

FOD Communication Network
(Fatty Oxidation Disorders)

'All In This Together'

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Volume 7 Issue 2

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

From The Editor

Another summer is upon us. I hope all of you are doing well. The Goulds are still 'hanging in there' as best we can. We just try to keep on moving. Of course, with both Kevin and Brian in the activities all year it is kind of difficult to be completely still ~ although, we do have some quiet moments. I, for one, look forward to them ~ great stress reliever!

This issue is packed with information and several family stories, as well as ideas for our Support Group LOGO. We greatly appreciate all your input ~ keep it coming! In this issue we learn more about GAI from the Myers family. This is one of the more rare FODs. I hope more of you that are coping with this disorder, as well as CPT, GAI/MADD, SCAD or the other more rare FODs will consider sharing your stories with our Network.

Eric Schmid and Judy Farrell have offered some ideas for our LOGO and hopefully by our next issue (Jan 98) we will have more ideas submitted. We would like to hear from you about those ideas or what you think about Eric and Judy's designs. So please **send your creative suggestions to us by December 1, 1997.**

I have also included a letter I wrote to the NC legislature and to some health professionals on the State Newborn Screening Committee to give you an example of what might be addressed in your own letters (if you choose to take this approach) in order to get your state/region interested and aware of the importance of making FODs a part of their mandatory newborn screening tests. Finding the necessary funding is a challenge, but connecting with a state or local politician may be an inroad to presenting a bill that would start the ball rolling. At the last moment, here in North Carolina the committee found the funding for a 1-2 year pilot study on testing FODs in newborns, so presenting a bill was not necessary, although I was able to enlist the help of a State Representative who was very willing to present a bill. My many letters did not go to waste ~ people do respond when they KNOW it is a VITAL issue that could help save future children. So try it ~ you may surprise yourself!

I hope this issue is helpful and spurs you to think of ways to make others aware of these rare disorders! **Each one of us CAN make a difference because 'We Are All in This Together!'**

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Letters to the Editor

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

Dear Deb and Dan: Thank you for the great work you do with the FOD newsletter and Network. When Maggie (age 7) was first diagnosed six years ago, I was frustrated with the lack of information available on MCAD. I know that families with newly diagnosed children are comforted by the info and network around them. Luckily, Maggie has no visible effects from MCAD. She has gone through several short bouts (12-24 hours) of intestinal flu where her intake of fluids and food were minimal without hospitalization. She is healthy and active with none of the symptoms that many of the other children suffer. We still maintain the low fat diet and carnitine and know that she could not survive untreated through extended fasting but we are one of the lucky ones and we are very grateful. Thanks again for all of your hard work.

Scott Dozier
Atlanta, GA

Dear Deb and Dan: Happy Holidays 1996 to you and yours...may they hold some incredibly warm and loving moments for your family that will become treasured memories of yours for many years to come!

First let me say that your newsletter is wonderful! I'm sure it brings much understanding camaraderie, power, and peace to many of us out here that have felt isolated, ignorant, inadequate and alone. 'We Are All In This Together'...and the time and energy you've shared have been a true gift to many. Thank you so very much!

I've attached our story. It's a letter I wrote several years ago to our first daughter, Hannah to let her know of what she started to teach us about herself on April 28, 1993 when she was only 4-years-old. She was then diagnosed with Glutaric Acidemia Type II. Those first few days were the scariest in our lives to date. Writing her the attached letter was very therapeutic for me at the time. I would highly recommend other Moms and Dads write such a letter to their loved ones.

We all (Hannah, her sister Emma, her Dad, Tom, and I) have come a long way since then. We now feel comfortable with Hannah's disorder and take life a day at a time, just like everyone else. Last month we made the bold move to have her 'specialist's visits' move to once a year, instead of the previous every several months check-ups. Hannah has been 'aware' and in control of her bodily needs from her first breath. She is more knowledgeable than any of the many doctors we have seen. We are proud of her and feel extremely blessed to have her in our lives.

I wanted to be sure to let you and others know what special medications she takes daily in hopes of sharing some knowledge with others that live with metabolic disorders. She's on a very limited protein diet ~ approximately 5-8 grams per meal (or about 24 gm a day). This doesn't seem to be much of a problem for her, as she craves and eats a lot of rice, hash browns, cold and hot cereals, non-dairy milk, potato chips, lots of vegetables and some fruits. Daily she takes one chewable multivitamin with fluoride and iron, one capsule of B-2 (riboflavin) 100 milligrams, and twice a day 1 teaspoon of Bicitra (sodium citrate and citric acid oral solution, UPS), and at each meal one regular Tums (calcium) and one 330 mg tablet of Carnitor®. I want to be sure to share that she takes the Tums and Bicitra, as we have run into others that haven't thought about them. The Tums is for the calcium, of course and the Bicitra is a stable systemic alkalizer containing sodium citrate and citric acid in a highly palatable sugar-free base. It is useful in the management of metabolic acidosis especially when the administration of potassium salts is undesirable or contraindicated. We were told that Bicitra will "widen her band of metabolic normalization."

