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From the Editor

Our Hopes for Good Health & FOD Awareness in 2015!

Now that we are starting a New Year, hopefully in a healthy way, please make a strong effort to share your FOD Stories with your Families, Friends and Professionals and anyone that will listen! Even though many of the FODs have been discovered for 20-30 years, most are still unaware of what they are and how they impact our children and adults and Families. We have had some members speak at teaching hospitals and also several have written school projects to spread awareness ~ we welcome ALL ways of getting our information out into the world!

A major way that we create awareness every two years is through our National Metabolic Conference that we do in conjunction with the Organic Acidemia Association—our next Conference won’t be until July 2016. In the meantime please share that tentative date with others so we can have an even bigger turnout than we did in July 2014 and you can begin saving for an unbelievable learning and networking experience! We are also searching for a Major Sponsor and Host for our next Conference—if a University, Hospital or Clinic is interested in hosting us (possibly in the Midwest) please contact me or Kathy Stagni of the OAA. You can read more about our past Conferences and our Speaker Presentation slides on our site.

This Newsletter issue is jam-packed with Stories and great Resources and Medical Information. If anyone, Family or Professional, would like to contribute to our July 2015 Newsletter please let me know in the next few months ~ our deadline for submissions is June 15, 2015.

As noted above, we had a large turnout for our Conference last July, but it comes at a huge cost for our two Groups. If you are able to DONATE to the FOD Group at any time throughout the year, it would be GREATLY appreciated so we can continue our efforts. We truly appreciate every penny that our members and others donate via cash, buying FOD Awareness items, and doing your own fundraisers for example. Thank you to all our members that have purchased Awareness items or donated throughout the year.

We have a NEW Awareness Item for sale ~ our Royal Blue Baseball cap is lightweight 100% cotton and has our yellow logo Embroidered on the front, the twill cap features a pre-curved visor and five-panel construction and has an adjustable strap with Velcro®-closure for kids and adults ~ I wear mine all over town!

You can find the Baseball cap and all of our FOD Awareness Items here. Also when you shop amazon be sure to bookmark and shop every time from our FOD amazonsmile link ~ we benefit from all of your purchases ALL year round by earning a certain percentage of your total purchase!

So please keep us in mind if you are able to donate anytime throughout 2015 and beyond!

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.

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/adults/families.

Professionals ~ we need to hear from you too! New Medical, Research, Nutritional, Counseling/Coping, etc articles are always appreciated.

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...
Deb Lee Gould, MEd, Director
Dear Everyone ~

We invite all those with an FOD in your Family to join, to provide de-identified medical information to the new FOD Connect Registry to help everyone in the global FOD community ~ patients, families, researchers, clinicians, and pharmaceutical companies ~ to learn more about Fatty Oxidation Disorders. We have had a slow start so PLEASE help us by filling out the form!

The goal is improved diagnosis and medical care, as well as empowerment of patients and families through knowledge, connections, and support.

FOD Adults 18 yrs+ can join on their own ~ if an FOD child is under 18 yrs old or if he/she is over 18, but does not have the ability to answer the questions for themselves, then parents/legal guardians can join. If you have experienced an FOD death, you can join the Registry.

If you have any questions about the FOD Connect Registry, or to opt in and Join, please feel free to contact me with any questions. Help our researchers find answers to help you live your life to the fullest ~ join our FOD Patient Registry and participate in future trials and studies!

In the future, we will be developing disorder specific survey questions so we can learn more about EACH FOD! I will be asking for VOLUNTEERS that can help create New Questions for the Registry ~ contact me if you are INTERESTED!

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Another way to CONNECT with other FOD Families and some Professionals is to join our facebook Group and/or our google Email List. We have over 1450 members on facebook and 1300 members on the google List.

♥ Please be sure you have completed the ‘JOIN OUR GROUP’ form BEFORE you request to join either group. ♥

To help EDUCATE and CREATE AWARENESS please also share our website and brochure with ALL in your Family and your Professional contacts! I often mail extra brochures when I mail out FOD Awareness items [bracelets, magnets, tshirts etc] that members have purchased that can be shared with their medical professionals or friends.

Professionals ~ I can mail a larger # of brochures if you contact me and send your address with the # of brochures you’d like for your office or clinic.

And for those that would like to create FOD Awareness in your own town by having your own fundraiser, PLEASE DO — donations to the FOD Group are tax-deductible! Please be sure to complete the Family Fundraiser form so you are aware that it is your own fundraiser and not one endorsed or solicited by the FOD Group. Contact me if you have any questions!

Make a CHOICE to SHARE your experiences with others ~ it MAY SAVE A LIFE!

~ Deb Lee Gould, MEd      FOD Director

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JULY is FOD Awareness Month and once again we will submit our Banner to USA Today to print in their Charity Spotlight (in print and online). Thank you Keith Widmann (MCAD dad) for updating our Banner.
UP! Story of an SCAD Dragonboat Warrior

Diagnosis: SCADD

In 2003 I started having difficulty walking. All of a sudden my foot started to drag, causing me to trip over curbs. My muscles progressively deteriorated and I could no longer stand on my own. I started using a wheelchair in my mid-twenties. Over the years I saw multiple specialists who were unable to diagnose me. When I turned 30 my geneticist diagnosed me with Short-chain acyl-CoA dehydrogenase (SCAD) deficiency.

I was relieved to finally have a name for my disorder and get proper treatment in hospital during a crisis. There was a lot to learn - i.e. the importance of glucose and a low fat, high carb diet, eating frequently and conserving energy. My family, friends and specialist advised against strenuous activity in order to conserve my energy. They worried that too much physical activity could trigger a crisis. I worried that I would lose more strength if I didn’t exercise. It’s so hard finding the right balance.

After many trips to hospital, I realized that I needed to try something new. I didn’t want to deteriorate and I was envious of my peers who were able to work and have active social lives. My coworker suggested that I try dragon boating and I joined a beginner’s team at the local rowing and paddling club.

