From The Editor

Here we are in the middle of a hot summer and only six months away from the new millennium ~ didn't we just celebrate the winter holidays? Time does go fast, especially when you have important goals to accomplish. One of our Group's goals, as stated in our January issue was to determine if testing/diagnosing of the FODs at birth versus much later, possibly after a child has one or more severe episodes, would make a difference in medical costs and complications. A big THANK YOU to all the families that returned our ‘Cost Benefit Analysis Survey.’ The results of that Survey (*in 2000, see our current website for Results), as well as some other articles related to advocating FOD Newborn Screening, are in a separate attachment in this envelope. Please take the time to read the Results and SHARE it with Professionals and Legislators in your state/region whom might be influential in advancing our goal to have ALL children in this country tested at birth for FODs (*as well as the many other metabolic disorders that can be diagnosed through Tandem Mass Spectrometry).

The Results were not surprising to us at all ~ now we just need to SHOW Insurance companies and Hospital Administrators that many of the Costs and Complications could have been PREVENTED if the disorders were detected and treated at BIRTH! Several states (NC, TX, SD, AZ, NE, MA, PA and WI) are making progress in either exploring the possibility of screening FODs or actually doing supplemental screening for some/all the FODs. It’s a good start but we have a long way to go before ALL newborns are tested (*see www.tylerforlife.com for more info on what your state tests for).

As in past issues, we have several families share their child’s story. Thank you for being so open about your family's pain and joy. As difficult as it is to write and to read, these stories DO HELP us deal with these disorders. Also mentioned in the last issue, we would like to hear from families dealing with residual effects of one/several episodes before a diagnosis was made, in regard to LEARNING DISABILITIES and/or ADD/ADHD (Attention Deficit Disorder and Attention Deficit Hyperactivity Disorder). Dawn Dougherty (VLCAD) would especially like to hear from you. Her daughter, Jordan, will be starting 1st grade in the fall. Her cognitive skills are on target but she has some concerns about Jordan's "attention and other executive function problems" and how that might affect her in school. Her doctors have told her that they are most likely related to Jordan's early seizures/metabolic episodes. Please refer to the Family List for Dawn's email and phone number. Also if any FOD Researchers are doing work in that area (ADD/ADHD), we would appreciate any information and then I can pass it along to our Group. Please email me at fodgroup@aol.com if you’d like to share your research or information.

PLEASE NOTE: Because there are so many computer viruses going around, type in FOD and then your topic in the subject of your email. Otherwise, if you email me there is a chance your email will be deleted because I either don't recognize the email address or the subject area has a non-specific topic such as “Hi” in the subject area.

PLEASE ~ to make sure I receive your email, place FOD somewhere in the subject line ~ THANKS!
Families keep sending your Stories and Professionals, keep us updated on your research.
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Letters to the Editor
(Letters/Articles from Professionals/Researchers are ALWAYS welcome too)

Dear Deb: It really made me mad to read about the problems so many people have with their doctors not believing them, and acting like the parents don't know what they're talking about. Our pediatrician and our endocrinologist and nurse practitioner at Children's Hospital of Philadelphia are wonderful, patient, and very caring and listening professionals. Every doctor and nurse should have good bedside manner like they have. However, I've had bad experiences too with other doctors, not related to the MCAD issue, and I know how it feels to get that kind of treatment. They come across like you're imagining things or you're making things up.

You know, for people in my generation and older, some of us have been raised to respect doctors just because they're doctors and they have 3, 4, or more years of schooling than we have. Well guess what? Nobody "gave" me respect when I entered the workforce ~ I had to earn it! Well, when you retain a doctor (or any professional), the tables are turned. That doctor works for you. And that doctor has to earn your respect by doing a good job and answering all your questions, or at least suggesting where you can find good answers, and by treating you with respect. If a doctor isn't listening to your information and your concerns, then that doctor has no respect for you and you need to "fire" that doctor and replace him or her with someone who will respect you.

There is an implied contract in retaining a doctor. That is, the doctor will provide services to the patient, and you will pay the doctor for such services through your out-of-pocket and your insurance. These services include listening to information and answering questions. That's what you're paying for ~ you're paying for the doctor's knowledge. When a doctor doesn't uphold his or her end of the contract, the contract needs to be terminated. In other words, "fire" the doctor. And interview other doctors until you find someone satisfactory.

Our bosses don't put up with disrespectful, poor treatment from us. We don't need to put up with disrespectful, poor treatment from people (doctors) who work for us. There is a phrase that goes, "hire slow, fire fast." Get rid of poor treatment, fast. And invest as much time as it takes to find a good professional.

I acknowledge this is hard to do in a rural community where the choices are limited. Maybe it makes sense to do something drastic like commute very long distances into the city where there are more choices in professionals. Another option even more drastic is to sell the house, quit the jobs, get new jobs and move to a larger community where there are more choices. I'm not saying this is the answer, but fear of the unknown keeps us in bad relationships. We need to learn to control that fear, get out of the bad or unsatisfactory relationship, and find a better, good relationship.

Also, I really suggest dealing with an endocrinologist for the MCAD deficiency and the other metabolic diseases. They fall under this area. And I would look for one in a Children's Hospital. ABCD (US-based Assistance for Babies with Carnitine Disorders), which has since merged with a Mitochondrial Group, did a recent survey and found that more than half of all pediatricians surveyed are unfamiliar with MCAD. It's a disease that calls for a specialist and not a regular pediatrician. I hope this advice helps some people having trouble with doctors.

Yours truly,
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Anna’s Story ~ MCAD

Anna was diagnosed with MCAD deficiency nearly two years ago. She was seven months old at the time. A few days after birth, she went briefly into intensive care with low blood sugar. At seven months old she came down with the flu and we again ended up in intensive care. All the stories leading to diagnosis seem to be very similar, so rather than go through those details, I’d prefer to focus on life since then.

