, and no added sugar!

The McDougall Health-Supporting Cookbook
John McDougall, MD
c/o PO Box 14039
Santa Rosa, CA 95402

The National Center for Education in Maternal and Child Health at Georgetown University has been awarded a cooperative agreement from the Maternal and Child Health Bureau, U.S. Health Resources and Services Library (MCH Library) services. The MCH Library technology to provide broad access to information about practice for health professionals, policymakers, program administrators, families, and educators.

• MCH Library Web site—an award-winning Web site (http://www.mchlibrary.info) with easy access to information compiled by library staff and electronic links to the best MCH information available elsewhere.
• MCHLine® and MCH Organizations databases - searchable, annotated electronic records on over 18,000 print, audiovisual, and electronic resources and over 2,000 government, professional, and voluntary organizations involved in MCH activities. In addition, the MCH Library provides information assistance available on site and via telephone, postal mail, and e-mail to aid MCH professionals and the public in locating resources.

The MCH Library Phone is (202) 784-9770; e-mail: mchlibrary@ncemch.org, Web address: http://www.mchlibrary.info.

Streaming video presentation: "How Autism Impacts a Family" From talkAutism.
Dr. Robert Naseef gives a heart-warming overview of the traumatic stress parents of children with autism go through, and the positive potential rewards the experience can bring. Running time is 16 minutes. To view go to: http://www.alternativechoices.com/video.htm

Medicaid Reference Desk is an online resource at www.TheDesk.info that explains Medicaid in basic terms, state by state. It gives people with cognitive disabilities, family members and advocates information about what is available through their State Medicaid Plans and waivers. The site also gives information on where to apply for services. We explain each Medicaid service in ordinary language. People can see and hear the information rather than read it. The following 21 states are on the site: AL CA CO DC HI IA IN LA MD MT NH NJ NY OK OR PA RI SC TN WA WI. These 10 states are in the process of analysis and will be launched by September 2005: AZ CT GA ID IL ME MO OH TX VT.
Follow-up: ‘Finally Hearing Our Voices’

When Mary Lingle shared her article (on losing her daughter to an undiagnosed metabolic disorder) with the Texas March of Dimes’ state director of public affairs, Jorey Berry, little did she know that she and her daughter, Erin, would be asked to lobby before three state senators on behalf of expanded newborn screening.

The March of Dimes’ Lobby Day held in March at the state capitol was an eye-opening experience for the pair. As filed, House Bill 790 would provide funding for Texas to acquire tandem mass spectrometry to initially pick up to screen for 27 of the 29 disorders recommended by the American College of Medical Genetics.

“Erin and I were asked to lobby at the offices of Senators [Todd] Staples, [Kevin] Eltife and [Robert] Deuell,” said Lingle.

“I’m so grateful for the March of Dimes’ hard work and the countless volunteers who are taking the time to contact our legislators to get this screening in place.”

The March of Dimes’ public-relations department interviewed Ms. Lingle and her daughter. Ms. Lingle’s article, as well as another expanded-newborn-screening article from The Wall Street Journal, was included in an information packet handed out to all legislators.

On May 4 House Bill 790 passed unanimously. The bill will be posted for a hearing in the Senate in the coming weeks. If all goes well and the expanded screening is mandated, it will take about one year for hospitals to get the necessary equipment and personnel in place to begin the screening.

Ms. Lingle says she knows all too well that numerous Texas babies could be affected during the wait. She will continue to spread awareness about expanded newborn screening.

“I had waited for more than 11 years to have a voice, to make a difference,” said Ms. Lingle. “I think about Candice every day, and I want the senseless deaths and serious health problems that result from lack of expanded newborn screening to stop.”

[Note: Mary is our FOD webmaster and also the mom of Candice, who died from undiagnosed MCAD in 1993. The bill was fast-tracked through the Senate and was signed by the Governor on June 18, 2005!]

