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From the Editor

Start saving for 2012 Conference!

An OAA mom visited the hotel and wrote an excellent description of the Hotel's amenities. You can look on the Hotel's website to see what they offer, as well as our Email List Archives for that summary, if you are on our google Email List. The room rates per night for singles and doubles will be $99, and moves up to $119 for a quad.

I HOPE many of you will be able to come! We will NOT be charging a Registration Fee for anyone at this Conference as we have in the past - you'll just need to fill out the Registration Form though so we'll know you're coming! And we most likely again will offer a Children's Activity Room—however parents will be responsible for having someone (ie., grandparent) watch their child. We will also try to get some volunteers as we did in Atlanta in 2010.

We will pass along more info over the Email List and our website as we get closer - if you look at our site you'll see past Conference videos, Agendas and Pictures. It's a fantastic opportunity to learn more about all of these disorders and to meet other parents in similar situations, as well as a chance to actually listen to and talk with some of our experts in the field! See you in Portland!!

Valerie Fulton (LCHAD) represented the FOD Group at the Feb 27 – Mar 2, 2011 Society for Inherited Metabolic Disorders (SIMD) Conference at the Asilomar Conference Center in Pacific Grove, CA. We shared a booth with OAA’s Director Carol Barton. Her summary and great pictures are on page 5 and 6. I will also be displaying our poster and brochures in Grand Rapids, MI for a NBS event September 24th.

Please also continue to create awareness of FODs with your family, friends, and medical professionals, as well as create your own ways to raise funds, via ‘Family Fundraisers,’ so we can continue to spread the word about FODs via our website, Conferences, speaking at hospitals, and other various ways that allow us to offer all of our services free of charge. Also, when buying online please remember when you use the iGive link on our site, the FOD Group gets a percentage of your sale. We also earn funds by using GoodSearch as a search engine, or using the Donate button on our site or on our facebook Cause page. You can also order your very own embroidered or screenprinted FOD polo shirt, cap, or any other item from the same embroidery company that I purchased our Speaker shirts! They have our logo on file.

Families ~ We welcome ALL new or updated Family Stories and pictures and we encourage Families dealing with the less common FODs (i.e. HMG, GA2, Carnitine Uptake Defect, TFP, CPT 1&2 etc.) to share their experiences. We’re also always looking for more low fat recipes, poems, ‘Silver Linings,’ pictures, and ‘Reach for the Stars’ accomplishments of our kids/families.

Professionals ~ we need to hear from you too! New Medical, Research, Nutritional, Counseling/Coping, etc articles are always appreciated. Thank you to Dr Fran Kendall and Dr Charles Roe for sharing your experiences in this issue!

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

‘We Are All in This Together!’

Take care… DLG
Editorial

Living with a chronic disorder is beyond challenging for many children, adults and their Families. Over the last 20 years I have spoken with many individuals that have voiced frustration and anger to me, among other feelings. It is especially discouraging for those still searching for a firm diagnosis - having to constantly endure being tossed from Dr to Dr because they do not know what to test for, or feel the parent or Adult is over-reacting or actually causing the illness symptom [ie., accused of Munchausen’s], or they ‘look too good’ to have anything wrong or their concerns are dismissed because false or inaccurate/incomplete information was in their medical file and any new professional often refuses to do any further testing based on what’s in the file [ie., doesn’t want to step on toes].

It is maddening for parents and adult patients when professionals and others [even their own familymembers] do NOT LISTEN to them and/or coordinate diagnosis or care. And because many of our adults have gone their entire lives without a diagnosis, they often have MANY complications and some in the field may not want to take that on for various reasons. It’s heart-breaking to hear an adult cry for help over the phone time after time and for some…year after year!

*Of course, this issue does NOT apply to ALL our medical professionals ~ we have some really wonderful ones around the world...just not enough of them!* Yet we do have many in the various specializations that might see our kids/adults based on their presenting symptoms [ie., GI, neuro-muscular, cardiac etc] that have no idea what FODs or mitochondrial disorders are and that they can present in a myriad of complex ways, as well as differing test results ~ there is no one test that can definitively diagnose FOD or mito 100% of the time. And when a dual diagnosis is involved [ie., having both an FOD and mito] test results can be even more difficult to decipher and to determine a diagnosis. So many are left in limbo to suffer, without having any more energy to fight the system.

That is why endeavors such as Dr Mark Korson’s Metabolic Outreach Service is so vital [see info below]! Other parts of the country and world could benefit from following such a model. We NEED more medical professionals to learn about FODs and mito, as well as train them to treat NOT ONLY our children but our ADULTS! Fortunately, we do have some pediatric metabolic Drs that open their practices to adults, but they are ‘few and far between.’ It’s a fact of life that children grow into adults and with expanded NBS taking hold around the world…we will need MORE Drs, RNs, Metabolic Nutritionists, etc to HELP our Families!

**PLEASE spread the word about FODs and/or raise more funds for CLINICAL METABOLIC TRAINING for NEW Professionals!**

Deb Lee Gould, MEd, Director

Metabolic Outreach Service (MOS)

The MOS has been very active. In March, I provided a 2-day set of workshops in Israel about metabolic diseases, originally directed to pediatric neurologists. However, as time went on and the conference was advertised, pediatricians and geneticists and pediatric house-staff also registered. The conference was sponsored by Assaf Harofeh Medical Center outside of Tel Aviv in conjunction with the Israeli Metabolic Society.

This was a very valuable opportunity for me since I believe such learning experiences can be helpful in getting general and subspecialty physicians to think more about the fatty acid oxidation disorders. I wanted to see what kind of response such a conference would elicit. The conference was a success—160 people attended and the evaluations are very positive.

During the conference I delivered the following lectures -

- Metabolic approach to encephalopathy
- Newborn screening
- Metabolic approach to lactic acidemia
- Metabolic approach to stroke
- Psychiatric phenotypes in metabolic disease
- Metabolic approach to neonatal seizures and hypotonia
- Elevated CK: A metabolic clue

The workshops were all case-based, interactive, and highly clinical and practical. The majority included some algorithm or set of specific questions to ask to help work though the challenge posed by the lecture topic. Six of them included discussion of the fatty acid oxidation disorders, their detection, diagnosis, and/or management.

The MOS is a diversified educational program that seeks to increase awareness among medical professionals and trainees, as well as encourage an interest in metabolic medicine among medical students. We provide:

- Educational workshops to general and subspecialty physicians and medical students, in a variety of settings
- Patient-As-Teacher Project, scheduling patient forums in which medical audiences hear patients and/or parents speak about their life experiences
- Metabolic Supervision Service, providing high-quality clinical supervision to medical and nutrition professionals without metabolic training who are taking care of patients with fatty acid oxidation disorders

The FOD Family Support Group is a co-sponsor of the Metabolic Outreach Service.

Mark S. Korson, MD
Tufts University School of Medicine  Metabolic Outreach Service
Alexandra is almost 9 years old. She has LCHAD. She is thriving. She has so much personality in her little body that just JUMPS out at you when you meet her. She is imaginative, playful, creative, energetic, bouncy... well, you get the idea. In almost every way Alex is a very normal 9 year old girl. We are humbled time and time again as she gets to experience so many wonderful things. She is our miracle. God must have some GREAT plan to allow us to have this little fireball in our family.

I (Lynne, her mom) developed HELLPs at 40 weeks. Because she was term they quickly delivered her and began to help me recover. This was a unique situation for this rare condition. Usually it is a "young" mom, first baby, mid-term. I was 32 (not "young"), second baby, and the pregnancy was term. I was recovering and our daughter seemed to be doing great.

When Alexandra was about 36 hours old she had cardio respiratory arrest while in the hospital room with us. She was rushed to a triage area where the NICU team would work to save her life. Eventually we were told she was stable, intubated and tests were being run. She was hooked up to a lot of things. It was scary to even think of trying to hold her.

The next few days were a roller coaster to say the least. Tests came back good. There didn’t seem to be any answers. Concern loomed over us; WHY did this happen? On her one week birthday she had a confirmed DNA diagnosis of LCHAD. The doctors read us what they knew as they researched. These were skilled and seasoned doctors and they had no experience with LCHAD. Now we ALL read all we could and we began to take action. The local metabolic doctor was out of town and another doctor in another state was consulted. They immediately put her on Portagen. She began to get stronger. She was amazing. We were scared.

Those first few months were filled with follow-ups with each of her nine doctors that seemed to say the same thing “we don’t know much, but each of these patients seem to be different. She has extra things to watch for because of her ‘bad event.’” Alex was happy, she was strong. We didn’t get much sleep. We didn’t know if we could breathe, because it always seemed like we were on guard for “something” but we didn’t know what.

By her first birthday you could almost hear the collective sigh of relief by us and her doctors. She was doing so well. She had only one short hospitalization and was developing quite normally. We celebrated BIG time.

During the next year we really learned what it meant to have LCHAD. She spent 40 days in the hospital in a 9 month period of time. We began to be able to see how SHE responded to illnesses. Our relationships with her doctors developed amazingly well. We learned how to communicate with each other and respect each other. While this was a difficult time we all learned a LOT. We kept a hospital bag packed. We built relationships with the nurses at the hospital. I learned how to help the staff be able to meet our needs and make things easier for her.

Alexandra has been doing exceptionally well physically and developmentally. She excels in school. She is actively involved with her friends. She plays hard.

Her normal routine does include six formula bottles (or cups at school) a day, breakfast, lunch, mid-afternoon, dinner, bedtime, and midnight (actually about 1 am) snack. She gets carnitine 2 times a day. Other than this, she eats a normal low-fat heart healthy diet, with emphases on carbohydrates and quicker energy sources on extra busy days and when she seems a bit sluggish. The wonderful thing is that she is able to have a bit of just about anything. It is interesting to see that her tastes lead her to lower fat snacks and other quicker energy foods. Her favorites are probably pasta with red sauce, deli sandwiches (with mustard), fresh fruit (almost any kind), cereal (loves Cheerios and Rice Krispies), fruit snacks, bagels with fat-free cream cheese, fat-free hot dogs, apple juice, and LOLLIPOPS! It is wonderful that she lives in a time like this one. There are so many choices for fat-free cheese and even children’s menu choices that finding good foods for her is not difficult.

We often plan ahead and make sure that we can easily accommodate Alex’s needs to conserve her energy, reload on calories, and provide lots of opportunities to experience life around us. We work on healthy habits. We emphasize hand washing (everyone knows her dad always has hand sanitizer at the ready). We try to balance a busy schedule and make choices that allow her to stay healthy.

It is hard to imagine how things would be without the medical professionals that take care of Alexandra. At first, they calmed us parents down (and still do occasionally), now they are so important to Alex.

She has special relationships with the staff at her pediatrician’s office. She looks forward to giving everyone hugs and often taking little surprises to them. This is truly a special group of people who love Alexandra. This practice had no other patients like Alex. They have chosen to go above and beyond to take care of our daughter.