Living with an infant/toddler with MCAD has been fine. I wish I had been able to read that two years ago. Anna is now two and a half years old, sleeping through the night (most nights!), and doing everything her peers are doing. We now hire babysitters and go out in the evenings. If Anna is tired, I let her oversleep, instead of waking her for a snack (*please note that every child is different and it is suggested that children under age 1 continue to have a night-time feeding, as well as older children when they are ill). Most of the time, she looks and acts like any other child. The only difference is when she comes down with a bad cold or flu, we might be in the hospital overnight.

Traveling with Anna has been relatively easy. We don't go to very remote places, but I'm not sure we'd want to take any toddler to a remote destination whether or not they had MCAD. We've had to use hospital facilities abroad (in the US and Spain), and this was relatively easy to manage. Although the doctors had never heard of MCAD, we produced a medical emergency letter that outlined the condition and treatment required. On both occasions, Anna was fine after a short hospital stay, and we continued the vacation.

Now that Anna is old enough, she goes to nursery school and gets her fair share of minor childhood illnesses. Most of these we are able to manage at home. With a bit of help from 7-Up and Coke, her sugar intake is adequate and she recovers quickly. I do hate to see her sharing drinks with runny-nosed children and eating biscuits off the floor, but I also know that she needs to be treated as a normal child as most of the time she is normal.

Anna has been taking L-carnitine supplements for nine months, and the difference has been extremely encouraging. Carnitine is not routinely prescribed for MCAD in the U.K. Through this newsletter and a friend working for a metabolic clinic in Oregon, we found enough information to urge our doctor here to try it. I am very pleased we did. Anna’s energy level is much more stable throughout the day, and in general she has more stamina. I understand that most children in the States are given carnitine routinely, and I hope it becomes the accepted practice here. I since have met with Professor James Leonard, the MCAD specialist at Great Ormond Street (London). He currently looks after 35-40 children with MCAD and is hoping to run a drug trial with carnitine in 1999.

Our pediatrician in London is superb. Dr Ed Abrahamson was on call when we arrived at the hospital. Anna had the flu, but started throwing up blood (in hindsight it was from severe acidosis as her body desperately tried to convert fat). Ed made some very good decisions about her care, and got her into intensive care at the right time. He then followed up with blood and urine tests to confirm the diagnosis he suspected when he first saw her. He provided us with all the information available at the time, and he keeps us up to date with new journal articles. We see him twice a year now for routine check-ups, and we really look forward to seeing him. With such a bright and supportive doctor behind us, I feel very confident. I can also relax a bit when Anna is in overnight as Ed, and his team, knows exactly what to do.

My only ongoing concern now is that Anna's energy level seems to swing quickly as do her moods. We are constantly reassured that MCAD should have no effect on her stamina or her personality. However, much like a parent's intuition about a sick child, I do think there is something going on with her moods, possibly linked to low blood sugar as she improves dramatically with food. She also has terrible tantrums before
breakfast. Have any other parents noticed anything similar with their child/children with MCAD? I would be really interested in hearing from you.

In the past two years, we have had to get through some very difficult moments. However, living with MCAD has become much easier. This is partly due to Anna getting bigger and physically being able to cope with more strain on her body, and partly due to our greater understanding and sensitivity to Anna's condition. It is a constantly shifting balance, but it only gets easier. If I had only known this two years ago, the diagnosis might not have seemed so overwhelming. I am sure that life now will only continue to improve.

The most encouraging thing I've heard so far was a passing comment from Professor Leonard. I asked him about what was in store for us as our daughter grows up. He said he really didn't know, as most children beyond eight years old no longer require any hospitalization (*please note that that statement is not always true in ALL cases of MCAD/FOD). However, an old patient of his recently got in touch to say he had just graduated from medical school! Best wishes to those parents who have only recently been given the news about their children. I know every family is different, but I can honestly say that MCAD has been very easy for us to live with.

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Update on Adam ~ LCHAD

Adam's story was first published in the OAA (Organic Acidemia Association, www.oaanews.org) and FOD newsletters in 12/93 and 6/95 respectively, and an update was published in both newsletters in 1996. It is hard to believe that he is now 8 years old. At the end of our last story we were starting to be concerned about long-term effects of LCHAD.¹

We were concerned in two areas at that time, one being the long-term effect of frequent breakdown of muscle tissue in Adam's body, and the other the long-term effect of a deficiency of DHA⁴ on his retinas. When Adam has an LCHAD attack his metabolism is out of balance. His body is in need of energy sources to perform the voluntary and involuntary activities that keep his body functioning. For most children, short-term energy needs are taken care of by eating carbohydrates. After a certain amount of activity, though, these short-term energy sources become depleted and unless more carbohydrate sources are digested, the average person must go on to utilize the long-term energy source in the body ~ the fat. Any lengthy continuous activity will require the use of fat reserves to keep the body functioning.

Adam, like all of the children with deficiencies of fat metabolism, can only use a very small portion of the fat in his system for energy. Since he does not have the enzyme to convert the fat to energy in the mitochondria of his cells, toxic substances are produced instead of energy. If this state is not quickly alleviated, his muscle tissue begins to break down in an attempt to provide an alternate energy source. The extreme symptom of this metabolic crisis is Myoglobinuria,³ which is the breakdown of muscle tissue to such an extent that it can be measured in the urine. Since our last update in 1996, Adam has suffered many of these Myoglobinuria attacks that have resulted in hospitalization for from 2-6 days. During these hospitalizations he has been given an IV solution of 10% dextrose at over 100 ml/hr to rehydrate his system and protect his kidneys.