Long-Term Follow-up of Infants Identified by Tandem Mass Spectrometry

Screening in Oregon, Iowa and Idaho

Judi Tuerck, RN MS Oregon Health & Science University, Sara Copeland, MD University of Iowa

Thanks to the World Wide Web, we live in an information and communication age. At our fingertips we have the latest and most up-to-date information about almost any subject one can imagine and we can communicate that information instantly with many thousands of people around the world. However, as wonderful as the Internet is, it cannot tell us how all the children and adults with FAO disorders, or any other metabolic disorder, are doing. We don’t know how many children are affected, what kinds of treatments they are receiving, whether those treatments are effective, what kinds of problems might be occurring in children over time or how they will do as adults. Basic information needed by every parent, metabolic treatment center and newborn screening programs. We search for journal articles and textbooks; we surf the Net and join various list serves to try and get as many answers as we can, but these answers are always incomplete and out of date. While millions of documents are available on the web regarding common medical conditions, the web pages for metabolic and genetic diseases are scarce. For instance, Google generates 1,000 times more pages regarding “diabetes” (46 million) than MCADD (41 thousand). The irony is that the answers we seek are there, hidden away in the medical records of all our children.

Long-term follow-up is the process of collecting and analyzing information on persons with the same disorder to determine differences and/or similarities in the ‘functional outcome’. That is, do affected children grow up, finish school and live independent and productive lives? Do they have complications or disabilities? If so, are these the result of metabolic crises, imperfect treatments, differences in gene mutations, or other unrelated causes? For most metabolic disorders these answers are either unknown or imperfectly understood. One reason for this lack of understanding is that there is no comprehensive long-term follow-up for most metabolic conditions. As a result, it can take years before a complication, such as renal failure in MMA, to come to light and many more years before effective treatments are devised and tested.
Long-term follow-up is difficult and expensive to do. It can take many hours to collect data from a patient’s chart, make sure it is accurate, and reported in a format that is confidential and can be compared with other patients. Also, in some cases it is difficult to decide which data should be collected. Some data, such as developmental and educational testing can be impossible to obtain if children are “doing well”, as insurance companies and/or parents do not see the need or refuse to pay for it. As a result, the most comprehensive studies, for example the PKU Collaborative Study and the Childhood Cancer Study, require large expenditures of federal money and have taken years to complete. Yet the benefits of those studies have been enormous to families and patients. Childhood leukemia has gone from a death sentence to curable in the space of one generation. In phenylketonuria (PKU) we now know that phenylalanine control is needed well into adulthood and while dietary treatment is better than it was 25 years ago, we must pursue more palatable treatments and a permanent cure. Without the PKU Collaborative studies for both children and pregnant mothers, we would still be discharging patients from clinic at age 6, with the advice that treatment is no longer necessary!

Metabolic treatment staff and parents feel strongly that expanded newborn screening offers the opportunity to identify many infants before their first crisis. We all know from grim experience the damage that can occur as infants struggle for their lives and physicians struggle to make a diagnosis with that initial crisis. We all expect that newborn screening will make a big difference in the outcome of our children. While we campaign for the addition of disorders to the newborn screening in our states or provinces, we must also campaign for a comprehensive long-term follow-up program. Without long-term follow-up the benefits of screening may not be known for another generation and valuable time will be lost.

Most of the disorders covered by tandem mass spectrometry (MS/MS), except for PKU and medium chain acyl-CoA dehydrogenase deficiency, are so rare that comprehensive studies may be years away. We could speed up this process if we could collect data on all the infants now being detected by newborn screening. These data would give us an “early warning system” that would monitor not only health and development, but also would catalog all treatments, laboratory results, complications, frequency and severity of crises, costs, burdens and barriers to appropriate care. If parents, treatment centers and the government worked together these data, collected anonymously, could be pooled and analyzed on a national or even international level, to give us important insights into what is happening to our children in real time. These data are critical to biochemical and molecular research, the development of clinical studies to improve or evaluate treatments, to advocate for the specialty care and services our children may need and to demonstrate the efficacy of newborn screening.