Her thankful heart shows through as she thanks the professionals who take care of her. This show up so vividly in the hospital. We had to ivPolePal. When she was young I created a paper face and taped it on her IV pole. She is so protective of her IV site that she doesn’t like to move around. I needed to get her up and out of bed. We named our new friend "Watery." Watery helped to change the mood in the entire room. Suddenly it was fun to take Watery with her around the room. Watery became a tradition. As soon as she is settled in the hospital room, she asks for Watery to be put in place. As the staff and visitors come into the room they notice and comment. Many staff remember Alex because of Watery. This is an important point of recognition for Alex. It often helps the staff remember her and her unique diagnosis. As Alex’s dad watched the reaction of people over the years he began to develop the idea that has lead to ivPolePal. These creative decorations that attach to IV poles have been a wonderful product of living with LCHAD and helping our daughter thrive through each challenge she faces. The opportunities we have had to encourage others struggling with medical issues have been a true blessing for our family.
Alexandra shows her compassion for anyone who is sick or in the hospital. Her area of most concern for those she loves centers around their IVs and going home. These are obviously the hardest things she has to deal with.

Our family knows the signs that let us know that an illness may be coming. We watch her appetite and listen to her. Paying attention to her little details help us identify issues as they develop. We work hard to avoid hospitalizations, but she tends to develop hypoglycemia and that is our “point of no return.” She doesn’t even protest too loudly when we actually go to the ER. It seems easier once the decision is finally made, she doesn’t have to “try” so hard. The staff at our hospital does a fantastic job. Many remember her from previous visits (even in the ER). She is still a difficult stick for IV’s and because of the D-10 that we run, it is difficult to keep these from infiltrating (fancy word for stop working). She really does make the best of the situation, especially as she starts to feel better.

School has been our latest “frontier.” We have amazing relationships with the faculty and staff at our school. As our oldest daughter worked her way through school we communicated a lot with the teachers. They were aware of our “unique” family situation and we encouraged communication with them and our daughter (Claire, now 12). As we prepared for Alexandra to start school we worked at communicating before school started and creating a plan.

The principal has become one of Alex’s “heroes.” She has taken responsibility for Alex being in the school. She has patiently educated us on the best way to help the school. She has watched out for her with concern. She keeps track of risk factors in the school to make sure Alex’s health would not be compromised. She occasionally allows her to “hang out” in her office when we need to protect her at critical times, and maybe most importantly she lovingly smiles and encourages her every time Alex seeks her out to hug her. The school nurse, teachers, P.E. coaches and office staff have all joined the cause of learning about Alex. They work to communicate with us all along the way about daily issues, concerns and needs. We are truly blessed that they see the amazing kids she is and help us at this step in Alex’s life.

Each school year seems to offer new adventures. We have learned to use “home bound” to keep her current on school work when she is out of class for extended periods of time. Alex has answered her peer’s questions about drinking her formula. She has endured wearing a mask at school a few times when we felt like it was necessary. While she is unique, she blends into the fabric of her school and class.

It is hard to predict when the enormity of having a child with unique medical needs will overwhelm us. Instead of her FIRST day of Kindergarten, it was her LAST day. The accomplishment of that goal and all the work that had paved the way caught us off guard. Sometimes it is just watching her play with a friend, or even when she fusses with her sister. Most of our days have become “normal.” We seem to like that status. New friends are supportive and amazed to learn that she deals with the challenges she faces.

We have been supported in so many ways. We have wonderful friends and family near us. We have a terrific medical team that works to take the very best care of her they can. Having the virtual community of the FOD support group has added confidence and community where even our closest supporters can’t help us. We have appreciated the shared experiences and stories we have read. The understanding we have gained about similar experiences have helped us prepare and cope with the challenges we are facing and creatively find ways to forge new solutions. The expertise and advice Deb has used to create and maintain the FOD support group is valuable for families like ours.

Currently Alex seems to be free from complications with her heart and liver. Her eyes seem stable for now. All of these are issues we watch closely. We never know what is coming next, but we have worked to learn to be thankful for each day we have had. We are a blessed family with two amazing daughters. They seem to be teaching us even more than we are teaching them. While this may not be the path we would have chosen, we are thankful for what it has made us.

Lynne Salser       salsers@sbcglobal.net

IVpolepals has recently won a national new products competition. Feel free to follow ivpolepals on facebook or at www.ivpolepals.com to follow this new product and our family’s journey.

Ways to Raise FOD Awareness and Funds for the FOD Group!

The past few years we had several Families plan their own FOD Awareness Projects and not only raised awareness, but funds for our Group, just as the Salser Family has done with ivpolepals! We even had Families have friends send donations into the Group in honor of their child’s birthday or baptism.

The Goodman Family/familymembers hosted several Silpada jewelry parties and raised several thousand dollars!

If you don’t feel comfortable doing your own fundraiser/project, you can help raise FOD funds by using Goodsearch as your browser or shop online using the iGive site – a portion of your purchases benefit our Group.

As for raising awareness, sharing your story at a local hospital or teaching hospital during grand rounds would be terrific ~ we NEED more clinical professionals in the field of metabolism and this would be a way of exposing them to the challenges of this exciting field!

We also added our information on facebook in order to raise Funds ~

check us out and join the FOD Family Support Group CAUSE and Group message board!
Jaime’s Story ~ Adult LCHAD

I’m not sure, but I am probably one of the oldest “diagnosed” patients with LCHAD. I have been poked and prodded by researchers across the country for many years. Once I was diagnosed, I became of special interest because I have done so well and don’t have the severe cardiac issues that many LCHAD patients do. Here is my basic story—keep in mind how old I am [51] and where medicine was when I was a child—polio was the most devastating disease at the time and little medical research was being done on “rare” disorders.

I was born in Rock Hill, SC a healthy infant. . .with no "apparent" problems until I was 24 months old when I suffered a life threatening episode where I became comatose and unresponsive. The doctors at that time said it was from an overdose of medication [my whole family was being treated for intestinal parasites following the recent birth of my sister—routine back then]. This was probably my first episode of hypoglycemia. I also was not walking well–weak muscle tone and was wearing leg braces. Following this episode–my parents say I began to "fail to thrive." I had chronic "sore muscles." Some of my fondest childhood memories is of my father massaging my legs, back and arms at night while the family sat and watched TV. Anyway, to say the least, I was a "sickly, weak" child who ate like a "pig!" I had a thing for sweets [It all make sense now—I was metabolizing sugar—not fat]. I also suffered from chronic kidney infections—diagnosed with dip-strips. Even today. . .urinalysis strips that are dipped into urine will recognize myoglobin as red blood cells.

When I was eight I had my first "muscle crisis" at a girl scout beach trip and was hospitalized with a severe kidney infection. At 16, I had my second severe "crisis" after a church car wash--this time my urine was black in color and my next door neighbor who was an anesthesiologist recommended a urologist. It was determined that I had a malformed kidney and ureter and that this was why I had been so sick. The kidney was removed and within two months I had another "crisis." The urologist told me and my parents "there is nothing else wrong with her . . she obviously has a low tolerance for pain."

Thank goodness this insulted my parents and they asked to be referred to a University Hospital. I was referred to Duke Medical University Urology Department. I was immediately diagnosed with an "enzyme" problem and transferred to the care of Dr Stephen Kahler. Dr Kahler was my Dr for almost 10 years.

At this point it is important to say that my health, while important to me, was not what I was focused on. I wanted to be like everyone else--not different. I focused on boys, school, then college, my first job, etc. I met my husband when I was 25.

Under Dr Kahler’s care, I had MANY, MANY, MANY tests. So many that I can honestly tell you that I don’t remember exactly what they did. One of the last things that Dr Kahler did before leaving Duke for Australia was a muscle biopsy that was sent to Dr Richard Kelly at Kennedy Krieger in Maryland where I received the diagnosis of membrane limited LCHAD. [I honestly don’t know what the difference is between membrane limited LCHAD and LCHAD]. I have also been to Children’s Mercy Hospital in Kansas City where studies were done to try and determine why I do not have the same cardiac involvement as most LCHAD patients.

At one time Dr Kahler had me on L-Carnitine, but I was eventually taken off that supplement. I was also on MCT oil for quite some time until I participated in a study at Children’s Hospital of Philadelphia where it was determined that I was not metabolizing the MCT oil–this was done through urine and blood samples. So now, Drs have determined that I do not metabolize long or medium chain fats—they believe that in all likelihood that I do not process short chain fats either.

When I married my husband, he worked as a staff photographer for a local newspaper. However, shortly after we were married he received a promotion to editor and eventually we started moving quite a bit for his career. Because of frequent moves, it has been difficult to find or keep doctors or/ specialists especially ones that had even heard of LCHAD, much less knew anything about the disorder. Plus, I have actually been relatively healthy. I average about one major "crisis" every two to three years that requires hospitalization. I have come to learn that a "crisis" is usually triggered by one or a combination of the following: fasting, physical exertion, and/or physical or emotional stress. So, I try to do the right things, listen to my body, eat a healthy low-fat, but well balanced diet, and RELAX. It doesn’t always work—but again, most of the time I do OK.

When I refer to a "crisis," I am talking about an episode that begins with muscle pain starting in my lower back, then proceeds to the rest of my body. I usually can not move, not because I can’t, but because the pain is so severe. My CPKs run over 50,000 during these episodes so I’m hospitalized and placed on fluid therapy to relieve stress to my kidneys. A crisis is usually accompanied by difficult breathing, tachycardia, and/or arrhythmia.
I have "sick muscles" regularly, when my CPK is probably over 1000. But because these happen frequently, and I don't plan to spend my life in a hospital, I bulk up on high sugar or fructose foods and drink lots of fluids and ride out the pain. I will usually feel back to normal within 24 to 48 hours.

As far as my heart goes, I have a left to right shunt, slight regurgitation, and occasional arrhythmia--nothing really severe.

As far as my eyes and RP. I have not been diagnosed with RP; however, I do have "night blindness" and avoid driving after dusk. According to my parents, I had 4 eye surgeries as a child to correct "crossed eyes"--none of these I remember. I am VERY farsighted with an astigmatism and have to wear glasses.

My husband and I made the very difficult decision not to have or adopt children. Not just because of the genetic risk, but also because we both doubted [even though I hated to admit it then and still do] that I was not capable of handling the "physical" demands of parenthood.

I have "recently" [ie., last 12 months] started to experience extreme fatigue. My general physician and neurologist, who really doesn't know the first thing about LCHAD, have ruled out the normal causes of exhaustion--the only thing remaining would be the normal progression of this disorder. So, a couple of days ago I started doing some on-line research into lipase enzyme supplements--that's when I discovered the FOD Support Group. I have spoken with Deb Gould and she put in contact with a local specialist in the Atlanta area--Dr. Fran Kendall. I have [had] an appointment with her on May 4th. I'm really excited--I haven't seen a specialist in quite some time. I'm looking forward to finding out what's new and see what her thoughts are on this new symptom.

Anyway, I hope I haven't exhausted all of you with all this BLAH, BLAH, BLAH. I look forward to keeping in touch and will be thinking about you and your families.