So attached please find our Hannah's story. One major addition that is not part of the attached is that through our in-depth medical computer searches regarding GAI, we learned via postmortem autopsies that individuals diagnosed with GAI also showed a deficiency in L-carnitine. I went to Hannah's specialist with a request to have her blood L-carnitine checked and he flat out told me no. I pushed him and told him I would find another doctor if he won't okay the test. He then agreed.

Well, it came back positive that Hannah is extremely deficient in L-carnitine and that she'll need to take it (Carnitor®) for life 3 times a day. The doctor apologized. I think **this speaks to the need to 'listen' to your heart/gut about things parents feel their children need. Many times doctors are held in such high esteem that they are never questioned.**

Please reiterate in your newsletter from time to time the need to **question and push the professionals ~ they too are human and make mistakes.** To say the least, Hannah's specialist and I now see each other differently and I now believe her care is topnotch quality.

I'm also attaching some 'sort of non-professional friendly' documents that help explain GAI and other deficiencies/acidemias that others may find of interest. We searched high and low for information in those critical first few months. With information comes understanding and power. We wish that for many.

I'm also sending this letter to the Organic Acidemia Association, as we receive their newsletter too and hope some of our learnings help them. We again wish you a wonderful holiday season and may 1997 bring health and happiness to you and many, many others. Much love from the Great Northwest,

Amy, Tom, Hannah (now 7) and Emma (now 4) Myers
Issaquah, WA

Hannah's Story ~ GAI

May 1993 ~ Hannah, you are 4 years and 3 months old. And this is a true story about you and what a scare you gave your Dad and me along with so many wonderful people ~ both family and friends. Someday you'll be able to read this. When that time comes, we hope you feel the love, support, and prayers again that came your/our way when all of this craziness happened.

It all started unknowingly, really at your conception. But this story begins on Wednesday evening, April 28, 1993 about 8pm, bedtime. Just before lying down you threw up. You had recently been 'enjoying' lots of salsa and onions, both green and yellow. And this evening with dinner, you had a whole green onion. SO when you vomited, we took it as a reaction to what you ate.

The next day, Thursday, April 29th, you went off to school ~ 'The Rainbow School' ~ in your usual manner. I remember singing "I'm A Little Tea Pot" with you on your way in. When we got to school, I talked to your teacher, Miss Rilla about the fact that you were sick the evening before, but seemed better now and that I would call the school a little later in the morning. Miss Rilla then asked if I was aware that you were falling asleep sitting up in chairs, and that the past two mornings you fell asleep at 10 am and slept through lunch, waking about 2 pm. I told her I hadn't heard that and maybe you were coming down with a bug. I again said I'd call a little later in the morning.

At about 9 am, Miss Rilla called me at work and said that you had just gotten again, and could I please come get you and take you home. I called your Pediatrician, Doctor Neuzil, and made an appointment for you at 2:10 pm. I got to your school about 10 am and found you asleep. We went home to hang out on the couch and wait for our afternoon appointment. You picked out the 'Nutcracker' video and shortly after it started you fell asleep again. But before you fell asleep, you were telling me about several different experiences from your past that really didn't make much sense. Just before we headed to the Dr's office, around 1:40 pm or so, you started to exhibit some very strange behavior while you were sleeping. You seem to be making panting noises and tossed and turned. I then knew you were very sick and was glad we were headed for the Pediatrician's office.

Now this is when it starts to get scary. Arriving at the doctor's office, you were a very limp noodle in my arms. When trying to respond to my voice commands, you tried to open your eyes, and then I noticed your pupils were dilating and closing very strangely. The doctor saw us right away, and after looking at you for probably a second, she said there was something very wrong with our Hannah. She did a few minor reflex tests and then instructed the nurse to call 911. The doctor said we would be transporting you down to Children's Hospital and to start "thinking about what we were dealing with." I said I knew your little stuffed toy puppy dog was in the car and that you'd want it. I ran to get it. When I returned, waiting just minutes for the firemen to arrive, three nurses tried to get your blood pressure. The first two couldn't hear or find one...and that's when I started to

cry. One could hear the sirens from the fire truck, then two firemen showed up. They took a quick look at you and realized it was beyond their control and instructed the nursing staff to call "Medic 1." Then the questions started flying...What had happened??? The only thing I could think of was that on the Sunday evening before, while eating dinner you leaned toward me to tell me a story and kept going and you hit your head on the hardwood floor. So everyone jumped to the conclusion that you were experiencing head trauma.