Dragon boating 101

One of the biggest challenges was getting in and out of the dragon boat. I was unbalanced and wobbly walking down the ramp and needed two people to guide me down it. Getting into the boat was also daunting and I was afraid of falling into the water.

But after pushing off the dock and making the first stroke, I felt a new sense of freedom. As I lifted and stabbed my paddle into the water, I was exhilarated. All the day-to-day challenges of living with a disability melted away and for the first time in a long time I felt normal. There was also something really special about paddling in unison with twenty other paddlers; we all had to paddle together to move forward.

At the beginning, I ran out of energy very fast. I could only paddle thirty seconds at a time before collapsing in the boat like a wet noodle. I felt terrible about resting while the others were paddling. I came to the second practice armed with bottles of juice mixed with Polycose and discovered that small sips of sugar gave me a little more energy. Eating a small high-carb meal before practice also helped. After practice, when my muscles stopped working, I sipped on a protein veggie smoothie.

After a month I was able to paddle for a minute and developed stronger core muscles. It was getting easier for me to walk and stand for longer periods.

Crisis

Two weeks before my first dragon boat race in 2012, I became very ill and was admitted to hospital for two weeks. I was too weak to swallow and was dependent on a feeding tube. I couldn’t lift myself out of bed and had to be transferred into my wheelchair in a sling. The medical staff were shocked that I was paddling and scolded me for “over-doing it.” They advised me to “accept” my limitations and “be more realistic” when planning my activities. My specialist said it was not possible for me to paddle because I didn’t
have enough energy reserves to draw from.

The dragon boat season was over for me and I was so disappointed in myself for making so much progress and then losing all that I had gained. My teammates also needed to find another paddler to replace me for the dragon boat festival. It was hard to imagine paddling again when I was struggling to swallow food and stand at the bedside. I had to re-learn how to walk yet again.

In early March, my family doctor and I drafted up a rehabilitation plan. The first goal was to improve my balance and strength. I started working with an exercise therapist at the local pool who helped me walk in the pool.

Second Chance

Despite my specialist's warnings, I signed up for dragon boating again. To my disappointment, I could barely sit up in the boat and paddle for more than twenty seconds. My family doctor encouraged me to stick with it.

Since starting the paddling, I have increased my strength and endurance. I can now paddle in sync with the other paddlers and have competed in over a dozen races. I now paddle at the front of the dragonboat and set the pace for the other paddlers. It is still difficult maintaining my endurance but somehow I am able to find strength from within.

At this year’s dragonboat festival my team won a bronze medal!

Every time my paddle hits the water, I am reminded of how far I have come these past couple of years. Paddling has helped me find a source of strength that I didn't know existed. I am not as worried about the future. Another crisis may happen but I know it's possible to recover.

I am also reminded of all the love and support I received from my FOD family. I hope I can give something back to this amazing community by paddling for FOD awareness.

The dragon boat is a great metaphor for the FOD motto: ‘We are All in this Together.’

Paddles up... ...Take it Away!

By: Marilyn, SCADD Dragonboat Warrior

Family Stories

Hunter’s pregnancy was seemingly normal...just a few minor complications. At 30 weeks I was admitted to labor and delivery for having contractions that stopped with a lot of IV fluids. I continued to have contractions on and off after that, but nothing too concerning. At 34 weeks the contractions were getting stronger so they gave me medicine to stop them. At 38 weeks I went into labor and Hunter was born weighing 9lbs. Our hospital stay was short and Hunter was sent home what we thought was a healthy baby boy.

Unbeknownst to us, Hunter’s newborn screening came back slightly elevated but “likely nonspecific. Please send repeat screen.” His repeat NBS came back normal. We were never told this until after our second hospital stay. I’m not sure what I would have done if I did know this then, but it is important for parents to know NBS is important and don’t assume everything is fine if they don’t say anything about them.

When Hunter was 6 months old he came down with a cold one week after getting his shots from the pediatrician. He wasn’t acting like himself and had no interest in eating. We called his pediatrician and he advised us to just continue to try to get some fluids in him and let the cold run its course. The second day of this “cold” Hunter could barely keep his eyes open and couldn’t lift his head up for more than a second. Enough of waiting it out—we rushed Hunter to the ER! After what felt like the longest wait of my life we were taken back to a bed.

Hunter’s Story ~ LCHAD
Hunter...cont’d

As soon as I laid him down the nurse’s eyes lit up—she realized something was wrong. Numerous tests were run and the only thing that came back abnormal was his liver enzymes were elevated. The ER we were at consulted with the University of Maryland and they decided he needed to be transferred there. University of Maryland ran more tests and all they could tell us was that his CK levels were elevated. They came back with the diagnosis (misdiagnosis) of Viral Myositis. I was relieved at the time this was a one-time freak thing that would never happen again...or so they said.

Then Hunter turned 1, went to the pediatrician- everything looked great! He was a happy 12-month-old weighing in the 75% and 95% for height. Hunter got his recommended shots and we were on our way. Exactly one week later we woke him up and he went back to sleep, put him in his car seat and he went back to sleep. When I opened the door to get him out of the car, he looked at me and I knew I had seen that look before. It was happening again, they told me it was a one-time thing and it was happening again! We called the pediatrician ASAP and he had us bring him in. Within a couple minutes of the pediatrician seeing him, he sent us to the ER. We got a room pretty quickly. It was the same nurse as last time (she remembered us) and the work up started. His blood sugar was 39 (normal 80-100) and again his liver enzymes and CK were elevated. We were transferred back to the University of Maryland for more tests; they assured me they would find out why this was happening again. Multiple tests were done (EEG, MRI, liver sonogram etc.) and aside from his MRI showing high lactate everything was normal. Lucky for us this time they consulted with a geneticist. The tests she chose to run were going to get us our answer. We spent 5 days in the hospital and Hunter was back to himself, but the tests weren’t back yet. We were discharged on a Friday afternoon with the suspected diagnosis of “Fatty Acid Oxidation Disorder” which if you’re reading this you probably know that could still mean a million different things. It gave us enough answers to drive us crazy but not enough to be able to do anything. The following Wednesday we got the call from the geneticist... Hunter had LCHAD.