Jaime
51, Membrane-Limited LCHAD [Juvenile On-set]
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Emma and Sophia’s Story ~
Canadian MCAD

We are the Nagel family and our story started almost 9 years ago. Our daughter Emma was born after many years of trying and several miscarriages. She was a healthy 7lbs 13oz girl, who ate constantly! She nursed at least every 2 hours for her first year and always ate like a horse. She was always thin and tall. I thought she was taking after my husband who is also thin and tall, so we weren’t alarmed. She got a stomach bug when she was about 18 months old. I remember taking her to the doctor because she wouldn’t stop throwing up and was very lethargic. They rushed her to the children’s hospital because they thought she was severely dehydrated. Thankfully somebody thought to put glucose in her IV. I was told she wasn’t producing ketones and that was odd, but after a 2 week stay in the hospital on IV, she was doing better. Upon discharge the doctor told us they weren’t sure why she was so dehydrated, but that we should always monitor her when she got sick. Fortunately we didn’t have to deal with that for a few more years. Emma never slept through the night. She was always hungry, even in the middle of the night. My husband or I would wake up every night to her being hungry. Because she was so thin, I fed her, because she would cry until she ate. Little did I know that was saving her life.

When Emma was almost 5, we welcomed a long awaited baby girl. All Emma ever wanted was a baby sister. Sophia was born weighing 6lbs 4oz. Because I had a c-section with her, I was required to stay in the hospital for 3 days. The night before we were supposed to go home, the nurse noticed she wasn’t keeping her body temperature up. They took her to the nursery to put her under the heating lamps. I remember calling my husband in the middle of the night crying because I was worried. Both he and the nurses assured me everything was going to be fine. When she came back to me a few hours later, she wouldn’t wake up to feed. The nurses said she was sleepy and to let her sleep. The next morning, she wasn’t eating well, nor was she having any wet diapers. I thought she looked quite jaundice and called the nurse. She agreed and called the pediatrician. He said she looked fine and would eat better at home, and discharged us. The last words he said to me was that I was an overprotective, overbearing mother ~ little did I know that listening to them and not going with my gut feeling that something was wrong, would haunt me forever.
A half hour later while picking up Emma at her grandma’s house, would be the last breath that our precious Sophia would ever take. My husband did CPR when we realized she wasn’t breathing and the ambulance came and worked on her for what seemed like forever. I remember getting to the hospital thinking she would have to spend a little time in the NICU and then come home. After an hour and half we received the news that she wouldn’t be coming home. She was gone to Heaven. It was the most heart-breaking thing any parent should ever have to go through. Two weeks later we received the news that Sophia had suffered a metabolic crisis and liver failure caused by MCADD. We did as much research as we could about MCADD and found that they had started doing the NBS for it in Alberta, where we live, only a month before Sophia had died. Unfortunately, we did not receive the news from the NBS in time.

We felt our older daughter should be tested, but trying to get a clean urine sample from a 5 year old is not easy. After 12 tries in the lab, the genetic clinic decided that if she was to get ill and end up in the hospital, she should have it done, if not, when the geneticist came back from maternity leave she could order the blood test.

Six months after Sophia died on Christmas Day, Emma fell violently ill at dinner that night. She had a high fever and was throwing up. We took her to the ER and they cooled her down and said she had bronchitis, and sent us home. She seemed to make some improvements and was keeping things down. Two days later she woke up at 4:00 in the morning and had thrown up. We cleaned her up and gave her some juice and put her back to bed. When I woke up at 8:00 in the morning, Emma was unresponsive. She was in a coma. After rushing her to the hospital, I told the doctor about MCADD and the test. He immediately called the children’s hospital and asked them what to do. They had to transfer him to the genetic clinic as they had never heard of it! He treated Emma and then transferred her to the children’s hospital. The doctor’s there still weren’t convinced that she had MCADD. They said she would have shown signs by the time she was 5. Emma turned out to have a pretty severe blood infection that required IV meds several time a day. We decided to go to our local hospital to have the treatments as we live about an hour and a half from the children’s hospital, and the driving was going to be a lot. I’ll never forget the look on the doctor’s face when he saw us coming into the ER for her meds. He came over and sat with us and told me he had been trying to get a hold of me that day. That was when he told me that Emma had MCADD. I believe that I knew something more was wrong in the big picture. I’ll always be forever grateful to him.

Almost 2 years ago, we were blessed once again with the birth of our son Cameron. He was born 6 weeks early and I remember the NICU being so good and following all the protocols given to them by the genetic clinic. He does not have MCADD, he is a carrier. He has some other issues, but we were very prepared for him to have MCADD. It is amazing how they treated as he had MCADD until it was proved otherwise, instead of the other way around.

People tell me all the time that Sophia saved Emma’s life. But I think of it as if there was a NBS test when Emma was born, Sophia would still be with us here today. Even though MCADD is a part of our everyday life, I try not to let it run our life, although, some days that is easier said, than done!

Dana and Richard Nagel – Alberta, Canada
Emma – MCADD 09/26/02
Our Angel in Heaven Sophia – MCADD 05/24/07 – 05/27/07
Cameron – Carrier 09/08/09
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Emma and Sophia...cont’d

FOD Awareness ~ Booth Display

SIMD Conference Summary
Since Deb was unable to attend, I was able to represent our FOD Group at the Society of Inherited Metabolic Diseases Conference February 28 to March 3 in Pacific Grove, California. I want to thank Deb and all of you for the opportunity to represent you. I had an awesome time. Previously I had attended several years ago for just one day. Being able to stay for the entire conference this time was much more rewarding. First, just being at the beautiful Asilomar Conference Center with many of the rustic buildings designed by the architect Julia Morgan in the early 20th Century was amazing. And the setting across from the Pacific Ocean on Monterey Bay could not have been better.
The SIMD Conference was 2½ days and 2 full days of presentations by world-renowned specialists in the inherited metabolic disease field. The talks were divided into groups of approximately the same theme. These groups consisted of 4 to 15 different seminars that were 10 to 20 minutes in length. These focused on updates to ongoing research and/or new discoveries/research. I attended all of the lectures that I felt were pertinent to our group.

The main goal of our FOD presence at the conference was to represent our group and share brochures. Our FOD Group shared a table with Carol Barton representing the Organic Acidemia Association in a room reserved for poster board research presentations, vendors, and parent support groups. Whenever this room was open to the medical professionals we greeted them and informed them of the importance our groups provided to our families and the medical community. The table was draped with our FOD banner and bracelets, newsletter and web page copies. I also brought Adam’s web page in binder form [adamsichad.com].

The doctors and other medical professionals were wonderful. They stopped at our table and browsed through our materials often taking our literature to add to their files. I was able to meet and renew acquaintances with many of my heroes and experts in the FOD field. These included Dr Melanie Gillingham and Dr Cary Harding, who will help support our upcoming FOD/OAA Conference next summer, Dr Arnold Straus who discovered the LCHAD gene, Dr Jerry Vockley, Dr Charles Stanley, Dr Neil Buist [he was first to diagnose Adam with LCHAD in 1991], and MANY more. Besides chatting with them at our table, there were many opportunities to visit with them during and after meals. It is very important that the medical professionals know about our group and the services we provide to families, especially the new ones who just had their child diagnosed with an FOD.

Although one of us was usually at the table, Carol and I did take turns attending the lectures we felt were pertinent to our members. I won’t go into detail about these but will just list the lectures I attended. You can access the complete papers at your medical libraries around the country. In Northern California, I always go to the Lane Library at Stanford University. Or, you can ask your health care provider to access a copy of the research paper for you.

1. Hypoglycemia in children: Interactions between the endocrine system, fatty acid oxidation and ketone bodies (SCHAD, CACT, CPT-1, and HLMG-CoA synthase deficiency) Charles Stanley, MD Children’s Hospital of Philadelphia, PA

2. Challenges in the diagnosis and management of VLCAD and trifunctional protein deficiencies Cary Harding, MD Oregon Health and Science University, Portland, Oregon

3. Short chain acyl-CoA dehydrogenase deficiency and other rare disorders of metabolism with elevated C4-carnitine detected by tandem mass spectroscopy newborn screening Dwight Koeberi, MD, PhD Children’s Hospital of Philadelphia, PA

4. Substrate oxidation and cardiac workload during exercise in long chain fatty acid oxidation disorders Annie Behrend, MS, RD [My son was in this study and he was pictured in the slides. I had to restrain myself from shouting out “That’s my son!”]

5. Long chain acyl-CoA dehydrogenase deficiency: A new inborn error of metabolism manifesting as congenital surfactant deficiency Kristen Suhriel, MD Children’s Hospital of Pittsburgh, Pittsburg, PA

6. The NIH Undiagnosed Diseases Program William A. Gahl, MD, PhD, Past-President, SIMD Clinical Director, NHGRI/Director, NIH Undiagnosed Diseases Program

7. Why are there no proven therapies for genetic mitochondrial diseases? Peter Stacpoole, PhD, MD University of Florida, Gainesville, FL

I will only comment on #6. I know many of you are frustrated knowing your child has an FOD but not knowing which one. In the NIH program that looks at undiagnosed diseases approximately 400 out of 1700 presented are accepted for review. Of these sometimes they are able to determine from gene elimination what the defective genes are. If a missing enzyme/chemical, etc. can be identified; possibly it can be replaced to reduce the severe symptoms of the disease. Before this can be done in humans, it must be tested on animal subjects [usually a mouse]. Then the drug/enzyme/additive/treatment can be tried on the mouse after one can be developed that has the same defect as the patient. I know this takes a lot of time. Dr. Strauss, for example, took many years to develop an LCHADD mouse for testing.

Then if the treatment works on the mouse model, the FDA must approve the clinical trials for humans. Then they will only approve of the treatment after much review and when the treatment is positively proven safe. But, what if the patient will die without the treatment? Isn’t it better in that case to try the treatment [I ask myself]?

Dr. Gahl said the public views must change to allow terminally ill patients to be given possibly unsafe treatments if it may save their lives. Otherwise they may go out of the country to have these treatments, some of which are used routinely with success, rather than wait for years for the FDA to decide what to do.

In summary it was amazing to be in an environment surrounded by medical professionals who knew what LCHADD was and not have to explain it. However, having had to explain it for 20 years, the description just flows off my tongue without even thinking about it...It was as good as being in a roomful of candy, knowing whichever one I chose, it would still be sweet. Is that a good analogy? Anyway, I hope to see many of you at the FOD/OAA Conference next summer, our first. Valerie Fulton, mom to Adam [LCHAD] Related articles: ‘Harnessing inner retinal cells...’ ‘Sharing with Congress...’
Bridging the Gap between Mito and Autism
Fran D Kendall, MD

[Presentation at the AutismOne - Generation Rescue Conference in Chicago –
video on http://www.virtualmdpractice.com]

Many recent studies have linked autism spectrum disorder (ASD) to poor mitochondrial functioning. Understanding mitochondrial disorders and evaluating their symptoms may help ASD families determine whether further testing may be important for their child.

What are the mitochondria?
Housed within our body’s cells, the mitochondria create energy, known as ATP. In lay terms, the mitochondria act as our body’s cellular power plants, busily converting the food we eat into the fuel we need to function. The mitochondria are long, cylindrical shaped organelles (parts of cells) composed of an inner and outer membrane. It is within the inner membrane that we find the well-oiled mitochondrial machinery that produces energy. Each cell contains many of the “power plants” needed to keep our internal engines running.