There are several reasons why Adam develops Myoglobinuria. If he has even a slight viral infection, his body seems to try to metabolize more fat than his cells can cope with. This also happens when he has a bacterial infection, and with any metabolic stress such as exercise, sunburn, altitude, cold or hot temperature or even emotional stress. Adam's body is very sensitive to dehydration and any of these metabolic stress situations. The first sign we have that Adam is in metabolic stress is when he claims to
have 'hurty legs.' This is severe aching in his legs that usually begins in his calves and works quickly up to his thighs and other parts of his body. Sometimes the onset of these symptoms is extremely sudden and very severe. The severity of the pain though doesn't always lead to hospitalization. Once, for example, we spent about one hour on a brisk sunny but windy afternoon at the ocean, soon proceeded by about a one-hour visit with a friend. After this we went directly to a semi-fast food restaurant nearby. Adam's legs began hurting as we seated ourselves. By the end of the meal, although he drank some fluids, Adam had severe muscle pain in his legs, arms, neck and chest. He complained it hurt to breath and talk. His Dad lugged all 87 pounds of him to the car and home where we continued to hydrate him with fluids like water and Capri Suns and he overcame this incident without hospitalization!

Another time a short round of Miniature Golf was all the exercise/metabolic stress he needed to be hospitalized. We are quite frustrated by these sometimes seemingly random 'hurty leg' attacks. Sometimes when we expect them, they don't occur, and other times out of the blue he gets sick. This is very difficult for us and even though we try to monitor his activities constantly, sometimes all of our attentiveness fails.

We would very much like to hear from other LCHADs and their health care providers who cope with diseases of fatty acid oxidation. We are concerned that if Adam's muscle tissue is so frequently breaking down, it is becoming weaker.

At this time we are particularly anxious because a 15 minute aerobic exercise session at Occupational Therapy 3 weeks ago has lead Adam to complain at least once or twice a week of 'hurty legs'. One week when he was complaining particularly frequently, I brought him for a CPK (Liver Enzyme) test to determine if his muscle tissue breakdown was worsening. His urine was still light so muscle breakdown could not be detected by sight. He was tested on a Friday afternoon (CPK 30,000), Saturday afternoon (CPK 17,500), and the next Tuesday (CPK 850). We don't usually run to the lab for CPK tests every 'hurty leg' episode but since it was the beginning of a 3-day weekend, I wanted to find out if he needed to be hospitalized over the weekend while we could still communicate with his doctors.

I'm sure a lot of you have had to experience a visit to the ER where even with a Protocol, unless they can speak to your child's doctor, very little is done. I don't know what normal CPK is, but when Adam has been seen in the hospital it has gone as high as 165,000. Does anyone else have many unexplained incidents of 'hurty legs' in their Fatty Acid Oxidation children? We think some of Adam's 'hurty leg' attacks are just caused by sore muscles from exercise but how do we tell the difference? We want Adam's Dr to give us some low impact aerobic exercises that he can do a few minutes each day. Hopefully this might help strengthen his muscles so they won't hurt after a little exercise and also help Adam metabolize some of the chubbiness off his middle.

Another long-term concern of LCHAD deficiency results in pigmentary retinopathy. This is gradual pigmentation in the retina causing severely impaired vision. Since 1996, Adam has been participating in a study through the Waisman Center at the University of Wisconsin (*see our current website www fodsupport org for more info), Madison which provides DHA, a component in fish oil that is needed for retinal health. Taking fish oil directly would be bad for children with a severely fat restricted diet. Martex Pharmaceuticals has taken the DHA out of the fish oil so those with severely fat restricted diets can still have an essential element for retinal health. Adam goes to Wisconsin once a year for extensive eye tests including an ERG to determine if the DHA he takes twice daily is helping maintain his retinal health. Since abnormal retinal pigmentation was observed in Adam's eyes in the summer of 1995, no further pigmentation has occurred and his vision is still within normal range.

We know the least about the long term LCHAD side effect of peripheral sensory motor polyneuropathy. We only know about what we have read in research. From our understanding this involves a gradual lessening response of skeletal muscle. Adam is a little delayed in gross and fine motor coordination which may be attributed to his first very severe LCHAD episode when he did lose some muscle mass or perhaps this is the result of some peripheral sensory motor polyneuropathy. We would be very interested in discussing this LCHAD symptom with older LCHAD patients or their doctors. Adam has come a long way from the very sick 5-month-old baby that he was in May 1991. It is very encouraging that with a few modifications of diet and exercise he can do so well. We try as much as possible to let him be a 'normal' little boy. He doesn't play soccer like his brother but does play baseball, is
on a summer swim team, rides his bike, roller blades, and participates in children's little theater. His favorite activity is watching the Disney Channel while dressing up in a costume and making huge messes in every room where he is watching TV.

We don't completely ban him from all very high fat foods. We let him try them so they won't be an enticing 'no-no' but always try to educate him as to what he can and can't eat and how much of each. He is allowed 20 grams of fat a day or 10% of his diet. Other LCHAD children we know are on much more restricted a diet. We sometimes wonder about this because Adam is chubby. Maybe he is eating too much fat or just too much food. His favorite foods are sushi (not raw fish but rice sometimes with fish on it), and plain white rice with Teriyaki Sauce. There are so very few LCHAD individuals older than Adam because most his age and some even younger died of severe LCHAD complications or weren't treated properly soon enough. We really can't know the life expectancy or what long-term problems may result as Adam grows up.

We would like very much to talk to any LCHAD families particularly those with older children than Adam and LCHAD adults. We would also like to communicate with LCHAD families and health care providers about various diets, quantities of carnitine and MCT oil taken daily etc. We have recently lost our marvelous Metabolic Nutritionist and feel at a loss about new diet considerations. We really need knowledgeable input about Adam’s diet. He is very chubby compared to all of the other LCHAD children we have met (four of them) and we are worried that he is getting too much fat or too many calories.