The next day we had our first appointment with the genetics team at University of Maryland and they were great. They explained everything and spent hours answering all my questions. Since Hunter’s diagnosis he’s been given a strict diet he needs to follow, MCT oil we need to work into his diet every day, and instructions to add cornstarch to his night time bottle so he can sleep through the night. With lots of doctors following him closely Hunter is doing amazing, I am excited to watch him grow up and see what he decides to conquer in his future.

Christina Abrams cabrams@harfordpublicsafety.org

Family Stories

Melody’s Story ~ VLCAD

Melody (diagnosed through newborn screening and genetic testing - zero enzyme activity) has been dancing with the Northern California Ballet since the autumn she was three. That first year, she just took one hour-long class per week and didn't participate in any performances or workshops. When given 5 ml of MCT oil on crackers before class, Melody was able to dance the entire hour without problems. A year later, she developed a sudden fever, so I took her to the local ER with protocol letter in hand, after having given her a fever reducer per phone instructions from our geneticist at the time. Upon arrival at the ER, the attending physician brushed off the severity of the situation. I even heard him on the phone with the on-call pediatrician, saying, "This child looks fine. I don't know what the mother's problem is."

Meanwhile, my "fine" child was getting worse by the minute and nobody, including the geneticist by phone, was heeding my insistence that Melody needed to be on D10 IV. Melody's blood pressure kept dropping and she eventually became unresponsive. Three hours after arriving, they finally put her on D10, only to take her off of it five minutes later! When seeing how frighteningly low the blood pressure readings were, the ER nurse said, "I think maybe it's our blood pressure cuff. I'll try a smaller one." New cuff, SAME scary readings. FIVE HOURS after arrival at the ER, they finally decided to admit Melody. The admitting nurse took one look at my unresponsive, dying child, pushed all of the furniture out of the way so that she'd have room for a crash cart, and told me, "This child is not well. I'll talk to you after I attend to her."
I’m a GA 2 Girl

By: Mackenzie Bird with Dancho, my companion dog

For as long as I can remember, my neck has hurt. It aches like a headache, but it’s lower, from the middle of my ears to just below my shoulder blades. It’s not just annoying in my life. It sort of feels like I have a house constantly sitting on me, and it makes me extremely tired all the time. In fact, it’s why I can’t go to regular school.

I went to elementary school through the third grade, but that was the last year I could make it through each school day without collapsing on my desk. After that year, my parents decided to take me out of regular school and began home schooling me so that I could have a modified schedule to give me opportunities at home to rest my neck. Some days I needed to stay in bed for most of the day because I just had no energy. My Aunt Debbie, a licensed teacher, reminds me how I sometimes used to lay out on a towel in the driveway, and she would read to me.

However, after returning to our normal routine, I realized that Melody had lost her former stamina for ballet. After a month of classes where she would have to sit out for periods of rest, Melody’s ballet teacher expressed concern and I made appointments with the geneticist and pediatric cardiologist. The geneticist dismissed my concerns, but the cardiologist recommended starting Melody on daily CoQ10. We also added in a stricter meal and snack routine to ballet days. As soon as we started the CoQ10 and new meal/snack routine, Melody was able to last the entire ballet hour without rest. Since then, she has performed in two seasons of Nutcrackers and a ten-day summer ballet intensive program. The Nutcracker begins with an audition in September and then lasts through months of weekend rehearsals, commencing in over a week of performing the full-length ballet every day, with a weekend of two performances per day. I won’t lie - it’s ROUGH, especially with a theater that prohibits food in the dressing rooms. BUT, manageable. The ballet director is now mostly aware of the nuances of Melody’s disease, and personally fed Melody her MCT and snacks during the summer intensive. Melody truly LOVES ballet. She is an artistic soul with a flair for drawing and a passion for ballet, nature, books, and baking. The excess energy I have to put into making sure she survives through ballet performances is worth the joy it brings to Melody, and therefore, to all around her.

Heather Rae Sprague  [picture— Melody (left, 6, VLCADD) and her little sister Evelyn (right, 5 untested carrier?)]
Paradise, CA
hrbrae@gmail.com
grid goes out in a neighborhood. When my grid goes out, my brain and other organs begin to hibernate. Not a good thing when you’re trying to do school work. But there are some bonuses to my disorder. One is I get to eat lots of candy. And another is when I go to amusement parks, I get to ride in a wheelchair, and you know what that means: lots of going to the front of the line.

My doctors and parents are working hard to find some solutions for my disability. There are some experimental treatments that are not yet approved in the United States, but some GA2 patients in England are benefiting from something called Alpha Hydroxy oil. We are hopeful that the US Food and Drug Administration will someday approve AH oil for use in America. Until that day, I take lots of vitamins, I eat food 6-8 times a day, I have snacks in the middle of the night, and I sleep as much as I can... unless I have to do a writing assignment like this.

[Note from mom, Patty [pattybird555@gmail.com]: We sent off a sample of Mackenzie’s saliva for Courtagen's Mitochondrial DNA testing in late Oct 2014. Dr Boles called to say they found a mutation causing phosphoglycerate kinase- (PKG1). Only 30 people so far have been diagnosed, but I'm sure many more are out there. It took us this long and so many avenues to get to Courtagen. This is added to Mackenzie’s Glutaric Acidemia 2 and Monocarboxylate Transport deficiency. http://ghr.nlm.nih.gov/condition/phosphoglycerate-kinase-deficiency]

[Note from Deb: Several of our members have written research papers and/or articles to not only complete a course requirement, but the information is valuable to create FOD Awareness ~ THANK YOU to both Mackenzie [above Story], Janet and Kelly for sharing with the world!]