What is mitochondrial disease?
Some people are born with changes in their mitochondria, or sustain injury to the mitochondrial system through other mechanisms, either of which can result in decreased energy production and the onset of disease. Mitochondrial disorders are a group of diseases that alter the body’s ability to adequately convert food into the energy needed for bodily functions.

These diseases, which affect up to 1 in 4000 individuals, can result in widespread clinical problems. These include vision and hearing loss, seizures, low muscle tone, muscle weakness, migraines, chronic fatigue, developmental delays, autism (ASD or autistic features), kidney and liver disease, diabetes and other endocrine problems, and alterations in blood pressure, heart rate and temperature regulation. Affected individuals can have some or many of these symptoms and problems. Often, but not always, the symptoms of mitochondrial disorders progressively worsen over time, particularly when individuals are subject to stressors such as illness or surgery. Although some forms of mitochondrial disease only affect one person in an extended family, most types are inherited, creating a greater impact on families at large.

The mitochondria create ATP through a complex series of biochemical reactions in the electron transport chain. The electron transport chain, also known as the respiratory chain, is composed of five complexes (Complex I-V) or groups of chemicals whose sole purpose is to create energy from the breakdown products of food using phosphate and oxygen. There are hundreds of different genes (37 inherited from the mother in the form of the mitochondrial DNA and over 850 inherited from both parents in the nuclear DNA) that encode for various proteins that ultimately come together like jigsaw puzzle pieces to create energy. Mitochondrial disorders alter one or more of these genes and proteins, resulting in decreased or ineffective energy production and subsequent malfunctioning of the body’s energy-producing processes.

Poor mitochondrial functioning has been linked to the onset of many other disease processes, including Alzheimer’s disease, Parkinson’s disease, schizophrenia and bipolar disorder. Some medications, such as HIV antiviral drugs, are also known to affect the mitochondria, resulting in poor energy production and mitochondrial disease symptoms. This secondary mitochondrial dysfunction is due to the mitochondria becoming “sick” or “toxic” due to changes in the cells.

How is mitochondrial disease diagnosed?
Some patients present with a collection of clinical features and findings that enable them to be diagnosed by a comprehensive history, examination and minimal testing such as blood lactate level and brain MRI. For example, individuals affected by Leigh’s disease (a particularly aggressive form of mitochondrial disease) have very specific brain MRI changes and often have elevated lactate levels. In this case, diagnosis can be made on the basis of clinical and laboratory findings alone.

In other cases, patients present with findings that are clearly seen in several of the commonly described mitochondrial diseases known to be associated with specific gene changes. An example is a patient who presents with elevated lactate levels, stroke-like episodes and progressive problems who is found to have the common tRNA mtDNA 3243 mutation seen with MELAS (mitochondrial encephalomyopathy lactic acidosis and stroke-like episodes). This diagnosis can be confirmed using a simple blood test.

In the past, for most others, definitive diagnosis of a mitochondrial disease required the completion of special studies on a tissue rich in mitochondria. The body tissues that house the most mitochondria are the brain, kidney, liver, heart, and skeletal muscles. Because collection of brain and heart tissue is impractical, and attainment of kidney or liver tissue for analysis is not only very invasive but potentially damaging and dangerous, muscle tissue became the tissue of choice for investigation of mitochondrial disorders. After collection of the muscle tissue, the mitochondria were removed from the tissue and studied using special instruments such as a spectrophotometer. Although laboratory use of these special instruments allowed physicians to interpret whether or not an individual appeared to be making energy at normal levels, the biopsies also always carried a risk of false positives and false negatives.

Recently, non-invasive enzyme tests have been developed that use tissues other than muscle tissue, such as buccal swabs and lymphocytes. Gene testing has also expanded, so that a simple blood draw can provide information on over 700 mitochondrial-related genes. These newer tests open the door to a larger patient population, facilitating widespread access to mitochondrial testing without the risks, cost and invasiveness associated with traditional muscle biopsies.

[Cont’d on pg 10]
BRIDGING THE GAP BETWEEN MITOCHONDRIAL DISEASE AND AUTISM

Although mitochondrial dysfunction has long been linked to neurological conditions, its association with ASD is a topic of more recent interest, research and discussion. ASD, a complex neurobiological disease, currently affects an estimated 1 in 110 individuals. ASD influences individuals’ ability to communicate and relate to others, while predisposing them to rigid routines and repetitive behaviors.

Studies completed by a group in Portugal in 2005 and 2007 (Oliveira et al., 2005; Oliveira et al., 2007) suggested that 4.1% of patients with autism had underlying mitochondrial disease. This analysis would classify mitochondrial disorders as a rare but definable cause of ASD. However, a more recent study in the US published in the Journal of the American Medical Association (JAMA) (Giulivi et al., 2010) suggests a much stronger link between autism and mitochondrial dysfunction, reporting that children with autism are far more likely to have defects in their ability to produce energy than typically developing children. In addition to other signs of mitochondrial impairment, the study discovered widespread reduced mitochondrial enzyme function among the autistic children. Complex I was the site of the most common deficiency, found in 60% of the autistic patients, and occurred five out of six times in combination with Complex V. Other children had problems in Complexes III and IV. Although many questions remain to be answered, the study results point to a stronger link between mitochondrial dysfunction and autism than was previously believed to exist. Importantly, this association was established utilizing a cell population (lymphocytes, a type of white blood cell) that is easily obtainable via blood draw.

Even more recently, a review in Molecular Psychiatry (Rossignol & Frye, 2011) reported findings that suggest that children on the autism spectrum also reside along a spectrum of mitochondrial dysfunctions of varying severity. This article, like the JAMA report, pointed to the need for more research to understand this association. However, the authors also emphasized the need for ASD children to be screened for possible mitochondrial dysfunction, citing improvements in children with ASD and mitochondrial abnormalities after initiation of mitochondrial disease management.

WHICH ASD PATIENTS SHOULD BE EVALUATED FOR MITOCHONDRIAL DISEASE OR OTHER GENETIC DISORDERS?

All ASD patients should undergo a basic genetics workup that includes studies such as Fragile X and chromosome microarray testing. Decisions regarding whether a specific patient requires a more in-depth investigation for mitochondrial or other rare metabolic or genetic diseases should be undertaken by a mitochondrial expert and/or a biochemical geneticist. Such a decision should be based on a number of factors, including screening results, laboratory testing, family history, physical findings, and clinical features. In general, the genetics workup and ongoing management of an ASD patient (should a genetics diagnosis be made) is best completed by someone trained in genetics with mitochondrial and metabolic disease experience and expertise.

WHY IS IT IMPORTANT TO KNOW IF AN ASD PATIENT HAS MITOCHONDRIAL DISEASE?

Most people or families seek a diagnosis for two general reasons. First, a mitochondrial diagnosis can lead to interventions that will improve the life and health of the affected person. Although mitochondrial disease is not yet curable, an affected person’s quality and duration of life can be improved by aggressive metabolic management by a mitochondrial expert. Knowing that a patient has a mitochondrial disorder is also important for ER staff and other healthcare professionals, as certain protocols should be followed to prevent the adverse affects that can occur particularly at times of illness and stress. Secondly, obtaining a clear diagnosis may assist families with future pregnancy planning, as well as providing a basis for determining risks to other family members. In addition, mitochondrial medicine is rapidly changing, with a number of clinical trials under way. Enrollment and participation in ongoing treatment trials and research protocols requires that a patient be definitively diagnosed with a mitochondrial disease.

CONCLUSION

Navigating the road of complex medical problems can be confusing and overwhelming. Many parents and families find themselves alone and sometimes bewildered as they try to determine the best course of action for their loved one. Understanding the facts and options, and what constitutes an appropriate evaluation and workup, can empower families to obtain the best care for their child or loved one and help provide them with the best possible outcome and quality of life. Making use of resources such as foundations, support organizations and chat rooms (particularly to seek opinions about subspecialists being considered for care) can alleviate stress, avoid potential conflicts of interest with providers, and guarantee the best care.

Regards,

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VirtualMedicalPractice, LLC
expanding genetic horizons...
My Experiences and Understanding of VLCAD Deficiency and its Treatment

Charles R. Roe, MD June 26, 2011 (Now retired)

This disorder is characterized by the deficiency of the VLCAD enzyme that is required for converting long-chain fatty acids (either endogenous or from dietary intake) into energy. It is an autosomal recessive disorder that has only been recognized and characterized since 1994 when, for the first time, the VLCAD enzyme was discovered. Prior to that time, during the early 1980’s, a disorder was recognized that was incorrectly called “Long-Chain Acyl-CoA Dehydrogenase” (LCAD) deficiency and subsequently found to be actually a deficiency of the newly discovered VLCAD enzyme. Prior to 1994 many so-called “LCAD” patients that were identified by skin biopsy enzyme assay died during infancy or early childhood with severe cardiac involvement. To my knowledge, there have not been any deaths due to metabolic decompensation and cardiomyopathy of VLCAD patients as adolescents or adults despite subsequent diagnosis of new patients in those age groups.

This difference between affected infants who often died with cardiomyopathy and the apparently less severe form of VLCAD deficiency in older surviving children, adolescents, and adults prompted investigation to explain why there were two very different clinical forms of the disorder. I will refer to these two phenotypes as “VLCAD-C” for the severe infantile form with Cardiomyopathy and “VLCAD-H” for the milder phenotype of older patients whose initial problems in infancy and childhood was Hypoglycemia (low blood sugar). The descriptions of the clinical courses of these two phenotypes is as follows:

“VLCAD-C”: These patients characteristically have episodes of hypoglycemia in the neonatal period that is followed at 3-5 months of age by cardiac failure, and evolution of “hypertrophic or dilated cardiomyopathy” requiring Intensive Care often for 1 or more months. Without an immediate diagnosis and appropriate dietary management, they usually died on that admission. They would also be found to have very high levels of creatine phosphokinase (CPK) signaling involvement of skeletal muscle (Rhabdomyolysis). In summary, this phenotype involves metabolic compromise of heart, liver, and skeletal muscle.

“VLCAD-H”: These patients also have episodes of hypoglycemia in the neonatal period and subsequently during childhood and adolescence when they will have recurrent bouts of muscle pain that may require hospitalizations for a few days to control the rhabdomyolysis. However, they do not have cardiac involvement and do not evolve hypertrophic cardiomyopathy as seen in the more VLCAD-C phenotype.