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Footnotes:
1) LCHAD = Long chain 3-Hydroxy acyl Co A Dehydrogenase Deficiency

2) Adam's Current Formula: 18 oz of fat free milk divided into 3 6 oz bottles, 1st one given in the morning supplemented with 1 tsp carnitine and 3T of MCT Oil; 2nd given after lunch around 2pm with the same ingredients as the morning bottle; 3rd given at bedtime supplemented with 1 tsp carnitine, 2T MCT Oil and 3T of Cornstarch. Adam also has ¼ tsp DHA twice daily in 6 individually stuffed size I gelatin capsules.

3) Myoglobinuria: The presence of myoglobin, a globulin found in muscle serum, in the urine as in a deficiency of muscle phosphorylase, in crush injuries, and after vigorous and prolonged exercise in susceptible persons.

4) DHA= Docosahexaenoic Acid

5) Retinitis Pigmentosa: A group of diseases, frequently hereditary, marked by progressive loss of retinal response.


Megan’s Story ~ GA II (MADD)

Our first child was born September 2, 1997. We named her Megan and were delighted to have a little girl. The only problem we had in the hospital was that Megan was a poor eater. We were told all newborns have
some trouble figuring out how to feed, but she would be fine. And, we were sent home with a healthy baby girl, as far as anyone could tell.

Megan continued to have little or no appetite, could not be roused for feeding, and was extremely lethargic. We were told to just keep trying to feed her. After six weeks of maternity leave, I, sadly, had to return to work. This prospect worried me greatly knowing how difficult it was for me to get her to consume even small amounts of milk. I took Megan to a new doctor at this time to get a different opinion. A meeting was scheduled for her to be seen by an occupational therapist to evaluate her feeding skill. The therapist said Megan looked like she was doing fine, although displaying a weaker than normal suck. No follow-up was attempted by the doctor.

Megan was then examined by a third doctor who agreed to authorize a barium swallow test to look for reflux. Result: No abnormality seen. Megan's feeding skill had even further deteriorated and she was not hitting other developmental milestones. She choked every time a bottle was put to her mouth, her lungs and throat were consumed by mucus, and she would turn blue due to lack of oxygen. We brought Megan to the ER, where the doctor looked at her, listened briefly to our concerns, and simply sent us home with no testing and no help.

Again, there was no follow-up done by Megan's doctor to determine what was wrong with her, so I took her to a fifth doctor. His diagnosis was a sinus infection. Her problems only got worse and I resigned from my job. I was appalled that these doctors collectively made light of my concerns when there was truly something wrong with my baby. I suggested that she be seen by an Ear, Nose, and Throat specialist, but the pediatrician did not think that was necessary. He seemed offended that a "mere mortal" would have the nerve to suggest to him what be done. I, however, made it happen.

This was the sixth doctor that had an opportunity to examine Megan. He decided to perform a bronchoscopy, but thought it could wait until after the Christmas holiday. He found no reason for urgency. He was wrong.

On Christmas Eve 1997, we drove four hours in the middle of the night to get Megan to The Children's Hospital of Minneapolis emergency room. We were not sure she would make it. She was so weak. She could not consume any milk. She could not breathe through the mucus, although I suctioned her constantly. She was pale and completely lifeless. At the ER, we were made to wait endlessly. When the doctor did arrive, he deep suctioned what he described as huge amounts of mucus from her throat and lungs. I desperately tried to convey to him how urgent it was Megan receive help. He didn't agree. His advice was to take her home and continue trying to feed her. We objected and asked that she be hospitalized. He said the hospital would be the worst place for her since there were several kids with RSV (very contagious) as patients. We even asked that he call Megan's pediatrician in the hope that we would get some action. The pediatrician said he trusted the ER doctor's recommendation that we go home. We were devastated and too naive to fight against the doctors; so, we took our fading infant home.

We returned to the hospital the next day and Megan had the bronchoscopy done. The result: No abnormal structures. The first turning point came that night, while Megan was in recovery from the procedure. A nurse, Andi, observed my husband trying to bottle-feed Megan. Andi saw Megan immediately choke and gasp for air, unable to swallow any of it properly. She told us Megan was drowning in the milk and made the call to finally have her admitted to the hospital. Andi was exactly right.

Megan's muscles did not work properly to direct the milk to her stomach. The milk was going straight into her lungs, and the doctors missed this! She had developed pneumonia on top of the underlying condition yet to be determined. An NT was placed while several swallow studies and many invasive tests were done to determine the cause of her muscle problem. A test for organic acids in her urine was never done. She was released after two weeks with a GT, a heart/apnea monitor, and a suction machine. No further follow-up was done on the part of the doctors.

We were told that hopefully Megan would “grow out of” her swallowing disorder. We tried to go on with our lives as best we could. Megan started therapy, as it had become quite obvious she was not
developing. At four months she could not even lift her head. Megan had no muscle tone, no reflexes. She made no sounds and had almost no facial expression for months. Megan made little progress in occupational therapy. I pushed her pediatrician to get me a different infant formula, since both the milk and soy formulas caused Megan to throw up. Nutramigen is a formula that has the protein in it already broken down. This formula worked much better for Megan. By nine months, she was able to have tastes of baby food, but still significantly delayed.

An infection or illness was devastating for Megan. Even though we had the advantages of a G-tube, once she would start vomiting, nothing would stay down. She had to be hospitalized with an IV in order to break the vomiting cycle. When Megan was a year old, we moved to Wisconsin and found a new pediatrician. Dr. Naomi Prieto agreed with me that Megan's delays and problems had to be caused by something and we had to know the cause. Megan was seen by the neurology physicians at Children's Hospital of Milwaukee, and urine and blood tests were ordered.