The "Medic 1" fellows arrived. There were two of them. By this time, several of the nurses, the two firemen, and now one of the "Medic 1" aides had tried about a dozen times to get an IV started in your arms. You were out the whole time, just waking up when a new needle was tried, and you'd scream "HUG ME, HUG ME, HUG ME"... I was crying, nurses were crying, and even one of the firemen could not hold back the tears. You had a whole room of people very, very worried. I called your Dad and let him know we were headed to Children's Hospital in Seattle and that we all suspected head trauma. He said he'd call Emma's caregiver and let her know what we were doing and that he'd meet you and me in the Emergency Room at Children's.

After several tries at getting the IV started, one of the Medics said "Let's Go." I picked you up...yes, you were breathing, but you weren't with us. We ran out of the office and they strapped me with you in my arms in the back of the ambulance. The Medic found a wonderful stuffed bear that he wanted you to have. With the sirens going we were off. We just made a mess of traffic, which delighted the driver, but the Medics were so concerned about you. One started a tube of oxygen that he just held close to your nose. We arrived at Children's Emergency Room around 3:00 pm. and they rushed you to a room. Dad showed up almost immediately and boy was I glad to see him. He held you close while I went to fill out the paperwork. By the time I got back to your room, they had successfully gotten an IV started in your right leg, close to your ankle. You were starting to come around by then.

Within minutes, they wheeled you up to have a "cat scan" of your head. Everyone was sure we'd see something really ugly and not good. But after two complete scans, the doctors let us know that all was well with your head. We sighed thanking God it wasn't a head trauma.

We returned to your room in Emergency awaiting the results of blood tests that were ordered. The questions they began to ask now had to do with digestion. Was there something that you had taken orally that might help explain your state? We couldn't think of a thing. We started talking about how you have always been a bit of a picky eater and it was hard for you to consume any type of meat and you didn't care for milk or dairy much either. The doctors in emergency started putting clues together and then the blood tests started to come back. We then thought there was something wrong with your kidneys or liver.

By this time, your hospital room was ready. It was close to 7:00 pm. and you were tired and hungry. We got to your room and there waited for your dinner ~ pretty non-descript ~

jell-o, bullion and saltine crackers. You ate very little and fell off to sleep. Daddy had to go pick up Emma and handle a few things around home. I slept in a small temporary bed right next to you all night. I think the seriousness of this all really hit me when your night nurse came by and said that she would be coming by every 2 hours and wake you up "So you wouldn't slip away."

We started letting others know, and that's when all the love, concern, support and prayers started flooding in.

Hannah, you were just great! A real hero in our eyes! You took all the needle jabs, tubes and all the other junk that goes with staying in the hospital really well. We were so proud of YOU!

Dr. Ron Scott (a specialist in Metabolic Genetics) came by to see you on Friday. He was the "Main Man," the doctor who would be your doctor and provide the diagnosis that we were looking for. He became very interested in you from the start. He took his pen and drew on your tummy where your liver was and you were so proud of that drawing that you showed several others that came to see you.

Friday was filled from beginning to end. We received phone calls from all over the country. Everybody who had heard about you were very worried about you and sent their love, support, concern and prayers. So many different folks popped in from time to time. Various doctors came by to examine you and give their support. Kelly and Lori Watts came by with a big bag of great things. Kelly and you ate Popsicles and colored. Lori read you "Aladdin." Dad, Lori, Kelly, you and I had a root beer party that ended rather quickly when we had root beer everywhere. You even made the comment at one point during the day, with your bed table covered with saltines crumbs, sipping on your 20th soda pop hanging on to the TV controls with the Disney channel on almost constantly, wanting yet another Popsicle, puzzle pieces and play doh hunks laying all over your bed, your leg propped up on a pillow with your IV hanging on to your right foot..."Mom, this isn't that bad of a place."

You tooted around the hospital in the "cutest" little person wheel chair being pushed by either your Dad or I, pulling along your IV pole. You asked about all the other kids and babies there! It was sort of sad because you were stationed on the cancer/immune deficiency floor. But you quickly made immediate friends with several little girls...getting together at a common table to blow bubbles and basically goof off.

Several different times we would get ammonia level counts on your blood from the nurses. It was continuing to be extremely higher than normal but it was coming down with each report and you were already starting to act like your normal self.

Late Friday afternoon (6ish), Doctor Scott showed up again. He had the news we were anxiously awaiting. After rushing blood and urine tests through his own personal lab within hours (which should have taken 7+ days), he told us that you have "Glutaric Acidemia Type II," a very rare genetic disorder. You received the recessive gene from

both your Dad and me. He told us that it meant that your body did not process or metabolize proteins in the normal manner. When you did consume proteins your body would accumulate toxins that would affect your brain. He instructed us to "listen" to you ~ that the reason that you had made it to the ripe old age of 4 without a previous episode was that you naturally steered away from proteins. That if we looked at the way you had previously eaten and the fact that we had to "battle" with you about drinking your milk, eating a piece of cheese (because we learned long ago that there was no way you wanted anything to do with meat), that it all made sense that you and Mother Nature knew about your physiological needs. I asked Doctor Scott why we didn't overdose you with proteins during your first year of life. He made the comment that breast milk was naturally low in protein (I breastfed you for 9 months) and the formula (SMA) we chose for you also is low in protein.