Most reviews of this deficiency describe the importance of the VLCAD enzyme in the “beta oxidation” spiral in mitochondria but do not address the degradation of fatty acids in other cell bodies such as the peroxisome. The peroxisome and mitochondrial pathways for fatty acid conversion to energy are linked together. The first thing to understand is the structure of fatty acids: dietary fats and stored fats, relevant to this discussion, contain 16-18 carbon atoms (C16 – C18) linked in a chain with the first carbon being the acid group. The oxidation cycle of fatty acids results in the reduction of carbon atoms, two at a time producing from C16 a C14 fat plus the two carbon unit known as “acetyl-CoA”. This cycle is repeated from C14 to C4, each time releasing another acetyl-CoA. When produced in the mitochondrion, acetyl-CoA is utilized by the “Citric acid cycle” finally producing the high-energy compound called ATP. Clearly, when the VLCAD enzyme is deficient, it is not possible to go from C16 to C4 and acetyl-CoA is not available for adequate energy production. This is why a severe energy deficit occurs when a VLCAD patient is fasted or receiving a high fat diet. The peroxisome contains different enzymes for beta oxidation and represents ~ 30% of total cellular fat oxidation, is limited to going from C16 down to C8. The C8 fatty acid is converted into an acylcarnitine, exported from the peroxisome, taken up by the mitochondrion, reactivated and oxidized down to C4 and ketone bodies in the liver. Normal mitochondrial oxidation can go from C16 or C18 down to C4 with the latter producing “ketone bodies” in the liver that can be exported to other organs as a source of acetyl-CoA for energy production from their citric acid cycle. In VLCAD deficiency the ability to produce and export these “ketone bodies” is severely diminished thus causing energy deficits in other organs that depend on them for energy production. This is especially true for skeletal muscle and the heart and results in muscle cell breakdown (rhabdomyolysis) and cardiomyopathy with heart failure during illness in the severe form of VLCAD deficiency.

Can these two phenotypes be recognized biochemically? Why does the milder phenotype of VLCAD deficiency never have heart involvement but has intermittent muscle breakdown (rhabdomyolysis) with illness?

The biochemical distinction between these two phenotypes was demonstrated with special studies of cultured skin cells to which was added stable isotope (non-radioactive) labeled palmitic acid in three publications between 1998 and 2001. [References 1-3]. Palmitic acid is the preferred substrate for the VLCAD enzyme that was deficient in fibroblasts (cultured skin cells) from biopsies of patients representing both “VLCAD-C” and “VLCAD-H”. When these cultured cells were analyzed following 72 hours of incubation with palmitic acid, the metabolites representing the attempted metabolism of this fatty acid could be measured. One could observe the metabolism of palmitic acid (having 16 carbon atoms) down to metabolites having 14, 12, 10, etc. down to 4 carbon atoms. The figure below illustrates the successful 100% recognition and differentiation of the metabolic profiles for “VLCAD-C” and “VLCAD-H”. Skin cells from patients with “VLCAD-H” reveal a ratio of C16 : C12 of ~ 1.0 in contrast to the ratio of C16 : C12 of ~ 4.0 observed with patients with the severe “VLCAD-C” phenotype.

These results suggested that in the milder form of VLCAD, the peroxisome was contributing more to the oxidation C16 and C18 fatty acids than in the severe cardiac form.

[Cont’d on pg 12]
Until recently, this method of identifying which form of VLCAD deficiency was present could be performed only at Duke University, Mayo Clinic, Baylor Medical Center in Dallas and and the Academic Medical Center in Amsterdam. Although some reviews have claimed that this test is “invasive” and takes too long, I have found it to be of enormous help in early management and of excellent prognostic value often providing great relief to parents when the milder form was identified. When a blood (not plasma) acylcarnitine profile detected an affected infant but could not identify the phenotype, the infant was treated with a high MCT containing formula such as Portagen and Carnitine supplementation while awaiting the skin fibroblast analysis to clarify the phenotype.

[For more detailed clinical and therapeutic aspects of these two VLCAD phenotypes, see Reference 4]

The Carnitine Controversy: Despite several reviews in the literature that suggest carnitine supplementation may be dangerous possibly producing heart rhythm problems from long chain acylcarnitines. This concern originated from a single LCHAD patient in Canada who had a rhythm disturbance while receiving carnitine. To my knowledge, there have been no other examples of this in any publications up to the present. In unpublished studies between myself and North Shore hospital, several VLCAD patients in the ICU in crisis were given intravenous carnitine in very high doses while being monitored for heart rhythm without any evidence of rhythm disturbance. In my 30 year experience with carnitine supplementation in all forms of fat oxidation and organic acidurias, Carnitine’s only side effects have included only a bad fishy smell or diarrhea but never any other dangerous effects. The fishy smell is due to bacterial breakdown in the intestine producing a fragment that is normally destroyed by the liver but if that action is reduced, the fragment (trimethylamine) circulates in the blood and may be found in breath, sweat, etc. giving the fishy smell. Both side effects which only occur rarely or due to very high doses can be eliminated by stopping or reducing the dose.

So, what is my rationale for using carnitine supplementation? Most physicians consider it important to provide carnitine only when the blood levels indicate a significant deficiency and do not explain why it might have other important benefits to the patient’s metabolism. My rationale is very different and based on compromised metabolism inside the mitochondrion during illness: In 1981, we documented that carnitine could act as a “scavenger” by combining with short chain fatty acids as “acylcarnitines” and eliminating them in the urine, This was felt to be a good way to remove potentially toxic short chain acids such as propionic acid found in the organic acidurias: propionic acidemia and methylmalonic aciduria. Although this scavenging role occurs with SCAD and MCAD deficiency, it could not eliminate fatty acid metabolites containing more than 12 carbons such as in LCHAD, Trifunctional Protein, VLCAD, Translocase or CPT II deficiencies.

However, carnitine has another important metabolic effect inside the mitochondrion. During illness in VLCAD and other long chain deficiencies, toxic long chain acyl CoA compounds accumulate. An acyl-CoA compound like C16-CoA consumes and sequesters CoA enzymes (CoA) and compromises oxidation of amino acids etc. thus interfering with energy (ATP) production. CoA can not pass through the mitochondrial membrane so that when it is trapped inside as acyl-CoA compounds from defects such as VLCAD, metabolism and energy generation are severely compromised. When carnitine is supplemented during this crisis, the acyl-CoA compounds are converted to non-toxic acylcarnitines and CoA is made available again for mitochondrial metabolism and energy generation. This is the most important contribution of carnitine supplementation for long chain fat defects.

Treatment Considerations: Fat mobilization during illness and why it occurs ......

When a patient with VLCAD and other long-chain deficiencies becomes ill and is unable to eat, there is an energy deficiency that can only be approached by mobilizing body stores of fat, amino acids and carbohydrate in an attempt to correct the energy deficiency. However, this is ineffective because due to VLCAD deficiency. Long chain fats cannot be oxidized to provide needed acetyl-CoA for ATP and ketone generation. All organs are therefore affected.

Since I have always been fascinated by basic science publications that might provide a better understanding of what we observe clinically, I discovered that Dr. Graham Hardie had characterized a mechanism that regulates metabolism either in the direction of “catabolism” (mobilization of body stores for energy production) versus “anabolism” (reversing catabolism and favoring “synthesis”). This is like a “switch” existing in each cell. This switch can be activated by reduced cell levels of cellular ATP and is an enzyme called AMPK-mediated protein kinase (AMPK) resulting in enhanced catabolism in an attempt to relieve the energy deficit. Activated AMPK enhances catabolism of precursors for the Citric Acid Cycle and ATP synthesis while inhibiting the mammalian target of rapamycin (mTOR) that is responsible for activation of synthetic pathways such as protein synthesis and cellular proliferation (reference 6). AMPK activation is known to enhance β-oxidation (for energy) while impairing fatty acid synthetic pathways. AMPK can only be inactivated by treatments designed to increase ATP concentration in the cell. In summary, decreased energy production with activation of AMPK is responsible for mobilization of stored fat, glycogen, and amino acids from body protein creating severe illness in long-chain fat disorders including VLCAD.
When the role of activated AMPK due to energy deficiency is considered, it focuses our attention on the fundamental goal of effectively treating VLCAD and other serious long chain fat disorders. The diet is usually low in long chain fat with enhanced carbohydrate and supplements of essential fatty acids such as linoleic and linolenic acids. We have shown that these essential fats are mainly involved in synthesis of important compounds and are poorly involved in beta oxidation where they might contribute to toxic compounds due to a defect such as VLCAD. In contrast, saturated fats (such as palmitate) and oleate are effectively oxidized by beta oxidation and produce the toxic metabolites that accumulate in VLCAD deficiency and are converted to the acylcarnitines that are diagnostic for this deficiency (C16 and C14:1 respectively) from blood analysis and expanded newborn screening. (Reference 7).

MCT oil is of definite value in the therapy because its medium chain fats do not require VLCAD or other long chain enzymes for beta oxidation. They slip in under the block providing needed energy and ketone bodies for export from the liver to other organs including muscle and heart ! However, they only provide acetyl-CoA for the citric acid cycle and ATP generation. For the citric acid cycle to function optimally, it requires a source of oxaloacetate in addition to acetyl-CoA for maximum ATP production. With VLCAD patients referred to me, I often found that they were receiving MCT supplement as little as a tablespoon (15 grams) ! For effective inactivation of the “switch” 35% of total caloric intake is more reasonable (for a 20 Kg [44 lbs] child this would equal ~ 65-80 grams of MCT per day). During crisis, 10% intravenous glucose as a source of acetyl-CoA is always started. However, sometimes the glucose is not effectively entering the cells and additional insulin is required to “force” the glucose into the cell while also inhibiting fat mobilization. As described above, carnitine supplementation also has a role for normalizing mitochondrial metabolism.

My Experience with VLCAD Patients:

From January 2000 until May 2009, 23 VLCAD patients were managed and investigated under an FDA IND treatment protocol at Baylor University Medical Center. As of August 2010, the age of these patients ranged from 7 to 52 years.

Twelve patients had the VLCAD-C phenotype and 11 had the VLCAD-H phenotype. All 23 patients were diagnosed by direct enzyme assay in fibroblasts, DNA mutation analysis, and identification of the phenotype in cultured fibroblasts as described above (See figure above). Results of the enzyme assay as performed by Dr. Vianey-Saban suggested that VLCAD-C patients had little if any residual activity while those with VLCAD-H had more residual activity. These findings do not allow for a clear distinction of the phenotypes but clearly documented the deficiency. There was no reliable correlation between DNA mutation analyses by Dr. Andersen in Denmark and the 2 phenotypes as was reliably identified by analysis of palmitate oxidation in cultured fibroblasts. Although blood acylcarnitine analysis confirmed VLCAD deficiency, one could not differentiate the phenotypes from those profiles. When I refer to a “milder phenotype”, I am referring to VLCAD-H in which there is no heart involvement but recurrent “rhabdomyolysis” requiring multiple hospitalizations will occur. Parents must pay attention to onset of episodes of muscle pain and if not treated will evolve into frank myoglobinuria. (See use of fructose for early treatment of muscle pain below)

Since 2000, there were only two deaths. One (VLCAD-C) was due to non-compliance with therapy. The other (VLCAD-H) was due to a torn superior vena cava during a routine mediport replacement. All of these patients received a diet containing triheptanoin – an odd carbon number MCT (7 carbon atoms instead of 8 & 10 carbons in normal MCT oil). The metabolism of this compound provides both compounds required by the citric acid cycle (acetyl-CoA, oxaloacetate and produces ketone bodies for other organs that can provide both acetyl-CoA & oxaloacetate to these organs. There were remarkable improvements in these patients and the mortality rate as describe above was only 2 of the 23 patients (7%) compared to 75% of VLCAD patients studied in France receiving conventional MCT oil diets. (See References 8 & 9 for more information. Now that I am retired, I have successfully encouraged Dr. Vockley in Pittsburgh to continue evaluating the utility of triheptanoin.