We received word on February 1, 1999, that Megan had a disease called Glutaric Acidemia Type II. Megan was 17 months old. At long last, we knew what was wrong and what to do about it. It is amazing how once we finally had an answer, the pieces of the puzzle came together and her problems over that past year and a half made sense. We had a lot to learn about this disease, and literature was hard to come by. With diligence, I have become as educated as one could be in GA 2. Within two weeks of being on a diet that restricts fat and protein, Megan could crawl for the first time! Within a month, she was babbling, showing some muscle tone, and able to move her body with more ease. Megan also receives carnitine and riboflavin, however I cannot locate the riboflavin in liquid form.

It has now been just two months since her diagnosis, and Megan can transition from sitting to crawling or lying, and back again. She can pull up as far as her knees and walks with speed and stability. She is also saying some words. Megan now has a big, beautiful smile that we are blessed with seeing every day.

I am still bitter about the incompetence we experienced from the numerous doctors that examined Megan. But, I know the most important thing is that we still have Megan with us despite the undue suffering she went through. Megan is the greatest joy in our lives and we are so thankful to have her.

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Taylor’s Story ~ MCAD

Jim and I were told that we would never be able to have children. You can imagine how lucky we feel now that we have two beautiful daughters ~ Casey, 4, and Taylor, 2. Both of the girls had milk and protein intolerance and reflux at birth. We had a lot of vomiting and trips to the ER. When the girls turned one, the problems seemed to go away and we have had a very healthy normal life.

One Saturday morning, Taylor came into our room and got into bed with us. She told me that she was cold. I felt her and tried to tell her that she wasn't cold, but hot. At that point, I realized that she had gone back to sleep. I knew something must be wrong. We talked for a second about the fact that she didn't have a nap the day before and had not been sick. Maybe she was just extra tired. I still did not feel right so I tried to wake her and she would barely respond. We picked her up and tried to get her to talk to us. Her eyes would not stay open. We called the children's RN next door. By the time she walked across the street, I couldn't get Taylor to even open her eyes.

We went to the ER and it took forever to get the triage nurse to realize that we had a serious problem. Finally, we were back in a room and the doctors and nurses were asking all kinds of questions about what kinds of medicine she might have gotten into. I explained that it wasn't that, but they wanted to treat for it just in case. By this time, they had put in an IV and catheter and still no response. The drug they gave her for an overdose did not work so they tried glucose in her IV. It took about 15 minutes and she woke up like nothing had been wrong. We were transported to Children's Hospital and spent the night. Her glucose level
stayed up and so they sent us home with a hypoglycemic two-year-old. They had run a few more tests, but were sure these tests would be nothing because she had ketones in her urine.

A week later, we got a call saying that Taylor had MCAD. We were shocked and terrified about the possibility of losing our little girl. We know now that her carnitine level was three and her glucose was 30. We have had a few other scares in the past three weeks. We now have a glucometer at home to test her when we are worried (*be aware that blood sugar levels can appear ‘normal’ but other levels, such as liver enzymes, could be changing and you don’t know that until they are tested for, so please do not totally rely on blood sugar levels – be open to other ‘observations’ such as lethargy, irritability etc. and trust your own instinct). We have met with a local specialist, Dr A.G. Wiltse of the University of Nebraska Medical Center, and a UNMC nutritionist. I've also consulted Dr Charles Roe of Baylor University. Dr Roe was great to take my call on the spot and did have Taylor's chart in his office. Our four-year-old does not have MCAD and we know how lucky we are to still have our little Taylor to put to bed each night. I will admit that I have a lot of trouble sleeping, wondering if she is okay in the room next door.

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Michael’s Story ~ LCHAD

First I would like to start off by thanking you for having this Support Group and Newsletter. We are new at all of this and it is comforting to know that we are not alone. I would like to tell you my son's story to better help you to understand this letter. I hope your sitting down because it's a long one.

It all started when my wife was pregnant with Michael. It was a horrible pregnancy. She was sick at least 4 times a week for the first 6 months. The seventh month was even worse. She was sick 3-4 times a day. The doctors said that this sometimes happens and that there was nothing that they could do. We weren't satisfied with that answer so we just kept going back until they finally admitted her to the hospital for tests.

After 4 days of tests, the only thing that they could tell us was that her liver was slowly failing and that they didn't know why. They sent her to Jackson Memorial Hospital at the University of Miami. It was there that they first introduced us to the possibility that she could have developed Acute Fatty Liver of Pregnancy. Although they couldn't be sure, they decided to treat her as such. Her liver was barely functioning at this point so they decided to do an emergency C-Section. The only known cure for AFLP is termination of the pregnancy.

Our son, Michael Dominick Gumiela, was born 3 months premature on March 28, 1998. He weighed 2.2 lbs. My wife, after losing almost 5 liters of blood (when the liver fails, the body's ability to clot blood stops), miraculously survived and spent the next 4 weeks recovering. My son spent 2 months in the Neonatal ICU were he won the hearts of all the nurses and doctors with his incredible strength and will to live.

During their stay at Jackson Memorial Hospital, we did a little research into AFLP and the possible causes. We found that one of the causes was a possible Metabolic Disorder called LCHAD that our son might have had. I took all the information we found to the doctors at the NICU and asked if he could be tested for it. They said that they would look into it. Just before Michael's discharge, my wife and I asked what they did about testing him for LCHAD. They said that they looked into it and that they don't think he has it. We demanded that he be tested. They said that they didn't see the need but that they would test him.