In 2002, the Center’s for Disease Control (CDC) recognizing the need, began a collaborative agreement with Oregon Health & Science University and Iowa State Department of Health to identify which data should be collected and to develop and test a data collection tool for use in the long-term follow-up of children identified by newborn screening with organic acid, fatty acid oxidation and urea cycle disorders. This tool, a database, has now been developed and is being beta tested by entering follow-up data from the infants identified in Iowa, Oregon and Idaho. By the end of 2005, we will have collected data on approximately 75 children (43% have FAO’s) in our three states ranging in age from weeks to 3 years. We have a functional database, which could be deployed to any metabolic treatment center or state genetics clinic in the country. We are interested in continuing our work on this project. Important next steps include evaluating the efficiency and costs of data collection methods, addressing confidentiality issues, adding additional metabolic disorders and most importantly continuing to collect data on the babies and children. Parents can help by continuing to advocate for comprehensive long-term follow-up for all children being identified by newborn screening.

Even though we have a small number of children, important data from our work are already coming to light. The first is that the incidence of all the MS/MS disorders in our states is about 1 baby in 3,000 births, three times as many as we expected. In Oregon and Idaho, where every baby is tested at discharge from the hospital and again at about two weeks of age, 10-12% of our cases have been identified only on the second test! We know that 15% of all cases were symptomatic before the screening results were known and of these, 2 died. Of the surviving infants, 92% are developing normally, although most are too young for formal developmental testing. Soon we shall know how many have developed ‘crises’ since diagnosis and treatment, and if there is any apparent difference between our states or by disorder in this regard. We want to be able to look at costs of care, not only routine metabolic care, but also emergency care. We want to know who is paying for care and whether there are barriers to getting needed care.

Our dream, ultimately, would be to see the development of a National (International) Metabolic Disease Data Center that could pool data from large numbers of individuals. Such a center could provide valuable information on the incidence of various metabolic conditions and the outcome data needed by parents, treatment centers, newborn screening programs and governments to ensure our children’s futures.

Judi Tuerck has worked for the last 26 years as the nurse coordinator for the Metabolic Clinic at Oregon Health & Science University. She has cared for hundreds of children with metabolic disease and their families. She has also worked to follow-up infants with possible metabolic disease found in the NW Regional Newborn Screening Program to ensure prompt treatment. This last year she resigned her position in the clinic to concentrate her efforts on the implementation of expanded screening, long-term follow-up and educational issues relating to newborn screening and metabolic disease. She was asked to present the Long-Term Follow-Up project data to the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children on January 13, 2005. She is a mom and a grandmother.

Sara Copeland is the Medical Director for the Metabolic Program at the University of Iowa and Medical Director for the Iowa Newborn Screening Program. She has been instrumental in the development of the database and invaluable in moving the project forward.

FOD website: Be sure to visit our website (In the News page) for the current articles on NBS efforts across the country. 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!
Please remember these families in your thoughts and prayers throughout the year

Tammy and Roger Clark
Jenna - Birth Feb 17, 2002 Death Nov 22, 2002

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

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

Sandy and Jon Cooper

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

David and Amy Deshaia

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

Carolyn and Terence Finn
Emily - Birth Feb 13, 2002 Death April 3, 2004

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

Lance and Dawn Goldsmith

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

Shelly and William Grabow
Noah - Birth Nov 18, 2003 Death March 23, 2004

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

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

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

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

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

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

Ralph and Angie Hedrick
Chelsea - Birth Jan 11, 1995 Death Apr 3, 1996
Nikki and Toby Hiatt
Reece - Birth Aug 1998 Death April 18, 1999

Pauline and Bill Hill

Amy and Matthew Hoffman

Brad and Kim Holmes

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

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

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

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

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

Brian and Patricia Karhu

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

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

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

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

Sondra Koehn

Jamie and Tom Lazzaro

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

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

Darlene and Larry Lopez
Marissa - Death Feb, 1999

Heather and Phillip Marsella

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

Randy and Misty McDonald

Christine and Mark McFarland

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

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

Lori and Jeff Michaud

Simone and Michael Miller

Kristen and Ken Mitchell
Nolan - Birth Aug 8, 2004 Death May 16, 2005

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

Verna Parker

Diana and Kevin Patterson

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

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

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

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

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

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

Brian and Cherryl Rosenberger

Janice and Steve Rowland
Welcome to new babies!

On Sep 27, 2004, Margie and George Hindley welcomed Timothy David (unaffected) into the world. Timmy was 6lbs .5 oz and 19 inches long. Brother Jimmy (VLCAD) is glad to be a BIG brother!