Janet D Longmore
University of North Texas
This research will explore the rhetoric used about one specific Facebook user group and its members, by the members of the group, to find how that rhetoric affects members’ feelings of belonging and group identity.

‘How a Facebook Group Creates Feelings of Belonging’

[Anya’s dad, Rodney, has MCAD]

• FOD Awareness Article by Kelly Huber and Deb Lee Gould, MEd ~ ‘The Challenging World of Fatty Oxidation Disorders’ in CoSozo Living magazine June 1, 2014
• FOD Awareness article by Kelly Huber and Deb Lee Gould, MEd ~ ‘Why us? Grappling with the Realities of a Rare Disease Diagnosis’ in CoSozo Living magazine August 1, 2014
• FOD Awareness article by Deb Lee Gould, MEd ~ ‘The Power of Support ~ Creating Worldwide Family Support for Fatty Oxidation Disorders (FODs)’ in CoSozo Living magazine Sept 1, 2014

Mackenzie...cont’d
Long chain fatty acid oxidation disorders (LC-FAOD) are caused by defects in the metabolic pathway that converts stored long chain fatty acids into energy, leading to a deficiency in mitochondrial energy production during times of physiologic stress and fasting. Severe and potentially life threatening clinical manifestations, including rhabdomyolysis, hypoglycemia, hypotonia/weakness, cardiomyopathy and sudden death. We have been conducting new clinical trials on treatment with triheptanoin (C7), a novel energy source for patients with LC-FAOD. We first conducted a retrospective medical record review study, sponsored by a clinical-stage biotechnology company, of data from 20 patients with LC-FAOD who were treated for up to 12.5 years with triheptanoin as part of a compassionate use protocol. Many of these patients were referred to me at the Children’s Hospital of Pittsburgh by Dr. Charles Roe. Clinical outcomes including hospitalization event rates, number of hospitalization days/year, and abnormal laboratory values were determined for specified periods before and after triheptanoin treatment. Other events of interest were documented including rhabdomyolysis, hypoglycemia, and cardiomyopathy.

Not surprisingly, we found that LC-FAOD was associated with frequent complications and hospitalizations. A trend to a lower hospitalization event rate was observed in the period after triheptanoin initiation compared with the before treatment period (1.26 vs 1.94; P=0.1126). Mean hospitalization days/year also decreased in the period after triheptanoin initiation (5.76 vs 17.55 vs; P=0.0242). Events of hypoglycemia were lower by 96% (0.04 vs 0.92; P=0.0091) and related hospitalization days were significantly reduced in the period after treatment (0.18 vs 8.42; P=0.0257). Rhabdomyolysis hospital event rate was similar but the number of hospitalization days/year trended lower in the period after triheptanoin initiation (2.36 vs 5.94; P=0.1224). In conclusion, in this retrospective study, treatment of LC-FAOD with triheptanoin appeared to improve the course of disease by decreasing the incidence of clinical manifestations and should be the focus of prospective investigation. Several patients with acute, life threatening cardiomyopathy improved dramatically with triheptanoin treatment but weren’t included in this study. My lab is now investigating several additional possible new medications to prevent episodes of rhabdomyolysis in FAODs.

Prospective studies of triheptanoin are now underway. One study, funded by the FDA and performed in collaboration with Dr Melanie Gillingham in Oregon, has just been completed and results should be available to review shortly after the first of the year. A second study in patients with symptomatic long chain FAODs, sponsored by the clinical-stage biotechnology company (the manufacturer of triheptanoin) is completing enrollment and results are expected to be announced in the second half of 2015. A larger, definitive study is expected to follow.

Additionally, a burden of illness survey is currently being conducted by Optio Biopharma Solutions, a US based independent market research firm that has extensive experience with rare diseases. The overall goal of the research is to help understand the impact that LC-FAOD has upon patients and their families, including symptoms and treatment as well as caregiver and financial challenges. The anonymous combined feedback from all participants may help develop and refine potential treatment options. The survey is being conducted by an ~1 hour telephone interview with a representative from Optio Biopharma Solutions. Participants will be reimbursed $150 for their time. To participate in this survey go to the following web address: https://www.engagehealth.com/survey/TakeSurvey.aspx?SurveyID=8l01662. Please note the survey enrollment will conclude Jan 16, 2015 and there are a limited number of spots, so please sign up soon.

Dr. Jerry Vockley and his colleagues at the Children’s Hospital of Pittsburgh of UPMC are conducting a 7-week evaluation of safety and biochemical changes of the drug Ravicti™ in MCAD patients. Ravicti™ is currently approved for treatment of urea cycle disorders, but laboratory studies in cells have suggested that Ravicti™ may also increase the amount of MCAD enzyme activity.

To be eligible for this study, you must be 18 years or older and have MCAD deficiency caused by at least one copy of the 985A>G mutation. Patients who have kidney or liver failure, or are pregnant or breastfeeding are not eligible. You must also be able to travel to Pittsburgh on four occasions and will be required to stay overnight in the Clinical Research Center for your first visit. For more information contact the research coordinator, Elizabeth McCracken, MS, CGC at (412)692-5662 or Elizabeth.Mccracken@chp.edu.
International Network for Fatty Acid Oxidation Research and Management

[Note from Deb: Stacie Poole, one of our Families, was our FOD Representative for an important and informative meeting in Austria this past September. This will be an ongoing meeting (next September it’s in Italy) that will hopefully generate and coordinate various projects that will benefit the FOD Community! THANKS Stacie for such a succinct and thorough Summary of this meeting!]

INFORM represents a new dimension in consolidated efforts toward research and the development of standards of care, best practices and rationale behind treatments for our children and ourselves, those of us affected by FODs. INFORM stands for the International Network for Fatty Acid Oxidation Research and Management. The Inaugural Symposium meeting was held this past September in Innsbruck, Austria and attended by over 250 doctors and researchers from all over the world, as well as newborn screening organizations and patient support groups. INFORM also is placing a premium on patient input. (More to follow on this exciting opportunity to provide personal input).