They told us that his blood was going to Texas to be tested so it would be awhile. After we took Michael home, we called the hospital once a month to find out if the results were back yet. Their reply was to be patient because it was going to take awhile but not to worry because they really don’t think that there
was anything wrong. This went on for 9 months until I finally said that no test takes this long and to find out what happened or I will. A week later they called me back and told me that it turned out that when the blood got to Texas, it wasn't testable for some reason. Texas called Jackson Memorial and told them to send more blood but the doctor who took the call never told anyone. At this point, I called and made an appointment with a Genetics Specialist to have Michael tested. We tried to get his pediatrician to test him but he knew nothing about the test. I couldn't get an appointment for a while with the Genetics Specialist so I decided to do more research on my own. Michael had already been hospitalized again for Bronchiolitis and he just wasn't right. I knew that there was something wrong but I didn't know how to handle it. The doctors weren't helping. Every time we took him to the doctor and said that he was very lethargic and sleeping all the time, they said that it was just his body fighting a virus and that there was nothing they could do.

It was then that I really started looking into LCHAD and the symptoms. The more I read about it, the more I thought, “This is Michael!” At that point I took Michael back to his pediatrician with all the information I had found and told him to have another look at him. He was admitted to the hospital for tests. He was poked and pricked for the next week and a half. At first he started to get better and look better. Then, all of the sudden, he got worse. They said that it looked like LCHAD but they couldn't make an official diagnosis until the test results came back. At that point, my wife and I thought that he would get better care at a University Hospital. He was sent back to Jackson Memorial Hospital and put into the Pediatric ICU. Ten minutes after he arrived at Jackson Memorial, he went into cardiac arrest. They were able to resuscitate him but he never really recovered. He died 3 days later on Easter Sunday.

So that is the story about my son, Michael Dominick Gumiela. Don't get me wrong though, his life wasn't all hospitals and doctors. For the short time that he was with us, he brought a lot of joy to our lives. But it wasn't enough. I was able to teach him a lot of things. But it wasn't enough. He would smile at least once a day (even when he was sick) and he would make us smile a million times a day. But it wasn't enough. You see, Michael was, in a way, my ‘drug.’ I was addicted to him. When I was with him, I was content. When I wasn't with him, I did nothing but think about him and when I was going to see him. My day was only complete when I would pick him up after work and he would give a smile. Only being able to be with my son for 1 year just wasn't enough and that's the reality that I have no choice but to live with. I keep finding myself thinking about the test.

Would things have been different if he was tested properly in the beginning? Some doctors say yes while others say no. I guess I'll never know and that is something that I'll have to live with. But I keep wondering if there was anything that I could do to prevent this from happening to somebody else. I keep wondering if I can prevent other parents from having to ask this dreadful question. So we've decided that we have to do something. The only problem is we don't know how to begin. I remember reading an article in your most recent FOD newsletter from a doctor stressing the importance of neonatal testing. My question to you is where would I begin to try making neonatal or even prenatal testing a requirement?

I realize that there's probably nothing that I could do and that I'm sure that people have tried in the past, but I have to try something in order to give my son's death some kind of meaning. If his death could change the medical profession and cause them to view Metabolic disorders differently, then maybe I'll think that he didn't die for nothing. I thank you for taking the time to read this and if there is anything that you could do to help me to get started, I would greatly appreciate it.

Michael F. Gumiela
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‘Semantics: Words play a part in how families perceive and deal with a chronic disorder’

One year after Kristen died suddenly from what they initially called Reye Syndrome, Kevin was born. Kristen is NOT here because her MCAD was UNDIAGNOSED and Kevin IS here because his MCAD WAS diagnosed at birth – which is when ALL children should be tested! At age 13, Kevin is
strong and intelligent and very active being a teen! We all KNOW how THAT can be! And we have Brian (11½) coming up quickly on the teen scene as well.

And to think it could have been so different if 14 years ago we KNEW about MCAD! Because it was, and still is to some degree, an unheard of disorder among MANY professionals, when we heard Kevin’s diagnosis we were devastated and frightened because we immediately thought it was another ‘death sentence.’ We felt Kevin would get ill and die. It wasn’t until we gained more information by traveling to see Dr Roe and gaining daily experience of giving the Carnitor® and making sure he ate often that we moved from perceiving his disorder as a disorder and NOT as an illness.

After the shock of hearing his diagnosis finally wore off, we CHOSE to view his disorder as a FAMILY CONCERN, not Kevin's problem ~ we were going to deal with this as a family and not try to make Kevin feel like he was so different, even though his disorder IS different than what most have heard about.

Even though we as a family chose to perceive his MCAD this way, oftentimes the outside world had a completely different view. At first I would just fume when people would say “He doesn’t LOOK ill.” And I would tell them (taking deep breaths and trying to stay calm!) ‘He’s NOT!’ He has a disorder that we have to watch closely on a daily basis, but it doesn't have to overSHADOW our lives as doom and gloom.

We CHOOSE to use ACTIVE, not passive words ~ words that TRANSFORM and ENLIGHTEN ~ instead of darken and cause one to stay stagnant. I used to get upset (actually I still do!) when people would say parents should just ‘ACCEPT’ their child’s death (or diagnosis) and just move on. Yes, moving on IS important and it takes a great deal of courage and grief work, but I, speaking only for myself, certainly DO NOT ACCEPT Kristen’s death! And I don't mean that I deny it.

If you recall from the last newsletter I referred to what I call ‘residual grief reality’ ~ I don't believe ANY parent ever FULLY ACCEPTS a child’s death ~ that’s part of their new reality. ‘Accept’ is NOT an acceptable word to me and for how I see my grief and working through it. It conjures up for me a “Get with it, Deb, and get over it” perception ~ not one I that I will own!

I prefer to use the word ACKNOWLEDGE over accept ~ I KNOW and UNDERSTAND that Kristen died from undiagnosed MCAD (and I KNOW Kevin has MCAD) ~ that doesn't mean I HAVE to ACCEPT that in order to grieve in a healthy way! People can argue and say that I’m playing with words ~ I’m not ~ I’m just trying to express myself with words that have more meaning to me and grasp how I SEE and HONOR my FEELINGS and THOUGHTS about life and loss and death.