On occasion, VLCAD patients in our studies who experienced muscle pain before a real crisis, were relieved by taking a dose of Fructose every 4 hours. [unpublished data] This supplement was ~3-4 tablespoons of Fructose powder in ~ 4 ounces of water. Relief was usually noted within one hour of that dose and aborted the attack thus avoiding hospitalization. Fructose is another form of anaplerotic therapy like triheptanoin. (fructose is in most grocery stores where powdered sugar is located.)

Other Observations:

To my knowledge, no deaths due to metabolic deterioration in patients with VLCAD-H have been experienced in my clinical experience or reported by other physician investigators before or after 1994. The prognosis for this phenotype is very good with, currently, no basis for concerns regarding lifespan. Now that physicians dealing with adult patients with recurrent rhabdomyolysis are aware of this disorder as a part of that differential diagnosis, more adult patients are being identified with this phenotype. As described, these patients do not have any cardiac involvement and are successfully managed with dietary therapy. (See Reference 5)

Two of my patients with VLCAD-H are good examples of the clinical course and tolerance for surgical procedures:

A 43 year old woman (DOB 12-29-1966) experienced recurrent rhabdomyolysis since adolescence. She has had two successful pregnancies (cesarian) since 2001 without complications. Since 2003, she has experienced only two episodes of rhabdomyolysis but only one required hospitalization for intravenous therapy. During that episode, she had cardiac evaluation with EKG, Echocardiogram, and 2 weeks on a Holter monitor that excluded any cardiac abnormalities. She continues to be healthy and active as of the date of this report.

A 41 year old woman (DOB: 8/24/1964) experienced recurrent hypoglycemia in childhood and subsequently recurrent rhabdomyolysis without cardiac involvement. She was diagnosed with VLCAD-H at the age of 18 years. She successfully completed three pregnancies (caesarian deliveries), underwent gall bladder surgery for stones, and had had 3 mediport placements prior to her death at age 41 years due to a tear in the superior vena cava during that mediport replacement procedure. Her pregnancies were complicated by repeated bouts of rhabdomyolysis. She was on no diet therapy during her pregnancies.

A 52 year old male: (DOB 9-25-58. 52 years old in 2010). He also experienced recurrent hypoglycemia in childhood and subsequently recurrent rhabdomyolysis without cardiac involvement. He has had only three episodes of rhabdomyolysis since 2002 all with good recovery with intravenous fluid therapy. He is a successful salesman. His surgical history was not available.

[Cont’d on pg 14]
My experience with the more severe VLCAD-C patients has been very good. Of note is that following successful treatment of a cardiomyopathy between 2–5 months of life, only one of my 12 patients had a recurrence of cardiomyopathy in later life. That occurred in a remote location where the treatment protocol was not followed. These patients are plagued by recurrent rhabdomyolysis in later life and are often confused with adult CPT II patients. All in all, they have had a good result and were able to participate in sports especially when taking a dose of fructose prior to exercise.

I apologize for, perhaps, excessive information but from following the literature and the e-mails to the FOD support group, I have been impressed (or depressed) by the confusion regarding VLCAD deficiency and its current management. I have always felt that the more understanding of the disease and reasons for its management are very important for families as well as their physicians. I remain a firm believer that “we are all in this together”!!

My best wishes to all of you. Sincerely, Charles R. Roe, MD

REFERENCES


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URGENT NEED for Medical Professionals

With more Families being identified with an inborn error of metabolism (through expanded newborn screening), our Families will need ongoing Clinical Care from knowledgeable and caring professionals. In addition to our Newborn Screening Advocacy by many of our Families, our Group is hoping to also bring awareness to medical schools and other medical organizations and facilities the need for educating and training new Professionals (physicians, metabolic nutritionists etc) in the field of Medical Genetics and Metabolism to treat our children, as well as our FOD adults. We are also raising funds for Clinical Training. [see our website for the donation box]

Once we raise enough Funds we will be able to offer grants to US Clinical Training institutions.

We NEED your help NOW and in the FUTURE so our children will thrive and grow into adulthood with the best of ongoing care!
ATTENTION FOD FAMILIES ~ FUNDRAISING EFFORT AT ITS BEST!
I will donate 95% of my proceeds to FODSUPPORT toward our RESEARCH FUND!
Ever heard of Silpada Jewelry? No, well let me introduce you to the beautiful line of
fine sterling silver jewelry we have to offer!

All our products are .925 Sterling Silver. What’s that you ask?
.925 is 92.5% pure silver mixed with 7.5% COPPER!
(NOT nickel or brass, which makes many individuals’ skin turn green or black)
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EVERY piece is handcrafted and exclusive in design!
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AND, in July 2011, over 200 new products are revealed!
I will donate 95% of my proceeds to FODSUPPORT toward our RESEARCH FUND!
Then send me an email with your order and your phone number and I will contact you personally for payment information.
Every order is shipped directly to the purchaser for just $4.00!
If you would like to have a catalog party to increase our donations, we can do that too!
Just send me an email and I will mail you the necessary equipment.

LET’S REALLY MAKE THIS HAPPEN! THE OPPORTUNITIES ARE ENDLESS WITH SILPADA!
In 2009 I WAS ABLE TO DONATE OVER $2500!!!
Brenda Goodman (FOD mom)
Independent Representative for Silpada Jewelry
Fine Sterling Silver Jewelry
email: doublebn@aol.com
home: 216.292.5938
website: mysilpada.com/brenda.goodman

Welcome to another FOD/Mito Meet in Denton, Texas! Enjoy a casual gathering of other parents, children and adults living with FOD and mito disorders. We talk about our children online but rarely get to actually meet in person. Make a weekend of it, or drive in for the gathering, but come to Denton on July 23rd!

We will meet at the Yogurt Fusion just off the town square, or if our group is too large for that space, I have an option of a meeting room nearby. Denton is a college town on I-35 with a beautiful town square and lots of funky little shops, many hotels and bed & breakfasts, and easy access to Dallas and Fort Worth. There are music and theater productions over the summer, and I will have a list of events from the universities and the City of Denton soon.

Yogurt Fusion has a selection of fat-free frozen yogurts, plus fresh fruits and toppings to make your own yogurt sundaes. They also have great coffees, coffee drinks, and teas. It’s a lovely place for FOD children because the food is FOD-friendly, kid-friendly, and parent-friendly. Come to Denton and meet the people you’ve been talking to all this time!

We’ll have a great time. Please contact Janet at wordminder@yahoo.com if you have any questions. Time is TBA.

Love Messages

‘Life without love is like a tree without blossoms or fruit’
~ Kahlil Gibran The Vision
The Inborn Errors of Metabolism (IBEM) Collaborative

Metabolic clinicians in the Region 4 Collaborative (Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio, and Wisconsin) and colleagues in six other states (New Jersey, Pennsylvania, New York, Missouri, Oklahoma, and South Dakota) recently learned that they have received a five-year grant from the National Institutes of Health to study long-term outcomes for children affected with conditions identified by newborn blood spot screening. Directed by Principal Investigators Cynthia Cameron and Susan Berry, this grant support provides exciting new opportunities for improving outcomes for children identified by newborn blood spot screening (NBS). All of these states screen newborns using tandem mass spectrometry (MS/MS) to identify rare, serious IBEM including fatty acid oxidation disorders and organic acidemias.

While long-term follow-up is critical for monitoring health outcomes and evaluating the effectiveness of newborn screening, standards of clinical care for screened conditions have never been subjected to evidence-based study. More information about outcomes for these disorders is essential to a better understanding of the natural history of the conditions and development of best practice models for treatment. The grant allows collection of information about the health outcomes, complications, and life progress of persons that have conditions identifiable by NBS.

This grant provides funding to continue work initiated by the Region 4 Genetics Collaborative Priority 2 Project Workgroup that began building an IBEM Information System with support from the Health Resources and Services Administration (HRSA) Maternal Child Health Bureau Genetic Services Branch. More than 300 persons have already agreed to participate in the project and have allowed collection of ongoing information about the rare IBEM affecting an individual in their family.

With the receipt of this grant, additional families who attend clinics guided by participants in this research will be invited to participate. Participation consists of allowing the site investigators and their team to abstract information about the progress and medical needs of the affected person. Information is stored in a secure, password-protected database; identifiable information about individual persons enrolled in the project is available only to the site investigator responsible for the individual enrolled. Privacy is carefully protected; information about individual participants will be examined in aggregate so that participating centers can accrue information about these rare conditions.

Over time, we hope that the data collected in this project will build the foundation for evidence-based medical practice and care for rare disorders ascertained through NBS because they will provide data to support treatment decisions based on larger cohorts of affected children than can be seen by any individual practitioner or specialty center. With the collaboration of multiple centers over time, a dynamic, longitudinal database will have the power to provide a foundation for evidence-based medical practice and care for rare disorders ascertained through newborn screening.

Our concept is that by developing a core series of agreed-upon strategies that examination of the differences in treatment plans may yield evidence about optimal treatment choices. Further, if families agree, participation in the project provides a point of contact for other research activities and a means to facilitate data collection about those related research activities. We hope to extend our efforts to collaboration with other researchers interested in new strategies for improvement in care for affected persons. We hope this research project will help both clinicians and researchers learn more about the real lives and outcomes of people who have these conditions.

For more information, please see http://region4genetics.org/

Or contact:
Michigan Public Health Institute
Cynthia Cameron, Principal Investigator
Director of Systems Reform
cameron@mphi.org

University of Minnesota
Susan Berry, MD, Principal Investigator
Department of Pediatrics
berry002@umn.edu

Dr Susan Berry with IBEM Poster at ICIEM
On April 13, Team Ella hosted its third FAOD Regional Meeting with the Yale Department of Genetics in North Haven, Connecticut. Keynote speaker Dr Cary O Harding of the Oregon Health & Science University School of Medicine gave a fascinating presentation on the latest research and promise of gene therapy for metabolic disorders, including the work being done at OHSU and elsewhere.

Dr Margretta Seashore from Yale joined Dr Harding for an open discussion with the audience, fielding diverse questions from the many families and medical professionals in attendance. The topics ranged from nutritional analyses and activity protocols, to the latest research and diagnostic tools. The discussion was quite intimate at times, as audience members shared their personal experiences and concerns, and sought advice from the speakers.

Lynne Wolfe from the Undiagnosed Disease Program of the National Genome Project and the NIH gave an important presentation on the hard work the department does to identify diseases for those with undiagnosed conditions. Following lunch, the three presenters held another open discussion with the audience that lasted the rest of the day.

Among the more than seventy participants, twenty families living with FAODs and metabolic disorders attended from several states, stretching from New Hampshire to Western Pennsylvania. Many new faces joined familiar old ones, as the FAOD community in the northeast continued to grow. Parents with FAOD infants spoke with families with teens and young adults. Old friends connected and new friendships were created in a span of hours.

Children spent their day together in the children’s room, doing arts and crafts, and playing with their peers. They enjoyed meeting other kids their own ages living with FAODs and had a particularly fun time comparing G-Tubes. The tone of this year’s Regional Meeting was more positive than in years past, owing to the energy and fun that the children brought.