So many times as parents or patients, we assume the doctors have “this all figured out.” In the world of rare diseases, which is the case with FODs, this simply isn’t true. Yes, our doctors know how to keep us safe, how to treat dropping blood sugars, myoglobinuria, and elevated ck levels, but there are times when doctors disagree with how to treat a problem within a disease, an atypical presentation within a particular FOD or a way to conquer truly troublesome issues such as persistent neurological, liver or heart complications. For those of us who have been caught in the crossfire of a debate on diagnostics or treatment options of an atypical FOD presentation, this is painfully true. If you’ve been in an extended hospital stay with doctors scratching their heads as to why your child’s ck levels won’t go down or blood sugars keep dropping as the D10 keeps increasing, you know your doctors don’t have all the answers. If you’ve told your doctor over and over you experience muscle pains daily only to be told the “literature” doesn’t support that finding, you’re not alone.

From all I witnessed, I truly believe, determining research based solutions to these incredibly important issues is at the heart of INFORM. Researchers and doctors posed questions many have asked on our support forum, questions that have begged answers, questions we have personally asked our doctors regarding our child, questions that have often been set aside. Debates regarding SCAD being mild, CPT2 also being triggered by VLCAD genes with delayed diagnostics far beyond birth, and promising research with C7 oil and the reduction of myoglobinuria and countless other complicated heavily scientific topics and angles were dissected. (All agreed one day was much too short and next year, the conference will be 2 ½ days to allow for additional discussion on treatment.) Everyone pushed to think outside the box, escape from research dogma and think critically about information before them as well as how it played out with the actual patient, the human beings they were treating.

Some of the most exciting findings were the following:

1. Patient trials continue and more are coming available. The C7 trials have been very promising. Patients have reported increased qualities of life with reductions in hypoglycemia, myoglobinuria and increased exercise tolerance. Additional areas of general research interest have been in insulin resistance, antioxidant therapy and continued ways to optimize the use of MCT oil, which still proves to be highly effective, especially with cardiac issues.

2. Mice trials, although they can’t be completely compared to the human system (mice have different fatty delivery systems than humans) are showing some incredibly intriguing results. Although they don’t immediately benefit the treatment of human illnesses, these mice allow study of a particular function within an illness. Once researchers crack the “code” that link the findings between the mice system and the human system, it allows them to overlay their findings onto the human disease process (I didn’t expect to be so completely excited about mice. These little, furry creatures are truly cheering us on and helping us uncover so much about the human condition). Specifically to FODs, these trials are proving to be very exciting.

3. A new, 3D mitochondrial cellular model has been developed by Dr. Gerard Vockley. This model has allowed for a much more accurate representation of the mitochondrial process as it relates specifically to FODs. It incorporates more accurate causal relationships and can allow researchers to pinpoint holes in their thought processes more easily. To see it, would make you laugh, as it resembles the world’s most intense rollercoaster. This model verifies the complexity of these diseases and just how intense research is in this particular field of medicine.
4. Studies in cellular stress seem promising. Illness, exercise and general stress clearly seem to all take its toll on people affected with an FOD. Researchers are looking at why. How does oxygen play a role? How do vitamins and minerals and the cellular absorption of these critical elements affect overall symptoms? This also takes a mitochondrial disease slant. If we improve mitochondrial function, can we improve the FOD process? And yes, these are mitochondrial disorders.

5. To be specific with the next area would be difficult. So many comments were made. By far, one of the most exciting things to hear, were questions...questions doctors asked researchers and each other. Questions regarding symptoms or diagnostic paths or secondary issues like neurological involvement in FODs that have been disregarded by so many. It is clear, beyond clear, that as much as they do understand about FODs, there are some components researchers and doctors are striving to understand. This validates so many people’s experiences within the FOD community.

Overall, INFORM members will continue to communicate with each other throughout the year. At the conference in September 2015, researchers and doctors will have the opportunity to present and discuss more regarding patient treatment plans and the patient’s physical experience. They are hoping to learn more about how doctors are treating patients in various parts of the world. What do protocols look like? What are the doctor’s rationales for doses and treatment plans? Although some of these elements are fairly obvious like the use of D10 and high carbohydrate diets, exact illness protocols and diet plans seem to vary. INFORM hopes to set research based standards that benefit the patient, take guesswork out of the equation for the local physician and keep the patient safe, beginning at birth.

Over the next couple months, I will be posing a few questions and asking for your input to prepare and submit to INFORM in preparation for the September 2015 conference. Questions like the following...

1. What clinical trials would you like to see happen? Why?
2. What is the most common treatment hurdle you experience when seeking medical care in an emergency?
3. Do you have an emergency medical protocol? If so, what are its instructions?
4. If you had the ear of the best doctors and researchers in the world on FODs, what are the two most important questions you would want to ask?

What symptoms of your FOD seem common that your physician will not address in you or your child’s treatment?

Your answers will be compiled anonymously and sent to the committee at INFORM throughout the year. This will allow them to use your input when developing their agenda for the next conference. With the addition of another 1 ½ days of conference time, this year’s conference should allow for much more in depth conversation focused on treatment, holes in treatment and struggles people with FODs face in the medical community.

INFORM represents hope. Hope that we and our loved ones have a stronger voice than last year. To sit among so many gifted and dedicated professionals was beyond sacred. The individuals present were passionate, interested and committed to improving medical care for all of us. Although it is a fledgling organization with likely a bit of a learning curve, INFORM is off to a solid start, with promising contributions that should eventually greatly impact the care of all of us impacted by FODs.

(Please feel free to email me if you have specific questions at staciepoole@gmail.com Much of the conference became very scientific in nature which led to the addition of the 1 ½ days, but I can see if anything specific regarding questions was mentioned in my copious notes. The symposium website is http://www.informnetwork2014.org/index.php )

Stacie Poole, FOD representative staciepoole@gmail.com
Reach for the Stars!

