FROM THE EDITOR

Changes are coming to the FOD Group. As I have posted in previous issues, we are no longer doing a Conference every 2 years. However, we are hoping to have our 1st FOD Regional MeetUp some time in 2020 or 2021. I am not sure where it might be though – we are exploring Boston, MA and possibly Portland, OR. We have to work around various other Conference dates, Speaker travel schedules, and site availabilities.

Mitoaction.org is hosting an FAOD July 24-25 Conference in Pittsburgh in conjunction with INFORM and Children’s Hospital of Pittsburgh. This is not an FOD Family Support Group event, but it is a way for Families to network and meet others living with similar experiences and to hear Dr Vockley and his staff. To sign up for more information click here. ALL conference planning will be through them. So if you have any questions, direct them to kira@mitoaction.org.

We also may be making changes to our Newsletter ~ with everything already online and/or posted in our facebook and google Groups, we may forgo an actual Newsletter and just post Family Stories, Medical Info etc on our website on those specific pages. There has also been a serious lack of interest in members sending me Stories and other submissions. I am still exploring this option. Until then, think about submitting for July 2020!

In the meantime, help create Awareness around the world by promoting and sharing your Family Stories with everyone willing to listen. Please also share our FOD banner (as posted in our facebook group) on your social media sites. Another way to create awareness is to purchase some of our Awareness items and wear them with Pride! 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! Online and check Donations directly made to the FOD Group are tax-deductible.

Always remember ~ Whether you’re a Family or a Professional, we are all striving to create more 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
EDITORIAL

Over the last 12 years, we have been raising funds for future Research and the Clinical Training of new metabolic professionals. However, because of slow growth of those funds, our Board made the decision to end raising money for those funds and to go ahead and disburse them to active FOD Researchers and Educators.

On December 2, 2019 we sent checks to 3 institutions:

Research: Children’s Hospital of Pittsburgh @$30,000
Research: Oregon Health & Science University @$30,000
Education: VMP Genetics @$10,000

The FOD Group has had long and ongoing relationships with Dr Jerry Vockley at CHOP and Dr Melanie Gillingham at OHSU. Both are VERY actively researching the various FODs and also seeing many of our FOD Families in clinic. We felt that they could best use our Research Funds for their current and future FOD efforts. Similarly, we have had a long relationship with Dr Mark Korson. He previously was researching and clinically active, but has since moved to educating medical professionals on a variety of metabolic disorders. We felt strongly that his worldwide efforts will create more awareness of FODs, as well as inspire new professionals to enter the world of Metabolism. We are in desperate need of MORE Metabolic professionals!

So if you would like to donate to any of the Research/Clinical and Education institutions above, you can do so on our Donate page.

THANK YOU to Dr Vockley, Dr Gillingham and Dr Korson for ALL your efforts to help our Families! ～DLG

www.fodsupport.org

‘All in This Together’
At 4:04 p.m. Feb 17, 2002, our beautiful baby girl came into this world. Jenna weighed 8 pounds 7oz and she was 19.5 inches long. Right from the start I knew there was something special about this little girl. She had vibrant red hair and beautiful blue eyes. I asked the nurse if there had been a mistake. Had someone switched our baby? My babies are supposed to have dark hair and dark eyes. We all laughed and the nurse reassured me that this was our baby. Two hours later, Jenna and I were settled in our room.

Jenna was a very sleepy baby. I had a difficult time trying to get her to breastfeed. The nurses reassured me that this was all very normal because of the delivery. After many attempts I finally got her to nurse.

The next evening our pediatrician came to the hospital to examine Jenna. He listened to her chest, moved her legs and declared that she was a “healthy” baby. He told me to call his office to book Jenna’s first check up. Then he was gone. Later that evening the nurse took Jenna from me in order to do her heel prick test (at the time, testing for P.K.U. and Congenital hypothyroidism). Little did we know how important this test could have been for Jenna’s very survival.

We were discharged from the hospital the following afternoon. I was ecstatic. Everything seemed so great. Our circle was complete. We were truly blessed. Jenna was a strong baby from the day she was born she could hold her head up. It was like she didn’t want to miss a thing. At six months she had started on some solid foods and was enjoying this new sensation. As most infants do she especially enjoyed razzing when she had food in her mouth. At 7 months she was crawling all over the place trying to keep up to her older siblings. She especially loved to crawl towards the sunniest place in the room to bask in its warmth. Almost like she remembered where she had come from. At 8 months she was able to climb upstairs. By nine months she had started cruising around the furniture. We were certain that she would be walking very soon. Jenna was a wonderful little girl who loved to babble dada and melt her daddy’s heart. She would giggle with delight when you gave her raspberries on her tummy. Developmentally she seemed to be reaching her milestones and more.