If you look at your own life, you can probably trace back where your own beliefs about life, adversity, loss and death were developed ~ and those early beliefs, role models, religious/cultural beliefs and rituals, etc., play a part in how you use language and how you act out those words in behavior. My ‘loss history’ started very early ~ as a 3-year-old with 5 siblings under 10 years old and a mom dealing with the sudden death of her husband and my ‘papa’ at age 36! I’m sure many of you have been ‘there’ yourselves ~ and it most definitely has an impact on how you SEE things as you grow up and how you grieve ~ IF you grieve ~ when a child of your own dies or has been diagnosed with a genetic disorder.

Kevin DOES have a chronic disorder that will be with him for a lifetime, but that doesn't mean he is ILL, as I mentioned before. He may have some days where he gets sick ~ and we don't take those lightly ~ but we try to keep them in perspective. We make sure he gets what he needs, we know what we have to do and we do it! Overall on a daily basis he is as ALIVE as any of us! Ask his brother, Brian!

Don't get me wrong ~ I'm not trying to sugarcoat these disorders or reality by using ‘New Age’ type words. I am just sharing with you how I SEE things and which words I CHOOSE to use to explain my vision of MY reality and how it's made a difference in how I cope with MCAD and the ramifications of it going UNDIAGNOSED. The point I'm making is that no matter what adversities are ‘thrown’ at us, we STILL have a CHOICE in HOW we perceive those circumstances.
It has been shown in research (in my own unscientific words!) that those that perceive the cup half full rather than half empty get through adversities in a more positive manner and are probably healthier for it! If you live in negative self- and other-talk ALL the time it takes its toll individually, as a family, and in all your relationships.

We choose to see both Kevin and Brian as they are – individuals trying to grow with each day, making mistakes along the way – as we ALL do – and trying to have some fun in the process. It doesn't negate that we are serious about his disorder but it sure does lighten things a bit as far as stress is concerned. That didn't come overnight though. Those of you that are just beginning this journey let me tell you that I DO understand where you are at!

It IS a reality in our family that we have already experienced one death due to UNDIAGNOSED MCAD and that there will be illnesses along the way for both our boys, but that doesn't mean I have to think of Kevin as ILL all the time just because he happens to have a metabolic disease. In fact, in order to defuse the ‘medicinal’ atmosphere of having to take the Carnitor®, we use the word ‘vitamin’ instead of medicine. To me, taking medicine is usually in the context of being ILL and Kevin is NOT ILL. So in order to promote a more uplifting atmosphere we use a word that signifies NURTURING HEALTH instead of trying to fend off ILLNESS.

After all my rambling, you probably get the idea that words impact perceptions and vice versa. Take, for example, how one views the percentages given each child for having an FOD. There IS a 25% chance of having it, but there is a 75% chance of NOT having it – which do you CHOOSE to focus on? Of course, when a couple is trying to decide whether to have more children, it all depends on your very own individual circumstances. You may already have a child that is experiencing several difficult effects of serious episodes and requires a lot of care and energy on a daily basis, so you may make the personal decision not to have more children. Yet, even in that circumstance, you still have a CHOICE of HOW you are going to perceive those circumstances ~ as a challenge or as a burden ~ it makes a difference! Your perception will definitely influence your actions toward your child and the rest of the family. If we as parents spend so much time in what we have ‘lost’ (i.e., the so-called perfect healthy child) we miss out on what we DO have – children that REALLY need us and love us and bring joy to us no matter what challenges are ahead!

If you recall the book reviewed in our January ‘98 newsletter, Special Children, Challenged Parents, Dr Naseef stated that point in a similar way – acknowledge that your child has medical concerns but try and meet him/her where he/she is at and move forward from there. Try to be aware of your own use of language when talking with others or even within your own self. It carries over to all aspects of your everyday living. By changing your words your overall perspective can be transformed from feeling victimized, burdened, and destined for years of pain and sorrow to a life where you learn to control what you CAN control, you view adversities/obstacles as challenges, and you make a commitment to always advocate for your child/children. And in so doing, you not only make a difference in your own life, but in the lives of your children, family, friends and others that come into your life! All by envisioning new ways of using words positively and actively.

I would give anything NOT to have to deal with Kristen’s death and Kevin’s disorder and I wish there was no such thing as an FOD ~ but the REALITY is, in MY eyes, I HAVE CHosen to deal with them, as much as I don't want to ~ I NEED to!

What it comes down to is this ~ You can choose to be DEFEATED by having to deal with these disorders or you can CHOOSE to be an ACTIVE ADVOCATE for yourself, your children, and children yet to be born! I have great FAITH and HOPE that the LOVE you have for your children, both living and deceased, will show you the way and give you the strength to NEVER GIVE UP!

Deb Lee Gould, Director
July 21,1999
Kristen’s 14th ‘anniversary’
Resources

**FOD Email List** ~ Great for Internet networking with others that are dealing with an FOD. Sign up by going to the FOD web page at [www.fodsupport.org](http://www.fodsupport.org), and click on ‘Join our Email List’ at the top of the homepage. We currently (*in 2000, we have 200 members on the List*) have about 45 members signed on from 16 different states/provinces and 7 countries!