Congratulations to **LCHAD Dad, Scott Schulte**, for publishing his 1st book ~ it will be out in March 2015!

*A Wrestling Life: The Inspiring Stories of Dan Gable*
By: Dan Gable and Scott Schulte

You can find Scott’s wonderful writing on amazon, but if you’d like to purchase it please use the [FOD amazonsmile link](#) so we can benefit by receiving a small donation from each sale!

Proud Mom, Sharon Little Allen, shares that her son, Joshua (MCAD, 17), is going to Wofford College in the fall to play golf for the Terriers.

The ‘Silver Linings’ of FODs ~ What is your ‘Silver Lining?’

In case others are interested in pursuing a Service Dog for their child or themselves, **Brenda Goodman** wanted to share her Family’s experience with finding one for her daughter, Kayla, SCADD, Epilepsy, PVS( repaired), PDD-NOS, Mito Disease (unidentified), HONORS STUDENT! ~

Kayla has a service dog ~ He is AMAZING!! Kayla used to have seizures regularly, but as she got older she became better controlled, so we couldn’t have Kain trained for seizure detection b/c she didn’t have them enough! So we got him simply b/c Kayla needed to develop confidence and independence AND BOY HAS SHE!!! Kain is her constant "pal!!" She relies on him for comfort and solitude...snuggling and all the time love!! He is her BEST FRIEND!! He adores her and she him. She takes care of him and controls him in public on her own! She is confident enough to tell people "you can not pet him, he is working" and she tells strangers what he does for her!

We got Kain through WAGS4Kids...here in OHIO! Please contact them for other recommendations...there is no monetary exchange with WAGS just an agreement you will fundraise $8000 for them.

We had a fundraiser in Feb 2014 that raised $4000 and planning another this Feb! It is a lot of work but SO WORTH IT!
Why Mitochondrial Disease Looks Nothing Like Medical Child Abuse

By Christine Cox on 04/16/2014
(Also an attorney & SCAD mom)


As media coverage of the Justina Pelletier case has grown, so has the misconception that mitochondrial disease is closely correlated with medical child abuse and its predecessor diagnosis, Munchausen’s Syndrome by Proxy. On behalf of MitoAction and the mitochondrial disease patients we represent, we are extremely troubled by this development and wish to highlight the obvious differences between mitochondrial disease and medical child abuse.

The plight of Justina Pelletier has riveted the rare disease community for the past fourteen months. Justina, a fifteen-year old Connecticut girl, was placed in the custody of the Massachusetts Department of Children and Families following a diagnostic dispute between doctors at Boston Children’s Hospital and her treating physicians at Tufts University. Justina’s parents were accused of medical child abuse for “overmedicalizing their child” through surgical procedures designed to improve her quality of life. The Pelletiers ultimately lost custody of Justina after physicians at Boston Children’s asserted an alternative explanation for her symptoms, which previously had been diagnosed and successfully treated as mitochondrial disease at Tufts.

Mitochondrial disease is not easily confused with medical child abuse. It is a devastating, multi-system disease that frequently results in death. Patients suffer from a wide variety of symptoms depending on which part of the body has been affected by the disease, which is due to problems with the powerhouses of the cells. Mitochondrial disease also can be degenerative and progressive, especially when high-dose vitamin treatments and other medications are withheld. In contrast, children who are victims of medical child abuse generally get better when separated from their families.

Before her most recent hospitalization, Justina was ice skating, going to school and living a relatively normal life. After fourteen months in the care of the Massachusetts Department of Children and Families, Justina is confined to a wheelchair, is in constant pain, has lost much of her hair and has had a significant resurgence of symptoms. In fact, Justina’s condition has deteriorated so much that Boston Children’s had to start using Justina’s abdominal port again, and the judge ultimately returned her medical care to Tufts. As of this past week, Dr. Mark Korson of Tufts is once again treating Justina for symptoms of mitochondrial disease.

Mitochondrial disease is relatively rare, and several media reports have cited an older statistic that places the prevalence rate at 1 in 8,000. More recent literature shows that the prevalence rate of mitochondrial disease is actually much more frequent at 1 in 4,000, http://www.med.unc.edu/neurology/files/documents/child-teaching-pdf/Mitochondrial%20Review%20DiMaro%2005.pdf, and some physicians assert that mitochondrial disease is as common as 1 in 2,000.

In contrast, medical child abuse and its successor diagnosis, Munchausen Syndrome by Proxy, are exceptionally rare. One study published in the medical literature showed an incidence rate for Munchausen Syndrome by Proxy for children under 16 years of age of 2 per 100,000 in New Zealand. http://www.ncbi.nlm.nih.gov/pubmed/11468037. The authors of this study further noted that this incidence rate was “higher than that reported from other countries.” Another study performed in the U.K. and Ireland showed an incidence rate of 0.5 per 100,000. http://www.ncbi.nlm.nih.gov/pubmed?term=8813872. Thus, an ailing child is at least 50 to 200 times more likely to have mitochondrial disease than to be suffering from medical child abuse at the hands of his or her parents. The speculative linking of mitochondrial disease with Munchausen Syndrome by Proxy is not only false but also is both disingenuous and disrespectful to mitochondrial disease patients who suffer from a dreadful and often fatal disease.

Mitochondrial disease affects multiple bodily systems at once, and therefore can be confusing to doctors who are accustomed to a medical system that compartmentalizes the body into discrete systems. It also is a good diagnosis to consider when symptoms do not fit a clear diagnostic picture, which is why mitochondrial disease is tested so often in cases that ultimately were the result of medical child abuse. However, once a family has a diagnosis of mitochondrial disease from a reputable physician, has documentation of other family members suffering from the disease, and has received successful treatment for mitochondrial disease, it is the duty of the hospital caring for the patient to obtain a complete history and speak with the treating physician who is familiar with the case. Had Boston Children’s taken that simple step, the course of the past fourteen months likely would have been much different for the Pelletier family.