The week of November 11, 2002, our son Justin was very sick with a virus. Jenna seemed fine except for a bit of nasal congestion.

On Wednesday November 20, 2002 Justin seemed better. He and Jenna seemed to have an average day. They played, and ate just like normal. I was relieved. I thought that we were finally free of the virus.

Later that evening, I went upstairs to put Justin in his PJs. Jasmine our eldest child, was holding her baby sister for me. Just as we were making our way down stairs, Jasmine yelled at me that the baby had just vomited all over the place. I cleaned everybody up. Jenna seemed fine. I thought that she had vomited from gas. Silently I hoped that she wasn’t coming down with the flu. I offered her a bottle and she drank the whole thing and kept it down. She seemed really tired so I put her to bed.
Thursday morning Jenna woke up around 7:45 a.m. which was late for her. She didn’t seem herself. She was very lethargic. I called the pediatricians office right when they opened at 9:00 a.m.

When I spoke with the pediatrician’s nurse I told her Jenna was not herself and that Justin had been sick the previous week with what I suspected was the flu. She asked me if Jenna had wet her diapers. I replied that Jenna hadn’t and that since she had woken up she was very lethargic. She asked if Jenna had a bowel movement recently, I replied that she hadn’t had one as of yet. The nurse advised me to come to the office for 1:30 and to keep pushing fluids in the mean time. I asked her if it was ok to give a nine month old pedialite. She said it was fine.

At 1:30 Thursday November 21, 2002 I brought all three children to the pediatrician’s office. When the doctor entered the examining room I was holding Jenna in my arms and she was quite sleepy. He commented that while she was quiet he would look in her ears. I then placed her on the examining table and she suddenly seemed more alert. I guess it was the change of environment or being out of the warmth of mommy’s arms. The doctor listened to her heart. He informed me that Jenna was running a fever and to give her Tylenol or Tempra. He told me that he didn’t think anything serious was going on. Her diagnosis was the flu. I was told to keep pushing fluids. Once again I asked if it was alright to give Jenna pedialite and he replied that it was fine but that I should try gastrolite as it was a less expensive alternative to pedialite. The doctor then switched his attentions to my other two children. After reassuring me that they were fine he left the room. I was not happy with the diagnosis but I said nothing. Something I deeply regret.

We arrived home from the doctor’s office around 3:30. I made dinner for my two older children. After making dinner I sat on the sofa with the baby. Her condition had not changed from when I had brought her to the doctor’s office. She was still very lethargic so getting her to drink was a challenge, but she was drinking so I took that as a good sign. When my husband came home the baby seemed to perk up a bit at the sight of her daddy.

We were having some flooring installed in our house the next day so my husband was busily preparing the house for the next morning. Around 11 p.m. my husband was getting ready to go to bed. I asked his opinion regarding the baby. She had just finished about two ounces of Similac and wasn’t vomiting any longer. I wasn’t feeling the best about putting her to bed. My husband reassured me that Jenna was in need of a good nights rest as I had been handling her all day long. I agreed and put her to bed. About half an hour later I heard her cry out. I checked on her and she seemed fine.

On Friday November 22, 2002 at 4:00 a.m. Jenna cried out. I bolted from our bed to check her. She was lying in her bed with her eyes closed. I decided to change her diaper and try to get her drinking again.

I tried giving her a bottle of formula but she would not drink. She just kept moving her head side to side as if to say no. I was worried that she was becoming dehydrated so I put some pedialite in a sippy cup. But she still would not drink. Eventually I did get her to take a sip of pedialite and she seemed content. A while later Jenna started making noises as though she were going to vomit. I thought that if she just vomited that she would feel better. At the same time my gut instinct started telling me that something wasn’t right.

I heard my husband in the shower. I went into the bathroom to look at the baby where there was more light. She didn’t look good to me. I mentioned this to my husband. Remembering the toll that the flu had taken on our son the week earlier, my husband reassured me once again that Jenna probably just needed more rest. We agreed that if she didn’t seem any better we would take her back to the doctor when his office opened.

Unfortunately, we didn’t get that opportunity at round 6:30 a.m. all hell broke loose in our home. The baby stopped breathing I tried to perform CPR while my husband was on the phone with the 911 operator. After what felt like an eternity, the paramedics finally arrived at our home. They whisked Jenna off in the ambulance and told us to meet them at the children’s hospital.

My husband sped off after the ambulance in his car while I waited for someone to come and stay with our other children. Finally, a police officer arrived and after some coaxing he agreed to take me to the hospital. It was the longest ride of my life. I kept my composure throughout the ride. I envisioned episodes of the television series ER. I thought that I would get to the hospital and they would tell me that my baby was going to be alright after a brief stay in hospital.
When we arrived at the emergency my husband was waiting outside for me. I figured that this meant one of two things.
1. That they were working on Jenna and he didn’t know what was happening. or
2. That Jenna had died.