**New Canadian-based ABCD Support Group (Assistance for Babies and Children with Carnitine Deficiencies)** ~ Contact Shelley Levesque, 146 Becker Avenue, Belleville, Ontario, Canada K8N 3T7 (613) 968-5264.  [shelley_lev@hotmail.com](mailto:shelley_lev@hotmail.com)

**New CPT Website** ~ Go to [www.spiralnotebook.org](http://www.spiralnotebook.org) or contact Diane Martinson for more info at [dianem@pclink.com](mailto:dianem@pclink.com)

**‘Stacey’s Cake’**

*A Low Fat Birthday Cake Recipe*

- 2 small boxes Fat Free French Vanilla Pudding
- 3 C skim milk
- 1 8oz container Fat Free Cool Whip
- 1 box low fat Graham Crackers
- 1 can reduced-fat chocolate icing

Combine pudding, skim milk and cool whip in large mixing bowl. Cover the bottom of a 9x13 glass dish with a single layer of graham crackers. Pour ½ of the pudding mix into the pan and spread evenly over graham crackers. Add a second layer of graham crackers on top of the pudding. Pour the remaining pudding mix in the pan and spread evenly again. Add a third layer of graham crackers and top with the chocolate icing. It’s best to make this a day ahead of time so that the graham crackers soften. Approximately 3 grams of fat per serving.

Lisa and Jeff Schmidt  
[jschmidt@cintemet.net](mailto:jschmidt@cintemet.net)

**Pharmaceutical Update**

Sigma-Tau Pharmaceuticals, Inc., makers of Carnitor® can be reached at 800-447-0169 or on their webpage [www.sigmatau.com](http://www.sigmatau.com).

**Medical Update**

**DHA Supplementation in LCHAD**

NOTE: *The Results of our ‘Cost Benefit Analysis Survey’ and additional articles about advocating FOD Newborn Screening are enclosed as a separate attachment (*in 2000, see our current website for the Results, under Newborn Screening)*.

For the past two years, the Biochemical Genetics Clinic at the University of Wisconsin has been conducting a clinical trial to investigate the effect of docosahexaenoic acid (DHA) supplementation on the eye sight of children with long chain 3-hydroxyacyl CoA dehydrogenase deficiency (LCHADD). One of the unique symptoms of LCHADD is pigmentary retinopathy, a disorder in which pigment gradually deposits in the retina of the eye and gradually leads to loss of vision. Most children with LCHADD develop some vision problems, even if they have good metabolic control with strict dietary treatment.
DHA is a fatty acid found in all membranes. It is found in the membrane of the retina at very high concentrations. In fact, about 40-50% of all the fatty acids in the retinal membrane are DHA so it clearly is important for the functioning of retina. **Several studies in animals and infants have shown that deficiency of linolenic acid and DHA decreases retinal function.**

**Children with LCHADD are at risk for DHA deficiency** because of the tight restriction in fat intake that is necessary to treat this disorder. DHA is made in the body from a fatty acid called linolenic acid. Linolenic acid must come from the diet because the body cannot synthesize it from other fatty acids. Thus, if the restriction of fat does not include sufficient sources of linolenic acid, it is possible to be deficient in linolenic acid and thus, deficient in DHA. There is also evidence from this study that synthesis of DHA from linolenic acid may be adversely affected in long chain fatty acid disorders.

Currently, 9 subjects are enrolled in this study. Their age range at the initiation of DHA supplementation was 18 months to 13 years. Baseline evaluations of eyesight showed below normal or low normal Visual Evoked Potential (VECP) results in all but one of the subjects. Seven of these subjects have been on DHA for over six months and have had at least one set of eye tests after starting the supplement. **In six of seven subjects the VECP results significantly improved with supplementation.** Because the initial eye testing results are positive, we are currently seeking funding to add additional subjects and expand the study to further investigate DHA metabolism in LCHADD. If you have questions about this study or are interested in participating, please contact the Biochemical Genetics Clinic at the University of Wisconsin-Madison or the Oregon Health Sciences University.


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**Love Messages**  
(Please see our most current online issue)

‘Your heart will always be where your treasure is’  
~ The Bible
FOD Notable ~ New Baby!

Congrats...New Baby! John, Jenny, and Jane (LCHAD) Carroll are pleased to announce the birth of their daughter and sister, Megan Rose, born on April 27, 1999 ~ weighing 8lbs and 20” long. She is not affected by LCHAD. Jenny, THANK YOU for being so instrumental in helping to network many of the LCHAD Families!

Family and Professional Donations

The FOD Family Support Group would like to thank recent contributors: Barbara and Joseph Brown in memory of Amber (VLCAD), Greg Bingham in honor of Lindsay (SCAD), Dr. Larry Sweetman, Institute of Metabolic Disease, Dallas, TX, Deb O’Brien (Adult FOD), and Mr. and Mrs. Charles Tuggle in memory of Trevor Dean Patterson (MCAD). We greatly appreciate donations to help with postage and copying. Be aware, however, that because we are not officially a non-profit organization, donations are not tax-deductible at this time. Checks can be made payable to Deb Lee Gould. Please note on the check that it is for the FOD Family Support Group.

Thank You!

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

Reminders

FAMILIES: Please send TYPED stories by December 1, 1999. 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 December 1, 1999.
Kids Korner Scrapbook

The Cajuste Family -
Jay - 3, Ashley - 1 1/2 (MCAD), Nicole - 8

Henry Furler -
9 months
(Undetermined FOD)

John, Jane (LCHAD), Jenny & Baby Carroll

Jennifer Miller -
2 1/2
(MCAD)

The Pennington Family:
Kim, Terry, Justin (LCHAD), Autumn (LCHAD), Christopher and Nathan

Dakota Eads - 2
(LCHAD)

Brian, Kristen (MCAD) and Kevin (MCAD) Gould
‘Allow yourself to fully take in what this day will bring…
knowledge is a way of loving ourselves.’*

~ Virginia Satir

‘The unfolding of a new day’ in Virginia Satir Meditations & Inspirations
Edited by John Banmen & Jane Gerber, 1985, Celestial Arts

*"Use with permission of Avanta The Virginia Satir Network,
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All rights reserved.”

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

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

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