Dr. Scott instructed us to keep you in the hospital until your ammonia levels were back in the normal range. He also asked that we bring Emma into the hospital the next day (Saturday) and have a urine test done on her to see if she too carried the affected gene.

The Doctor prescribed that you continue to take your multi-vitamin with iron and fluoride, along with 100 milligrams of riboflavin (B-2) and three TUMS (for calcium) daily along with following a very strict vegetarian diet. He said you would be a VEGAN (total) VEGETARIAN all your life ~ and that you should stay away from all meats, fish, fowl, eggs, and dairy products (***be sure to talk with your child's Dr and/or nutritionist about diet ~ opinions and information on diet may differ from what is written here**). We quickly asked about ice cream and milk in your breakfast cereal. And what about eggs baked in things? He said that the Dietitians he would refer us to could instruct us more on questions like these, but that a very small amount of ice cream and watered down milk in cereal would be fine. And the amount of eggs in baked products was minimal.

Dr. Scott explained that for growth and development reasons you did need to get some protein. He instructed 1.5 grams per kilogram (or since you weighed 32 pounds, your target consumption was 22 grams of protein per day ~ no more, no less). So that began the first step of us reading labels and books and understanding what foods had how many protein grams, what vegetables, grains, legumes, seeds and nuts complemented each other to provide the appropriate amino acids.

The Dietitians that work with Dr. Scott said it would be necessary for us to track your food intake for a couple of months so we could see your eating patterns and have it analyzed for protein and calorie content. They said that in a couple of months we could get into the right "nutritional groove" for you and then just be aware of your physical well being.

Dr. Scott also asked your Pediatrician, Dr. Neuzil, if it would be okay for you to come see him every now and then and then he could watch your development, supply us with information (*see below) and most importantly take information he will learn from you regarding this disorder and document it for others in the future who must deal with it.

some sort, there may be periods of low blood sugar, called hypoglycemia. This can be very severe and make your child feel weak, shaky, and dizzy.

GAI is currently treated with a low fat, low protein, high carbohydrate food pattern. Foods with very little fat and protein should be eaten since their bodies lack the enzyme to break these elements down. Foods high in carbohydrates are necessary for calories for energy.

Supplemental Carnitor® and riboflavin are often prescribed because this disorder may eventually lead to a deficiency in these nutrients. Carnitine is an amino acid. Amino acids are the building blocks that join together to form proteins. Because individuals with this disorder must eat very little protein, they don't get much carnitine in their diets. Carnitine is essential for muscle energy production and helps to transport fats to cells in the body to carry out its many chemical reactions. Contact the doctor to see if these treatments are appropriate for your child.

Length of Treatment

GAI does not go away over time. Treatment must continue throughout the life span to maintain health. A doctor and a nutritionist will closely follow your child because glutaric acidemia is a life-long condition that could at times be very severe.

Doctor visits offer a lot of advantages for children with GAI. At these visits developmental, physical, and neurological assessments are available. Family interaction, which is important to development, may be evaluated. The staff needs to know how your child interacts with your family in order to help you guide your child. Their job is to teach you to help your child develop the skill needed to manage their own disorder. The staff can provide the support that you need as a parent and as a family.

During visits to the nutritionist, a food pattern will be created and changed as necessary. Most importantly, these visits offer you an opportunity to ask questions and have them answered. Buying a 3-ring binder with tab dividers is helpful to record information, questions, and food patterns. Here you can collect treatment plans, food lists, recipes, and other information. Designate a section for food records and a place to chart your child's growth and development. Make a list of questions as you think of them so you'll remember them at your next doctor visit.

Nutrition and Dietary Guidelines

Depending on the unique needs of your child, the doctor and nutritionist at your clinic will develop a food pattern appropriate for your child's needs for growth and development. The food pattern is usually one low in fats and protein, and high in carbohydrates. Following is a list of foods high in carbohydrates and a list of foods high in fat and protein, which need to be limited according to your nutritionist's recommendations.