Unfortunately, it was number two.

To say the least, we were in shock. How could this have happened to our baby who only two days previous seemed so healthy? This was the first time Jenna had ever been ill. Surely there had to be some mistake. Initially, we were told that the cause of death was Reye’s syndrome.

About a month later we received a call from the coroner’s office stating that the cause of death had been changed to a fatty oxidation disorder called Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD). Adding to our devastation, we have learned that this disorder would have been detected at birth with a simple $25.00 US ($40.00 CDN) blood test, similar to the heel prick test that is currently done for PKU. Since Jenna's death her PKU card (ie: heel prick test) from when she was first born was obtained from the Ministry of health. It was sent for MCADD screening. The results came back positive for MCADD. This proves that if Jenna had been screened for this disorder as a newborn we would have known what we were dealing with right from the start. A treatment plan would have been devised, and Jenna most likely would have had a healthy normal life. We were told that the general treatment plan is an adherence to a low fat and high carbohydrate diet. In times of metabolic crisis such as the flu or ear infections treatment is generally nothing more than an IV with glucose. Again let me repeat with early detection Jenna would most likely still be alive today.

The carrier rate for MCADD amongst Caucasians is estimated to be between 1 in 60 to 1 in 70. MCADD is thought to occur in approximately 1 in 9000 live births. The incidence rate for this disorder is just as high as PKU, which newborns are currently being screened for in all provinces.

Our child is not the first case of MCADD in Canada. Through the Fatty Oxidation Disorder Family Support Group we have learned of children and families who have been affected by this disorder all over Canada and the world. We have heard of many tragic stories like Jenna's and also many stories of hope. The stories of hope come from families who have been fortunate to receive their child’s diagnosis before a metabolic crisis occurs. Early diagnosis of these kinds of disorders via expanded newborn screening is saving lives, and offering better quality of life to those individuals diagnosed early enough to prevent severe neurological damage. Please use your voice to advocate for expanding newborn screening in your community. You will help to spare innocent children and their families a lifetime of needless heartache.

Sincerely,
Tammy and Roger Clark
Jenna 9 mos. – deceased MCADD
Justin
Jasmine

Editor’s Note – Thanks to the hard work and tireless efforts of families such as the Clarks, the Government of Ontario decided to implement expanded newborn screening in 2005, and currently tests all babies born in the province for 28 conditions, including MCADD.

[Note: from Tammy] Status of newborn screening programs Worldwide, including Canada 2015

In 2016, The Canadian Federal Government announced that Prov. Health Ministers had agreed to a list of 22 core conditions for newborn screening programs across all provincial and territorial jurisdictions, although some provinces were already routinely testing for 30+ diseases. These requisites are all considerably lower than the number of rare diseases that could be tested with a single bloodspot, and this is true of most countries globally (except for the United States, where some states test for 50 or more diseases).

***Not certain if this Canadian initiative towards greater uniformity for NBS programs has been implemented yet.
Current FOD Research Updates

For many years the idea of increasing the small amount of residual fatty acid oxidation capacity among patients with Long-chain FODs has been proposed as a potential treatment option. Several trials in Europe used a drug called Bezafibrate to stimulate residual fatty acid oxidation in patients with CPT2 and VLCAD deficiency with mixed results.¹⁻³ A new company (Reneo Pharma Ltd.) is trying a novel drug that is much more specific than Bezafibrate, called REN-001. The drug comes as a capsule consumed 1X per day. The pre-clinical studies in animal models and the studies in healthy normal volunteers suggest the drug selectively increases fat oxidation in muscle, is well tolerated and is safe. They are currently conducting a safety study recruiting 12 patients with CPT2, VLCAD or LCHAD/TFP deficiency who are 18 years or older. The clinical sites enrolling patients include OHSU, the University of Pittsburgh, Children's Hospital of Chicago and the University of Utah. I'm very optimistic about this compound. It appears to be more specific for muscle with little if any effect in the liver and more potently upregulate fat oxidation than other previous compounds, like Bezafibrate. The company will be evaluating the results in April or May of 2020 to determine next steps for further research on efficacy in patients with FODs. The goal is to mitigate or prevent the myopathy and rhabdomyolysis that occurs among patients with LC-FODs. If you want more information, see the clinicaltrial.gov listing.