[cont’d on page 13]
The fact remains that physician hubris combined with a low tolerance for assertive parents led to the result in the Justina Pelletier case. Until hospitals, patients and caregivers can find a better way to communicate about the health of a child and can tolerate differences in opinion with more resilience, parents of children with rare diseases will continue to run the risk of medical child abuse accusations.

About The Author/Speaker:

Christine S. Cox, Esq. is the Director of Outreach and Advocacy for MitoAction. Christine has a child with a mitochondrial disorder and first discovered MitoAction three years ago. She has been working with Cristy Balcells and others to establish the MitoAction Advocacy Task Force, and also volunteered at the MitoAction Clinical Conference in Los Angeles in February 2014. Christine has practiced law in Atlanta, GA, for the past 10 years. Since 2010, she has been working on legislation in the Georgia General Assembly that would mandate coverage of medical foods (formulas used for metabolic and GI disorders) by private health insurance companies. This bill is very similar to House Bill 977, the legislation MitoAction has been working on to obtain insurance coverage for the Mito Cocktail in Massachusetts. Having seen firsthand the beneficial impact that MitoAction’s work can have on both a family and an entire community of mitochondrial disease patients, Christine is very excited to be a part of this organization.

~ NEEDED FOR THE JULY 2015 ISSUE ~

Medical Update ~ Please Submit to Deb

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

The ‘Silver Linings’ of FODs ~
What is your ‘Silver Lining?’

FAMILY STORIES &
Pictures for KidsKorner

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.
Welcome to New Babies!

Zachary David was born on December 16 at 5:56 pm, weighing 8lb 15oz and 20".
Little brother to Aaron, 2.5 VLCADD
Zachary is unaffected, praise the Lord! We got his NBS results yesterday morning.
Everyone is home, resting, and healthy for the holidays.
Dave, Dana, Aaron, & Zachary Mattini
Indiana, PA

Kingston Zion Chambers, MCAD
Parents: Anna-Nicole Leckie and Patrick Chambers
8lbs 8oz, 21 inches long
May 31st, 2014
leckieanna@yahoo.com

NBS Update

The rare disease community received an early Christmas present from Washington last week when President Obama signed The Newborn Screening Saves Lives Reauthorization Act of 2014 (H.R.1281).
The law is designed to eliminate critical delays in the newborn screening process.
http://www.raredr.com/articles/President-Signs-Newborn-Screening-Act

Through the Eyes of a Mother
By Kaylee / In Newborn Screening

‘We are very honored to share the story of the Wilkerson family and their battle for improvements in newborn screening. Baby Genes was privileged to perform supplemental newborn screening for the newest Wilkerson family member! Isn’t she darling!’

[Sarah Wilkerson is an FOD mom. Diane Wilkinson of the March of Dimes shared this info with us]

We are incredibly thankful that this option existed for families like mine who know that they are at risk.
-Sarah Wilkerson
This personalized support program was created specifically to help dietitians and their patients/caregivers to save time and make the process easier for getting the Vitaflor products needed.

- 1-888-848-2356

**Living with CPT2**

www.cpt2.me [sign into facebook first]

I am one of the very few people to have been diagnosed with a genetic disorder called Carnitine palmitoyltransferase II Deficiency, or "CPT2 Deficiency."

In between episodes, life with CPT2 is very normal. However, during an episode CPT2 patients experience muscle pain, rhabdomyolysis, myoglobinuria...please read more on my blog!

Gary Shuster adult CPT 2

**Gluten free cookbook**  [http://howcanitbeglutenfree.com/](http://howcanitbeglutenfree.com/)


**Lab that identifies the best quality health and nutritional products** through independent testing

**Suggested by Christyne B**

Rady Children's Hospital announced it will create a **genomics research center** that will focus on helping children suffering with diseases that are difficult to diagnose. The Rady Pediatric Genomics and Systems Medicine Institute will assemble scientists, researchers and clinicians to work on treatments and cures for childhood diseases.

**Book Suggestion for Spouses:** I'm reading a book that I highly recommend. *"Mainstay: for the well spouse of the chronically ill."* I was able to get a copy through inter library loan, as it is no longer in print (I'm told Amazon has used copies). I have found additional support at [Wellspouse.org](http://Wellspouse.org).

An MCAD grandmother that I met at the FOD conference told me about the group. I'm very grateful, so I'm sharing. The group is not for any specific illness, and encompasses a wide variety of life challenges.

Deb Porter  porter.deb@sbcglobal.net
[Note from Tara: I work directly with families all over the world who may wish to make a donation by shipping them a sample collection kit (postage paid both ways) and the necessary paperwork. I am also available to them for any questions they may have regarding the consent and submission documents. I’ve included an updated list of the samples we have - there are still plenty that we don’t have any representation for and even for those diseases that we do, we would welcome more samples. We’ve also got a new webpage describing the sample donation process in more detail and that link is included as well.]

The NIGMS Human Genetic Cell Repository is a biobank that collects blood or tissue samples and clinical information from individuals with inherited genetic diseases like fatty acid oxidation disorders and makes cell lines and DNA for scientists to use in their research. Samples and corresponding clinical information that are donated to the repository are de-identified and made available to qualified researchers all around the world through an online catalog. Having a centralized source of well-characterized cells lines and DNA allows scientists to spend more of their time and funding on studying how cells function, identifying new mutations, and developing ways to diagnose, treat, and possibly prevent fatty acid oxidation disorders.

Our goal is to continue to build our collection of fatty acid oxidation disorders to create a larger, more diverse and more valuable resource for scientists studying the causes of and potential treatments for these inherited metabolic diseases. Details about the diseases currently represented in the repository are in the table below. There are many diagnoses for which we still do not have any samples and we would welcome additional samples for diagnoses for which we do have some samples.

If you are interested in donating a sample to help us build this valuable research resource for fatty acid oxidation disorders, please contact me either via email tschmidlen@coriell.org or by phone at 856-757-4822 for more information. You can also read more about this opportunity on our website: https://catalog.coriell.org/1/NIGMS/About/Information-for-Patients

Thank you again to those who have already donated samples!