Foods HIGH in Carbohydrates

Cold cereals	Fruits	Croutons	Peaches	Popcorn
Hot cereals	Broccoli	Rolls	Vegetable Juices	Asparagus
Pasta noodles	cauliflower	Pita bread	Lettuce	Carrot
Rice	Apricots	Animal crackers	Squash	Oranges
wheat germ	Cherries	Crackers	Sweet Potatoes	Bananas
Beans, Peas	Fruit Juices	Potatoes	Breads	Berries
Lentils	Mushrooms	Vegetables	English muffins	Pears
Corn	Radishes	Apples	Buns	Onions
Special low protein breads and pasta	Yams	Cabbage	Tortillas	Spinach
	Grapes	Graham crackers		

Foods HIGH in FAT ~ to be Limited

Butter, Margarine, Oils, Most Cheeses, Whole Milk, Whole Milk/Cream Products like ice cream and sour cream, Nuts, Seeds, and their products like peanut butter

Foods HIGH in PROTEIN ~ to be Limited

Meats, Poultry, and Fish, All Cheeses, All forms of Milk and Dairy Products, like ice cream and sour cream, Nuts, Seeds, and peanut butter, dried peas and beans

Because your child will be limiting certain foods, the food pattern may not always have enough vitamins or minerals to meet your growing child's needs. For this reason, a general multivitamin and mineral supplement that includes calcium and iron is essential. Your child's nutritionist will help you to develop a specific food and supplemental pattern to insure that your child will be well nourished.

(*Please be aware that these lists may not be totally appropriate for YOUR child. Be sure to talk with a nutritionist familiar with your particular FOD before making changes with the diet.)

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Taylor Renee Schmid ~ MCAD

Taylor turned one on November 6, 1996 and was diagnosed with MCAD when she was a newborn. When Taylor was 3-days-old, she stopped breathing, had several seizures, and her blood sugar was very low. I had been trying to nurse Taylor since her birth but was not having much luck. The nurse was trying to feed her a bottle of formula when she noticed something was wrong. Taylor was transported to Children's Hospital and was there for a week before we found out she has MCAD.

Before being released from Children's, Taylor was put on Carnitor® 3 times a day and we were told she had to be fed every 3 hours the next 3 months and then I decided to bottle feed. The stress of worrying if she was getting enough to eat and setting my alarm clock for every 3 hours each night was too much for me. Taylor did not wake up to be fed ~ we woke her every 3 hours around the clock. We were even told to put cold wet cloths on her cheeks to wake her to eat! My husband and I started taking turns bottle-feeding her at night. As time went on, it was harder and harder to get Taylor to eat. At one point she was just about refusing the bottle completely.

Our pediatrician decided to hospitalize her as a precaution even though she was not showing any signs of having an 'episode' and her blood sugar was normal. While at the hospital, we explained to the doctors that Taylor didn't want to eat every 3 hours. We were forcing her to eat when she wasn't hungry. We were told to 'relax' the schedule and feed her when she acted hungry but were told not to go more than 5 or 6 hours between feedings at night. Since then we gradually increased the amount of time between feedings at night.

Taylor goes to bed at 10:00 pm and we set our alarm for 7½ hours later ~ 5:30 am. We give her a bottle and put her back to bed. She usually wakes up around 8:30-9:00 am. Our goal is to let her sleep from 10 pm to 8:30 or 9:00am without any feeding in between.

Taylor has been sick twice in the last year with minor colds but did not need to be hospitalized (thank goodness!). Taylor is now a happy, healthy 1-year-old. She is walking and has not shown any side effects from her 'episode' when she was 3-days-old. We continue to give her Carnitor® 3 times a day and follow up with her pediatrician every 3 months. Our concern is still her eating. She seems to be picky about food and we can't help wonder if force-feeding her as an infant has caused this. We would like to hear from other MCAD Families who have a child close to Taylor's age. There seems to be so much conflicting advice on feeding and Carnitor® for MCAD children. It would be nice

to hear how other families deal with illness and the day-to-day stress that comes with MCAD.

Eric and Lori Schmid
Columbus, OH

Update to Taylor's Story ~ MCAD

Taylor is now 18-months-old and doing much better. She is completely off bottles and formula and drinks (whole) milk from a cup (*please note ~ low fat or skim milk is often recommended over the high fat whole milk). She is also doing much better eating. We saw a doctor that specializes in child behavior and he helped us a lot with her eating habits. We also stopped the night feeding in January. Taylor now gets a snack and a cup of milk at bedtime. She sleeps from 9:30 pm until 8:30 or 9am the next morning. She has never shown any signs of an 'episode,' so we now know that she can go 12 hours without eating (*when well). Knowing this has definitely made things easier. We can relax and not worry about her eating every 3 or 4 hours. She eats when she's hungry and has a better appetite than when we were trying to force her to eat every 4 hours. We can finally treat her like a 'normal' child! Of course we get concerned when she is sick, but fortunately she has only had a few minor colds and has not needed to be hospitalized. Thank you for letting us share our story. I think it really helps to know other families are going through this too.