The Regional Meeting got off to a rousing start the night before, at a reception to meet the speakers, where more than fifty friends of people with FAODs welcomed Dr. Harding to the East Coast. Team Ella wishes to thank its volunteers, its expanded list of contributors, and owes the greatest debt of gratitude to Pat Grodski, who once again put forth tremendous effort to make the meeting such a success.

Yesterday ended nine years of the "g-tube" era for us. It is amazing to me to look back and see where we have come from with Stephen. I could not have predicted he would do so well in the beginning, and was filled with uncertainty when we began with metabolic crisis at 8 months old. We worried that our child who had double lung failure, partial heart failure, and spent 9 days on ECMO life support would not have a happy, full life, if he even lived. The literature made TFP look pretty bleak. We spent about 7 years using the g-tube for any illness management. Stephen observed, "Now I am completely human, with no plastic parts." That is being debated by his therapists. We've done several years of therapy to overcome the effects of the crisis and just plain TFP issues. We still fuss over thin tube for at least night time feeds, and much more than that for the first several years. The g-tube kept Stephen out of the hospital, except during times when he was both vomiting and having diarrhea for several days....that only happened twice. Once with Roto virus, and once with food poisoning. During the past 1 1/2 years he has taken cornstarch at night instead of having the pump going, and we have not needed the g-tube for any illness management.

Yesterday, I was amazed to see a very alert kid with great color coming out of surgery to permanently close the stoma. Our anesthesiologist was amazing, along with the surgeon. Stephen observed, "Now I am completely human, with no plastic parts." That is being debated by his three older sisters. We've done several years of therapy to overcome the effects of the crisis and just plain TFP issues. We still fuss over things we hope we can still help (still has gross motor skill/strength deficits, and struggles with anxiety.) But he is starting fourth grade in the "highly capable" class next year, and seems excited about it. He’s smart, funny, handsome, an age appropriate pain in the toosh at times, and destined for greatness as far as I am concerned.

I’m so happy to be at this point. I hesitated to share, because I know what it feels like when you are in ICU with your child day after day, week after week, and you see all these other people leaving because their child is getting better, and yours is not. Ours finally did, but there were some whose journey ended there. Every case is so different. And there are times when we all wonder if life will ever feel normal and good. Ha Ha! I’ll probably feel that way next week! But I hold out hope for every one of you that you can find joy in the journey with your children, and that you may seek and find every great moment there is available for celebration.

Summary Information on this Regional FAOD Meeting:
Email GoTeamElla@AOL.Com
Team Ella includes family and friends of Ella, 6 yrs old with VLCADD, diagnosed through newborn screening
‘Reaching for the Stars’
Adventures and Accomplishments of our FOD Kids

Jordan Dougherty, 18, VLCAD, graduated from Catonsville Sr High this year. She won an award for gaining the most volunteer service hours. Jordan will be attending community college in the fall.

Mom - Dawn Oella, MD
dawnd39@comcast.net

Maximo, 2 yrs old, GA2/MADD, learning to ride his scooter on his 2nd birthday, along with dad!

Argentina

Doug, Teen LCHAD
Wrestler and Champ!

Dad - Scott Schulte
Utah
schulte_scott@yahoo.com

~ NEEDED FOR THE JANUARY 2012 ISSUE ~

Medical Update ~ Please Submit

PROFESSIONAL ABSTRACTS/ARTICLES OF ALL KINDS
(Drs, Nutritionists, Genetic Counselors, Social Workers, etc.)

FAMILY STORIES
&
Pictures for KidsKorner

The ‘Silver Linings’ of FODs ~
All too often we are reminded of the difficulties associated with FODs. Hopefully our Email List support will help us remember the ‘Silver Linings’ to these disorders as well ~

What is your ‘Silver Lining’?

Complied by Kim ~ please send your ‘Silver Linings’ to Kim at garethsmommy@yahoo.com to be included in our January 2012 issue.
I have started an open Facebook group MCAD / FOD Recipe Exchange where we can all get together and post ideas and recipes. To post a recipe just use the "Create Doc" link to the right of the page so you can post unlimited characters. You will need to sign in to Facebook to access the Recipe Exchange page.

Teresa, mom to Slayer Dane, MCAD Oklahoma
osricsmom@gmail.com

I made a batch of fat free (and in our case also sugar free) oatmeal raisin cookies today and they tasted so good that I thought I would send out the recipe. They are soft and taste like Quaker Oatmeal to go that you could also use them for a grab and run breakfast.

**Nearly Fat Free Oatmeal Raisin Cookies**

- 1 cup whole wheat flour
- 1 1/2 cups quick-cooking oats
- 1/2 cup sugar (I used Xylowsweet)
- 1 teaspoon baking soda
- 1/2 cup unsweetened applesauce (I added an extra couple of tablespoons to make the dough come together)
- 1/4 cup honey
- 1 teaspoon vanilla extract
- 1/2 cup raisins

**Directions**

1. Combine the flour, oats, sugar and baking soda (I also added a teaspoon of cinnamon); stir to mix well.
2. Add applesauce, honey, vanilla and raisins; stirring well to mix.
3. Coat a baking sheet with fat free non-stick cooking spray.
4. Drop by rounded tablespoons onto baking sheet 1 1/2 inches apart & flatten slightly with spoon. (I roll them into balls and flatten by hand so they cook evenly.)
5. Bake at 350 degrees for 10 minutes.
6. Cool and enjoy.

**Nutritional Information**

- Total Fat 0.2 g
- Cholesterol 0 mg
- Sugar 6.5 g (lowered with sugar substitute)
- Sodium 32.1 mg
- Carbohydrates 11.1 g
- Protein 0.8 g

Teresa, mom to Slayer Dane, MCAD Oklahoma
osricsmom@gmail.com

I used to have more time for "from scratch" recipes or to find vehicles for delivering the MCT on a regular basis. I haven't had that kind of time with 3 teenage girls going going going, and we've had more episodes of "Sorry, you can't have that" and the ensuing disappointment because Stephen couldn't have whatever it was the girls were eating, and he was bored of the usual substitute.

I finally found another mix that works with MCT to make a palatable cookie. I recently found and tried "Krusteaz Bakery Style Sugar Cookie Mix". The mix is pretty low-fat -.5g for one serving. The usual problem with cookies made with oil and/or low fat is that they are too cake-like on the inside and tough on the outside. These cookies had a really nice texture on the inside—more cookie-like than cake-like, and had a nice flavor. They did not feel gummy in the mouth, and were tender, not tough on the outside.

Here's how we made it work (We're just doing 1/2 of the mix at a time):

- 1/2 of pkgd mix
- 1/4 c. flour (the mix was too sticky to handle without it. Maybe could have used less yogurt.)
- 6 envelopes MCT Pro-Cal
- 2 T Nonfat plain yogurt
- 2 T liquid egg substitute or egg white

Divided the dough into 6 to make large cookies (each delivers 1 pkt MCT Pro-Cal). Drop by spoonfuls and flatten onto cookie sheet sprayed with baking spray. Baked at 350 degrees convection for 10 min (375 regular oven).

Would recommend making 12 cookies instead...these were huge—about 5" cookies. My husband thinks they would be excellent with lemon or orange zest. My kids think they taste like vanilla wafers. The mix was not quite as much hassle as making it all from scratch, but the real value was that the cookies were a huge hit with Stephen and taste better than any MCT sugar cookie I've come up with from scratch. He put a little ff strawberry cream cheese on one for frosting. I'm into the frugal shopping these days, and was able to pick up the mix for $.75, so it very affordable too.

It was so nice this morning to give Stephen a choice for his lunch...."do you want a brownie or a cookie for your MCT snack today?" and know that it's going to get eaten! Hope this helps someone else.

Diane
Stephen, age 8, LCHAD/TFP
3 girls, ages 12, 15, 17, unaffected
dnielsen6@gmail.com
CDC’s National Center on Birth Defects and Developmental Disabilities announces a new Pediatric Genetics Web site, which contains easy-to-read information on genetic disorders, family history, genetic counseling, and newborn screening; a compilation of important data and scientific publications; and an individualized page for health professionals.

CDC is working to provide a research-based site that is user-friendly, up-to-date, and appealing.

Visit the site:
http://www.cdc.gov/ncbddd/pediatricgenetics/

•   •   •

This is an excellent allergy site if you want to read up on allergies.
http://www.allergy-clinic.co.uk/

Caroline, MADD/GA2 and unknown mito
United Kingdom
caroline@tilley46.freeserve.co.uk

I am a parent with a child with MCAD and a professional in Special Education. For good information, I suggest the NICHCY website, the National Dissemination Center for Children with Disabilities. The site provides information and support for families just learning about services as well as detailed information for support all through the special ed system. I directing you to a particular page from NICHCY http://www.nichcy.org/FamiliesAndCommunity/Pages/findinghelp.aspx. Please scroll down ¾of the page and you will see highlighted titles for PDF links - Parent’s Guide to Accessing Programs for Infants, Toddlers, and Preschoolers with Disabilities & Your Child’s Evaluation - will give you a good start on understanding the evaluation and procedures and a good overview of early childhood special education. The site also has links for each early childhood special education and state specific resources and agencies. Early intervention has a good efficacy research for intervening early and supporting families.

Kate         Mom to Shannon, 17 MCAD         Central PA         katemckinnonpsu@gmail.com

**Mito Gene Testing Expands Significantly**
[From Dr Fran Kendall’s VirtualNews Winter 2011 Newsletter]
http://hosted.vresp.com/580812/5ed3f06f5f/1731513173/dd9e93a533/

For about twenty years we have known a lot about the 37 mitochondrial genes or those inherited exclusively through our mothers. The remaining genes involved in energy production are inherited or passed on to us through both parents, and are located in the center of our body cells known as the nucleus, and consist of hundreds of genes. Up until very recently, technology allowed us to look at only a handful of these nuclear genes.

A number of studies suggest that 75% to 90% of mito disease in pediatric patients is due to changes in the nuclear genes inherited through both parents. As such, up until now DNA testing did not allow us to find the gene causing mitochondrial disease in all patients.

All of this is changing. In November, a laboratory on the West Coast rolled out expanded nuclear gene testing by offering the first run to a select few practices around the country, including VMP. This testing looks at up to 700 genes involved in mitochondrial energy production including the genes encoding for the proteins in Complex I, II, III, IV and V, support genes such as OXPHOS complex assembly factors, OXPHOS cofactor synthesis, and mitochondrial genome replication/ transcription factors, including the 86 ribosomal protein genes.

These noninvasive studies are a huge step towards eliminating the need for invasive biopsy testing. This will also negate the ambiguity that can come with those often not-so-clear testing results. Ultimately, identifying the gene cause for each patient’s specific mito disorder will lead to better treatment for and understanding of disease sub-types. Please contact us to determine if you, your family member or other loved one may benefit from this expanded testing.