Sincerely,

Tara Schmidlen, MS CGC
Genetic Counselor, NIGMS Human Genetic Cell Repository
Coriell Institute for Medical Research
403 Haddon Avenue Camden, NJ 08103
tschmidlen@coriell.org P: 856-757-4822

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Number of Samples</th>
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</thead>
<tbody>
<tr>
<td>2,4-Dienoyl-CoA reductase deficiency</td>
<td>0</td>
</tr>
<tr>
<td>3 Hydroxy Acyl CoA Dehydrogenase Deficiency (HADH)</td>
<td>0</td>
</tr>
<tr>
<td>3-Hydroxy-3 Methylglutaryl-CoA Lyase (HMG) Deficiency</td>
<td>2</td>
</tr>
<tr>
<td>Carnitine palmitoyltransferase I deficiency (CPT I)</td>
<td>0</td>
</tr>
<tr>
<td>Carnitine palmitoyltransferase II deficiency (CPT II)</td>
<td>2</td>
</tr>
<tr>
<td>Carnitine/acylcarnitine translocase deficiency (CACT)</td>
<td>3</td>
</tr>
<tr>
<td>Carnitine Transport Defect (Primary Carnitine Deficiency)</td>
<td>4</td>
</tr>
<tr>
<td>Electron Transfer Flavoprotein (ETF) Dehydrogenase Deficiency (GAI &amp; MADD)</td>
<td>1</td>
</tr>
<tr>
<td>Long chain 3-Hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)</td>
<td>1</td>
</tr>
<tr>
<td>Long chain acyl-CoA dehydrogenase deficiency (LCAD)</td>
<td>1</td>
</tr>
<tr>
<td>Medium chain ketoacyl-CoA thiolase deficiency (MCKAT)</td>
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</tr>
<tr>
<td>Medium chain acyl-CoA dehydrogenase deficiency (MCAD)</td>
<td>15</td>
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<tr>
<td>Short chain acyl-CoA dehydrogenase deficiency (SCAD)</td>
<td>8</td>
</tr>
<tr>
<td>Trifunctional protein deficiency (TFP)</td>
<td>2</td>
</tr>
<tr>
<td>Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)</td>
<td>4</td>
</tr>
</tbody>
</table>
It is with great sadness that we learned of a baby’s recent death within our ‘FOD Family’ in the last few months...please send your prayers and thoughts to Justina and Andrius Žukauskai from Lithuania.

From mom Justina, in her own words ~

First off all I want to sorry for my English.
Me and my husband Andrius are very glad to find such an organization. Reading and listening to your videos with the same situation word to word. I will try to tell short our story.

Our daughter was born in 2014-05-10 too early in 34 weeks, in tummy stopped growing, heart tone began to fall so went to emergency and they did cesarean section. Born small but beautiful 45cm, 1750 kilogram girl, Gabija.

Only now I understand why she was born too quickly... One month in hospital. She was true champion. Every day better and better. Everything was ok. Every month we visiting doctors, attending pool, everyday doing exercises, hire a baby massage. Everything was perfect and she almost catch up with peers.

We did exercises every day and I notice that my girl was sluggish, weak not like every day. After two days 2014-11-24 Doctor did Complete blood count (determine the amount of hemoglobin in the blood, the children often premature determined anemia). Next day 2014-11-25 I and my baby went to the doctor to know for results, doctor checked my baby, listen hard, results off blood was good, checked baby and reassure that everything was alright but that my girl has small runny nose and because of that she was tired, lost appetite, sometimes vomiting. The body is struggling with a runny nose.

Next day 2014-11-26 she didn't wake up...she was in coma (hypoglycemia). From one hospital we went to another because no one know what was happening. In third hospital genetics said that our daughter has very rare multiple acyl-CoA dehydrogenase deficiency (GA 2/MADD), and if the treatment does not work she will die... 2014-11-29 we lost our beautiful princess.

I hundred times thinking about that horrible week what we did wrong , what happened ... In Lithuanian language there is no information at all... As said doctors there was two families in Lithuania with similar situation but so serious multiple acyl-CoA dehydrogenase deficiency we are first...

Our geneticist told us that 25% our future children will have the same disease. Now do genetic studies in order to investigate the genotype, in Lithuania there are no such studies...maybe you can recommended where , and what cost to do that?? Geneticist says when I will be pregnant at 12 weeks do research and if my future baby is sick do an abortion ... So simply in 12 week when you can see it’s a boy or a girl when there is small human.. Now I know I am reading your web site, stories no killing ... I will procreate... But we have to get ready...

Can you advise, share your experience, I would like to talk to the same fate family...

Best regards,

Justina and Andrius Žukauskai

j.petkute@gmail.com
TeenKorner!
Sisters Lucy, 16 & Abbie, 18
Both MCAD
United Kingdom

Katie
8 yrs old
LCHAD
North Carolina

Leah
Almost 7 mos old
MCAD
New Hampshire

Please note that we also have an FOD KidsKorner/Adults Gallery and other Pictures on our homepage. To submit a pic please email Deb.

TEAM ELLA ~ UPDATE
Regional FAOD Meeting

There will be NO Spring meeting for 2015 ~

More Information will be posted when a date is scheduled
Email GoTeamElla@aol.com

Ella
DONATIONS
[since our July 2014 Newsletter]


Tshirts, Bracelets, Ribbons, CafePress, GoodSearch browsing, MissionFish/eBay selling, or iGive shopping: Jesse Rocha. Daniel Burbott.

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 http://www.fodsupport.org/donate.htm


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. ALL donations go toward FOD efforts and programs.

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

Reminders

Families - Please send TYPED (preferably in word document) stories etc, by June 15, 2015 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 June 15, 2015 to Deb.

‘Don’t be afraid to stand for what you believe in, even if that means standing alone’

~ Unknown