Lori, Eric and Taylor Schmid
Columbus, OH

Truly a Miracle

Rachel Marie Gibson was born on January 8, 1995. After an uncomplicated pregnancy and delivery we were blessed with a 7 lb 5 oz baby girl. Upon arriving home the next day she seemed unusually sleepy: and not very interested in eating. Being our third baby, an alarm went off telling us that something was not quite right. We kept our eye on her and even tried to force a bottle but she just wanted to sleep.

By 4 am we decided that was long enough, and by the time we arrived at the emergency room she was limp like a rag doll. Her body temperature was 96 degrees and she was very lethargic. We were told to wait in the waiting room and, needless to say, we wouldn't stand for that. We called our clinic's after-hours number and got some action. Rachel's doctor admitted her for dehydration and failure to thrive after drawing blood for several tests and a spinal tap. All tests for infection of any kind came back normal, however, her blood sugar was 26!

After a week of stabilizing what we thought was a blood sugar problem, and testing for a few other scary things, we were sent home. We were overjoyed! Things were going fine. Rachel was a very happy and content baby. She remained a poor eater, however, and her slow weight gain was a concern. With weekly weight checks and monitoring how much

she ate we kept on top of things. But when she was 3½ months old she stopped gaining weight and her muscle tone was so low she could not even hold her head up on her own. This prompted her doctor to proceed with some of the 'rare testings,' though none of us ever dreamed something rare would turn up.

However the weekend before her clinic appointment, Rachel started throwing up, and to be safe, she was admitted for an overnight stay in the hospital with IV fluids to prevent dehydration. The specialists told us that if she did have one of the metabolic diseases, dehydration would be very serious.

The next day, April 23, 1995, her breathing became labored and her eyes were glassy. She was transported by ambulance to a town 90 miles away where she was to be further tested. Ten miles from the hospital she quit breathing and had to be resuscitated the rest of the way. We ended up in the emergency room instead of the pediatric unit where they were expecting her. Rachel resumed breathing on her own but stopped again while being examined. She was intubated, placed on a ventilator and sent to the pediatric intensive care unit. Within four days the diagnosis was made ~ LCAD (***many LCADs are now being retested and re-diagnosed as VLCAD, as was Rachel**). Thus began a three and a half month stay to stabilize her.

Rachel failed an extubation, and had to be re-intubated with plans for a tracheostomy and long-term ventilation. She was breathing on her own and we were elated but after 6 hours she got pale and her breathing became labored. Her liver and heart were enlarged from the fatty acids she could not metabolize all her little life. Since her heart was so big and thick, it did not want to pump properly, and the effort it took her lungs to breathe was too much for her. Such a high and low in one day was so mind boggling. It almost seemed like making the diagnosis was the easy part. We had a long road ahead of waiting and prayer. Her tiny frail body fought pneumonia and several infections while battling out her metabolic 'seesaw.'

We finally got to bring Rachel home on August 8, 1995 (after much training!). She was still on a home ventilator 24 hours a day with several other monitors and she required 10 medications around the clock. It took a year for her heart to heal and to wean her off the ventilator.

We are pleased to announce that on August 22, 1996 we took her back to the hospital to have her tracheostomy removed. She is still tube fed but is now learning to eat by mouth. She also has physical therapy once a week and is starting to take her first steps at 20 months. We are so proud of her progress and feel that God has something special planned for her life. She is truly a joy and every second of agony for us was well worth it. Please feel free to call or correspond with us via mail or the Internet if we can be of any help to other families.

Bill and Lisa Gibson
Cedar Park, TX

Questions and Answers

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

Question: Can you explain what a cell's 'mitochondria' is and what they do? Also, what does carnitine have to do with mitochondria?

Answer: Mitochondria are structures that exist inside every cell of our body except for red blood cells. There are from one to several hundred mitochondria in EACH cell and each mitochondria contains the complex protein molecules necessary to carry out energy making chemical reactions. Like a furnace that uses coal to create heat to warm our homes, mitochondria act as 'powerhouses' for the body by using food to create the energy molecules (ATP) [taken from Mitochondrial Disorders Foundation of America newsletter, June 18, 1996] that we use to keep our bodies working properly. Carnitine (*see our website under Pharmaceuticals) helps get the fatty acids from foods into the mitochondria so the fatty acids can then be used as energy. Additional carnitine is also helpful at taking out the toxins or wastes from the mitochondria so there is no extra buildup of fat. If there is a severe buildup, the mitochondria are not able to do its job and a metabolic crisis will likely occur.

Question: If my FOD child eventually marries an FOD carrier, or a non-carrier, what are the chances of their children having the FOD or being a carrier?