**MEDomics Headquarters**
426 N. San Gabriel Avenue
Azusa, CA 91702
Telephone: (626) 804-3645          Fax: (626) 529-0907
To contact us for additional information, please send an email to info@medomics.com http://www.medomics.com/
Nutritional Ideas & Parent to Parent Suggestions

[Always check with your specialists before making any changes to your/your child’s diet or supplements]

A Variety of Ideas from Chris :)  
Kraft makes some fat-free cheeses that are pretty good.  
I make almost zero-fat little pizzas with a toaster oven:  
Lightly toast a corn tortilla.  
Put a tablespoon or two of pizza sauce on top, and then Kraft fat-free shredded mozzarella. I like to put a little Kraft fat-free cheddar on too, plus extra oregano and thyme, but I think most kids like less spice.  
Add toppings and toast again until the cheese is melted.  
Slice with a sharp knife to make it easier to eat.  
I like to make Hawaiian pizzas with Hormel all-natural ham lunchmeat sliced up and pineapple. Or else I make veggie ones piled with mushrooms and peppers.  
Ham and turkey lunchmeat are low in fat, but I look for ones without the preservatives.  
You can also find low-fat tostadas that you could microwave with the toppings on instead of toasting, but they have a little more fat. The tostadas are crunchier.  
Beans are usually very low fat. I follow the same procedure above to make bean tostadas. I put a layer of refried beans on, then fat-free cheddar cheese and toast it. Then I add salsa and lettuce and chopped tomatoes and sour cream.  
The nice thing about the toaster oven meals is you/your child can put them together just how you/he likes them.  
If you can eat wheat (which I can’t) bean & cheese burritos can be made very low fat with the Kraft fat-free shredded cheddar.  
You can make a fat-free cheesecake with the fat-free cream cheese and egg whites—and then add strawberries or blueberries or cocoa or pumpkin...without the fat it’s quite healthy. I use crunched up cornflakes & sugar on the bottom, which isn’t very crunchy without the fat to crisp it up, but tastes good.  
Almost all fruits and veggies are very low fat of course.  
There are very low fat rice crackers at the health food store in different flavors that he/you might like. They are thin crunchy wafers. I like to eat the plain ones which are zero fat, with Kraft fat-free cream cheese and jelly on top. I also like the cheese flavored and tamari-seaweed ones.  
Pollock and shrimp are very low fat, but the fat they do have is about half essential fatty acids. I wonder if he would like little salad shrimps with ketchup? They are fun to eat. The little scallops boiled in a little salt water are very low fat too, and are sweet and bite-sized for a 4-year old. Also taste good dipped in ketchup. I buy them in a bag in the freezer section of the grocery store.  
You can make fat-free pudding with skim milk and cornstarch. You can also find it in the store, but I can’t eat it because it all has gluten, so I have to make it from scratch. Jello with fruit in it is fun. I used to make “jigglers” with my kids which is jello with half the water added, set, then cut up into shapes. Add black cherries cut in half or oranges or pears or other fruits when it is soft to make faces and things. Playing with food is fun :) We had plastic popsicle molds too, to make chocolate milk popsicles (using skim milk) or fruit juice popsicles.  
Smoothies are good, made with frozen fruit and juice or skim milk or yogurt, blended in a blender.  
A lot of kids like V-8, and they have new carrot-fruit juice kinds that are really good.  
A lot of baked goods can be made by substituting applesauce for the fat and egg whites for the eggs - like soft pumpkin cookies with raisins and cream cheese icing...mmm...  
The Kraft fat-free cream cheese makes a really nice base for icings instead of butter. Add powdered sugar, a little skim milk, and whichever flavors (cocoa, vanilla, orange zest, etc) you like.

I’m always sneaking vegetables and fruits into ‘bad for you’ foods. For instance finely chopped black cherries added to super lean ground beef (5 g of fat per burger) make the burgers softer and tastier - that’s a Michigan trick I learned. I also add shredded zucchini to cinnamon buns and things like that. Not a big veggie fan myself so I have to sneak them in somehow.  
Salads are really good with applesauce on top instead of salad dressing. I make grits in the microwave all the time. I put raisins and other dried fruits in, with brown sugar and cinnamon, then add the water and microwave for three minutes and let cool - quick and easy. Tastes good with real maple syrup on top too!  
Best Wishes, Chris acbliton@gmail.com

I’ve often heard that avocados are a no-no for MCAD, because unlike most foods, avocados contain some medium chain fat. However, they actually contain only a very small amount of medium chain fat, compared to the amount of long chain fat they contain.  
According to nutritiondata.com, 100g of avocado (less than half a cup, about 1/3 of a cup), contains 14g fat. Of that, only 1 gram is medium chain. The rest of it is all long chain and very long chain. Which is one of the reasons I’ve never given my daughter (LCHAD) avocado...at least nothing more than a taste.  
When you’re on nutritiondata.com, when you look at the screen where it tells you how much of each nutrient there is, at the bottom of each section, you can click on a blue tab which says “more information.” That’s where it breaks down the fat by carbon chain, so you can see how much of each kind of fat is in each type of food.  
Incidentally, California avocados, which are apparently a different variety than Florida avocados, have NO medium chain fats!  
I hope this helps! Warm wishes,  
Taryn Mom to Katie, 5, LCHAD  
Queens, NY tpaladiy@gmail.com

My daughter Katie has had problems of dilated cardiomyopathy/heart failure since she was born. In the last year we went back to MCT oil and other MCT products as her heart condition worsened after having a bout of Swine Flu.  
I think it was a combination of factors that helped to improve her heart, but MCT did seem to make a difference in the long run.  
She experienced terrible Vomiting & Diarrhea and the prescribed MCT oil was causing this from what we could determine. I looked at the make up of our oil and found it had some C14 long chains in it. We changed to a different brand with no C14 oil in it (MCT Gold).  
She tolerates it in mashed potato, added cold to sauces and casseroles, and made in to cookies, cakes and tea breads. Our dietitian has said it is better to give a small amount in the food several times a day and to build up very slowly if you want to increase it.  
Mel 45 and Katie 17 Possible CPT2 or Mito mrowan@seven-peas.co.uk
Q: My son has a g-tube and sometimes we have problems with the area around the tube—does anyone have some tips to help with that?

A: This is what we do for G-tube problems: mix a pasty mixture of baking soda and Maalox (regular)...applied with a q-tip. 2 x day. That helps with the oozy stuff and crusty blood BUT not bleeding. IF you want any info on G-tubes, go to the site, below — these kids are on continuous feeds, so they know all the tips! http://hydranencephaly.com/Care/tipsandtricks.htm

Michelle Michelle Janczewski  janczewski@cox.net

Q: What do some Families do to get their child/self to take riboflavin (my daughter has GA2)?

A 1: Riboflavin is the worst thing I have ever tasted! It makes Ethan throw up. We can stick it in peanut butter and he wont even know it is there and he still throws up :( We get ours microencapsulated from Solace nutrition. A lot more expensive, but so worth it.

Christy Ethan 4 SCAD  Erin 2 SCAD  daisy7843@hotmail.com

A 2: We had great success with it being compounded into little squares sort of like gummi bears- but it was really expensive. Now we have it added to his carnitine along with added flavor to mask the taste. Our insurance only covers riboflavin for our son if it is mixed this way. The only downside is that the mixture has to be refrigerated.

Susan Tipton, TN
Jack, almost 5, carnitine deficiency, unspecified FOD  Samuel, 3, unspecified FOD  5 older unaffected children  rieshtipton@gmail.com

A 3: We cut a 100mg pill in half and crush it in with his Flintstones, DHA and CoQ10. We mix the whole thing with blueberry baby food. Anything he likes will work. Just experiment.

Beth mom to Luke 5 1/2 LCHAD/TFP Northampton PA  folcherb@eastonsd.org

Q: We are having problems in the hospital with staff listening to us! What can we do so I can best advocate for my child?

A: Good for you for advocating for your son! Two departments you should be aware of that can help if the attitude gets stinky: first ask to speak to the floor manager. The nursing floor manager usually can get things done ASAP. When a problem gets to her (or him), her job is to smooth things out so you don't have to take step two. If she doesn't address the concerns, ask to speak to someone in Risk Management. The slick trick here is to call the switchboard and ask them to transfer your call to Risk Management. Tell them what room you are in and they'll come find you. By calling on your own, the offensive person is not prepared to defend themselves when Risk Management asks for their side of the story. Risk Management's job is to avoid lawsuits.

Expect to be educating the staff today as you go. They don't get a lot of experience with complicated cases let alone FODs and Mito. You might want to have the FOD web site available to give them as part of their education.

Personally, I'm praying that you receive a TON of compassionate support from the staff and 'Dr Obnoxious.'

Barb Mom to John, 21, partial CPT2 and partial That's John Syndrome Wife to Steve  swybayjsy@aol.com
Welcome to New Babies!

Introducing Sam my beautiful Grandson ~ he is one month old today (June 29, 2011) and was diagnosed with MCADD following the very important heel prick test. After a wobbly start for both Mum and Sam he is now making very good progress.

Kathy
Grannie to Sam - United Kingdom
wigwom@live.co.uk

Kids Korner

Juan
Teen VLCAD
Argentina

Zach, 13 & Renee, almost 8
GA 2/ MADD
Canada

Maximo
& mom, Paz, celebrating 2nd bday!
GA2/ MADD
Argentina

Connor
2 yrs old
VLCAD
Oklahoma

Please note that we also have an FOD KidsKorner/Adults Gallery and other Pictures on our homepage. To submit a pic please email Deb.
**DONATIONS**  
[since our January 2011 Newsletter]

**Family Donations:** Pamela Haviland in honor of Isabella Pennimpede (FOD) and in memory of Bella’s great grandmother Virginia Troncone. Mimi Lee Hogan and Deb Lee Gould in memory of Cyrus Mayer. Kelly Lusis. Brittany & Patrick Henagan and Diana & Justin Reneau in honor of Evangeline’s (LCHAD) 1st birthday. Paul Rosenasser.

**Tshirts, Bracelets, Ribbons, CafePress, GoodSearch browsing, or iGive shopping:** Jeannie Johnson. Barry Utter.

Thank you to all that have bought products from companies on the Internet that support the iGive and Cafepress.com program of donating a certain percentage to Groups like ours. All of those links are on our homepage, right sidebar boxes.


We greatly appreciate donations to help with daily costs, website fees, supplies, Conference costs, phone calls around the world, rent for the Grief Consult office, and raising funds for FOD Clinical Training and FOD Research and long-term investments.

**US Checks can be made payable to ‘FOD GROUP’ and mailed to:** FOD Group PO Box 54 Okemos, MI 48805

We also have a Secure PayPal link on www.fodsupport.org  
**ALL US donations are tax-deductible.**  
Our Tax ID # is 83-0471342.

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**Reminders**

**Families** - Please send TYPED (preferably in word document) stories etc, by Dec 10, 2011 to Deb. Continue to spread the word about FODs and the need for screening ~ it will SAVE LIVES!

**Professionals** - Please let us know about your research and/or clinical work with FOD Families. Send articles, summaries, etc by Dec 10, 2011 to Deb.

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‘People are like stained-glass windows. They sparkle and shine when the sun is out, but when the darkness sets in, their true beauty is revealed only if there is a light from within.’

~ Elisabeth Kubler-Ross

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**Communicate With Us**

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

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Please **REMOVE** me from your mailing list:  
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Please include ideas for future issues or your questions