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

Jackie Shears

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

Leah and Paul Sofranko
Kyle - Birth Feb 7, 1988 Death Feb 5, 1989

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

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

Anne and Gary Stitt

Lisa and Doug Tennyson

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

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

Darren and Karen Wade

Sirpa and Jay Waananen

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

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

Denise and James Westman
Benjamin - Birth March 11, 1987 Death Dec 20, 1988

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

Karen and James Whiteside

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

Christi and Ronnie Williams

We who lived in concentration camps can remember the men who walked through the huts comforting others, giving away their last piece of bread. They may have been few in number, but they offer sufficient proof that everything can be taken from a man but one thing: the last of the human freedoms – to choose one’s attitude in any given set of circumstances, to choose one’s own way.

Viktor Frankl

Condolences

It is with saddened hearts that we mourn the death of Christy Axsom’s son, Kagan (Ethylmalonic Encephalopathy/SCAD), on January 28, 2005. Kagan was 18 months old.

Our thoughts and prayers go out to the Axsom Family.

We also send our condolences to Kristen and Ken Mitchell ~ their son, Nolan, died May 16, 2005 of undiagnosed LCHAD. They share their 9-month-old son’s story on our NBS page.

All of our FOD children will ALWAYS be with us in our hearts!

Welcome to new babies!
Kids Korner

Simone Andrews (Undiagnosed FOD), UK

Grandma Darlene and Hayley (MCAD), Australia

Michael Larson (MCAD)

Nathan and Abigail Hebab (Still Testing)

Grace (MCAD) and Ben Mauro, Italy

Nickolas (SCHAD), Zachery (SCHAD), and Hope Brent
**Family & Professional**

**Family Donations:** Kim and Tom Pfister in honor of Aedan (MCAD). Patricia and Edwin Charlesworth in memory of Matthew Christerson (adult VLCAD). Jane and Donald Strawser in honor of grand daughter, Abby (MCAD). Madeline and Charlie Dreifus, Joanna Dreifus and Sanford Weisburst, Melissa and Jason Goldman, Jennifer Scharer and Ross Katz, Lori and David Schlewitt, Stephanie and Doug Bushell, Meryl and Andrew Gutterman, Grandma Stephanie Kalfus, Farah and Kenneth Shapiro, Gerri and Jeremy Rothlieich, Amy and Michael Rollins, and Ross Rosen in honor of Alexis Hammer’s (MCAD) 3rd Birthday. Stephanie and Andrew Plaisted in memory of Drew’s (undiagnosed MCAD) 8th birthday. Anne and Lawrence Lodding in memory of Nolan Mitchell (undiagnosed LCHAD). Thank you to all that have bought products from companies on the Internet that support the iGive program of donating a certain percentage to Groups like ours. Please remember, however, that donations are NOT tax-deductible because we are not a nonprofit organization. For more info on the iGive program, visit [http://www.iGive.com/html/refer.cfm?causeid=24970](http://www.iGive.com/html/refer.cfm?causeid=24970).

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

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

**Reminders**

**Families** - Please send TYPED stories by DEC 15, 2005 To be listed on the FAMILY LIST, please return the SIGNED Family Questionnaire or hand-write your information as seen on the current Family List and sign and date it. Continue to spread the word about FODs and the need for screening -- it will SAVE LIVES!

**Professionals** - Please let us know about your research and/or clinical work with FOD Families. Send articles by DEC 15, 2005. Also, please return to Deb the Professional Questionnaire even if you are already listed on the printed Professional List.

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Thank you to Erika Wallace - [erikawallacepa@yahoo.com](mailto:erikawallacepa@yahoo.com) (Mailing Lists), Mary Lingle - [Mcartwrite@aol.com](mailto:Mcartwrite@aol.com) (Web Page) and Brian Gould - [briangould@triad.rr.com](mailto:briangould@triad.rr.com) (newsletter) for all your hard work.

Special thanks to Sigma-Tau Pharmaceuticals, Inc. for their continued financial support.

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

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‘We must become the change we want to see in the world.’

~ Mahatma Gandhi~