Answer:

- 1. Marry a carrier:** Each child would have a 50% chance of having the FOD and 50% chance of being a carrier
- 2. Marry a non-carrier:** 100% chance of being a carrier and 0% chance of having the FOD **It is suggested that the spouse be tested for FOD status/carrier state with an acyl carnitine profile just to be sure he/she is not affected by one of the many possible mutations of any of the FODs.** (* in 2000, there are some adults just now being diagnosed AFTER their own children were diagnosed either through NBS or through diagnostic testing).

Pharmaceutical Update

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

Medical Update

GAI Patients Needed For Research

Dr. Charles Roe, at the Institute of Metabolic Disease at Baylor University Medical Center in Dallas, is interested in obtaining skin cells of GAI patients for an investigation of alternate pathway activity in modifying the severity of the disease. This is strictly for research and there is no charge for testing. Contact Dr. Roe at: 214-820-4533
Fax 214-820-4853

Emergency Room Protocol for FOD Individuals

Dr. Roe is presently trying to propose some changes in Emergency Room Protocol across the country. There have been **too many children that have died in ERs because a Glucose IV was not given to the child in time.** Mostly because it is not part of the usual ER protocol. If a **child comes into the emergency room lethargic** (very sleepy, unresponsive) **and/or with a low glucose (sugar) level, it is vital that a glucose IV be given immediately** in order to prevent hypoglycemia (low blood sugar) and to restore consciousness more rapidly. **Having one child die because Glucose IV was not given or not given in time is one child too many!**

Resources

March of Dimes Resource Center: They can answer questions about genetics and related topics. 888-MODIMES, 914-997-4763(fax) E-mail: resourcecenter@modimes.org

NORD Rare Disease Database: Information on 1,085 disorders and in-house articles on 2,700 rare disorders; 203-746-6518 203-746-6481(fax) www.rarediseases.org; orphan@rarediseases.org

Online Self-Help Resources: The Future of the Self-Help Group Movement

By Loretta Klamik

Online computer systems have been bringing people with similar experiences together to share needed support and practical information. Several servers, such as America Online (AOL), Prodigy, and CompuServe offer online self-help groups. Different servers host different groups, so if a person is looking for a certain group, they should ask that server if any such group meets on their server.

The most economical of the online services are the hundreds of Bulletin Board Systems (BBSs) across the country that may be called for free almost 24 hours a day. These are often run out of individual's homes.

Self-help support networks on the Internet take two primary forms: The first are the "mailing lists" or listservs that allow members to receive messages delivered to their e-mail address and send out messages and responses that can go to all who are on the subscription list. A comprehensive listing of medical mailing lists and other resources is the "Medical List" by Dr. Gary Malet and Lee Hancock (to access The Medical List from its FTP location "ftp2.cc.ukans.edupub/hmatrix." Login anonymous. Password is your e-mail address. The "Medlist" file will be labeled by date of release).

The second way people share their experiences, strengths and hopes is through the USENET network on the Internet. Usenet provides access to thousands of news groups. A newsgroup stores messages on a computer in a central location, which can be read and replied to by users. Online groups and the Internet are especially helpful to consumers who need information beyond what is available in their local communities. The Internet and Online services are a wonderful source for self-help groups and professionals alike. They supply services such as information and resources, which can be easily accessed from one's personal home computer. If you would like more information about these and other online self-help group resources, please call the Self-Help Network at 1-800-445-0116. American Self-Help Clearinghouse Web Address: <http://www.cmhc.com/selfhelp>

Love Messages

(Please see our most current online issue)

*'May you find the strength of the eagle's wings,
the faith and the courage to fly the new heights,
and the wisdom to guide you there'*

American Indian Blessing

Kids Korner

Hey Kids & Parents: Is there anyone out there who would like to submit anything for this space? Maybe a favorite snack for other kids to try? How about a short story telling an advantage to being on a 'special diet?' Kari Farrell's Grandma would make sure to have something just for Kari so she could have a dessert too. It was different from the others' but it was hers only. This made Kari dislike her disorder a little less ~ for a while anyway. How about a picture and a little bit about yourself? Maybe some of the older kids have their own LOGO ideas? Give it some thought ~ send whatever you come up with to Deb Gould by December 19, 1997. THANKS!

Donations Received

The FOD Family Support Group would like to thank: Steve Bruski and Liz Pease for their generous donation in memory of Caitlin. We greatly appreciate donations to help with postage and copying costs. Please be aware that donations are not tax-deductible at this time since we are not officially a non-profit organization. Checks can be made out to Deb Lee Gould and please note on the check that it is for the FOD Family Support Group. THANKS!

Logo Ideas

Here are the ideas we have received so far.
Are there any others out there??



(Deb just added the outside info to identify our group)

"Angel's Wings wrapped around our children's hearts"

From
Linda Johnson
Coventry, RI



All 3
From
Eric G. Schmid
Columbus, OH

From
Judy Farrell
Menasha, WI



FOD Family Support Group



"All In This Together"

Reminders

Book suggestion from Steve Bruski: **Don't Take My Grief Away** by Doug Manning Harper & Row (1984); Short and conversational style, very readable and useful

WANTED!! FOD Kids ~ Please send your questions, stories and experiences with having an FOD. We'd love to hear from you!!

Professionals ~ Let us know what you are involved with as far as your FOD research and services.

Families ~ Keep your stories coming. We all benefit from reading about all your struggles as well as the successes!

Logo Ideas ~ We have some ideas, but I'm sure there are more out there! Send your ideas and articles by December 1, 1997.

Example of Newborn Screening Letter to Advocate FOD Testing

Date

Dear

I am the Director of a **National Family Support Group** for families coping with **rare genetic Fatty Oxidation Disorders (FOD)**. With many of these diseases, children cannot break down fats for energy because an important enzyme is either missing or not working correctly. Death can occur when the disease goes undiagnosed and untreated. Many of these disorders are **often misdiagnosed as Sudden Infant Death or Reye's Syndrome**.

My letter to you is prompted by a **very personal experience ~ the sudden death of my 21-month-old daughter, Kristen, on July 21, 1985, from an undiagnosed metabolic deficiency called MCAD, one of the many Fatty Oxidation Disorders**. Our second child was diagnosed at birth after we read about MCAD during his pregnancy and our third child is a carrier. To say the least, my family has been forever changed by this disease. It took doctors one year **after** her death to correctly diagnose our daughter's disorder after **initially misdiagnosing her with Reye's Syndrome** ~ a diagnosis we just did not accept! It was a year spent in 'hell' from not knowing why she died.

I strongly believe that **if all Fatty Oxidation Disorders were tested as part of the ROUTINE NEWBORN SCREENING for infants in our country, our daughter would still be alive today. Yet, because her disorder was not diagnosed AT BIRTH,**

Kristen went from a thriving, energetic, and beautiful little girl to a STARK DEATH STATISTIC! And our family is not alone.

My letter is to alert you that **CHILDREN ARE SUFFERING AND DYING NEEDLESSLY ~ WITH YOUR HELP, THAT CAN CHANGE!** I am asking you to **consider bringing this issue to your State Health and Medical committees and policymakers in order to change the law and PROMOTE THE INCLUSION OF FATTY OXIDATION DISORDERS IN THE ROUTINE NEWBORN SCREENING TESTING.**

Metabolic disorders are the second most difficult medical problem to diagnose, according to a recent Gallup organization survey of pediatricians. Undiagnosed disorders have been linked to complications such as cardiomyopathy, developmental disabilities, seizures, and for some, death. Only 52% of the pediatricians were familiar with various metabolic disorders, and of those, only 43% knew that blood tests were necessary to confirm a diagnosis. **Using the 'PKU blood spot card,' infants can now be tested for 38 different metabolic disorders for \$3-\$20 per test. Don't you think that SAVING THE LIFE OF A CHILD IS WORTH THIS SMALL FEE?**

Fatty Oxidation Disorders meet the 3 Criteria that have been established for metabolic diseases to be tested at birth. **1) High Enough Frequency:** Conditions already tested in many states, such as Cystic Fibrosis, PKU, and Galactosemia, occur in frequencies of 1 in 2000, 1 in 15,000 and 1 in 50,000 respectively, and may vary yearly and from state to state. Some of the FODs, such as MCAD, occur 1 in 10-20,00. **2) Established Newborn Test:** Many of the FODs can now be diagnosed using Tandem Mass Spectrometry on a blood spot. The child's PKU card is often used as the sample. Researchers have also developed prenatal diagnosis methods using amniocytes. However, the Mass Spectrometry offers a diagnosis in a shorter period of time. Because the cost of the necessary equipment may be expensive for EACH state to acquire, our group is suggesting possibly more cost effective Regional Testing Centers instead of individual state centers. **3) Established Treatment:** Many of the FODs are successfully treated with FDA approved medications, as well as diet restrictions and/or supplementations.

As a spokesperson for the FOD Family Support Group, I strongly urge you to be proactive on this issue and consider advocating the inclusion of these disorders for Newborn Screening, as well as appropriating future funding necessary for the cost of equipment and testing. I would be very glad to discuss this vital issue with you at your convenience.

Treatment cannot begin WITHOUT AN ACCURATE DIAGNOSIS! EARLY DIAGNOSIS THROUGH ROUTINE NEWBORN SCREENING OF FATTY OXIDATION DISORDERS would most definitely PREVENT NEEDLESS DEATHS AND IMPROVE THE LIVES OF OUR CHILDREN AND THEIR FAMILIES.

Respectfully,

Deb Lee Gould, MEd
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MCAD Parent and Grief Consultant
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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.]

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