There is also exciting research for developing a potential treatment for LCHAD deficiency retinopathy. Our group recently received funding to conduct a large natural history trial of 44 patients with LCHAD or TFP. We are currently finalizing the IRB approvals and consent forms to begin recruiting in January 2020. The goal of this project is use new noninvasive imaging to take pictures of the layers of the retina in patients with LCHAD deficiency and better describe how the retina changes over time and in different ages.⁴ The clinical centers enrolling patients will be OHSU and the University of Pittsburgh. In parallel, we are working on developing a retinal gene therapy approach in cells and animal models in the lab. Gene therapy in the retina was recently approved for another genetic retinal disorder that causes blindness, Leber’s congenital amaurosis.⁵ There are multiple retinal gene therapy trials currently using a similar approach to add a working copy of the mutated gene to the retina. Our basic science work will lay the ground work for developing a similar approach in LCHAD deficiency and the natural history study will provide detailed information about the course of the retinopathy to plan for a future treatment trial. If you want more information about the natural history trial, contact the study coordinator at OHSU, Ashley Gregor, MS at gregora@ohsu.edu.

It is an exciting time in science for rare diseases and new treatments are being developed in many areas. I’m encouraged and excited to see the same interest and treatment development happening for FODs. I look forward to the day when I can write about approved treatments in future editions of the newsletter.

Melanie B. Gillingham PhD, RD, LD
Associate Professor
Molecular and Medical Genetics
Oregon Health & Science University
3181 SW Sam Jackson Park Rd
Portland, OR 97239
gillingm@ohsu.edu

Studying rare metabolic diseases in the Baltic Sea Region

In early December 2019, I participated in a congress on public health that was organized at the Warsaw Medical University in the capital of Poland. Round table discussions featured members of the Polish Health Ministry and policy makers, members of the Polish National Health Fund, chief physicians and heads of major hospitals, and a few representatives of patient organizations to name a few. Among panels and round tables that focused on issues such as “Cancer Strategy” (Narodowa Strategia Onkologiczna in Polish), psychiatric health, the so called “obesity epidemic,” and vaccinations, there was one round table devoted to “Health-Based Healthcare.” A rather heated discussion revolved around efficient and implementable ways of replacing the current practice called “fee for service” with a more quality-driven one called “health-based healthcare.” As is often the case in Poland recently, the discussion focused on prices of drugs, including orphan drugs. When it comes to rare diseases—said a panelist and doctor who works within the Health Technology Assessment (HTA)—the current situation may be described as “prevention” (profilaktyka). Nobody questioned this statement. As a socio-cultural anthropologist who has been working within the field of rare diseases in Finland and Poland, I was surprised. Are special diets that constitute part and parcel of living with a rare metabolic disease mere “prevention” as the speaker said? What about hope for a pharmaceutical and/or technological cure that allows many families whose kids are diagnosed with a rare disease to carry on in their daily lives? Such questions remained salient, but unanswered.

Since 2016, I have been leading social science research projects that focus on experiences of people with rare metabolic diseases in the Baltic Sea Region in Europe. Together with other researchers and doctoral students, we have conducted comparative ethnographic research among families that have kids and teenagers who suffer from inborn errors of metabolism, adults, physicians, dietitians, geneticists, and members of patient organizations in Finland, Poland, and —beginning in January 2020—Sweden. Our research focuses on fatty acid oxidation disorders (FAODs), such as LCHADD, MCAD, VLCAD, and organic acid disorders (OADs) for a number of reasons. Let me name two here. First, according to medical literature, some rare metabolic disorders seem to be more frequent in European populations, especially around the Baltic Sea (in Poland, Finland, Sweden, Denmark, and Estonia) than in their North American, Australian or Asian counterparts. For instance, LCHADD is the most frequent FAOD in Finland and Poland. On the other hand, although GA-1 prevalence varies considerably between countries, it is relatively frequent in Sweden where it affects 1: 30,000 newborns. Secondly, the above-mentioned metabolic disorders have been included in newborn screening in all three countries in recent years. This allows for a faster diagnosis, the introduction of dietary treatment, and a lower mortality rate. However, the implementation of newborn screening raises ethical and emotional problems for the families; these are still understudied in the European context.

In the following, I briefly describe the healthcare policy on rare diseases in the European Union (EU) as a background for research topics that are examined by the projects. Finally, I will shortly address ethnographic methods utilized in our study.

In the European Union (EU), a disease is considered rare if it affects no more than 5 in 10,000 people. However, Member States may adopt a slightly different definition of a rare diseases; this is the case in Sweden, for instance. In Sweden, a disease is considered rare if it affects no more than 1 patient in 10,000 persons and it is combined with a severe lifelong disability. Rare diseases have been considered a healthcare policy priority at the EU and Member States levels since 1990s. The 1999 Orphan Medicinal Product Regulation was the first European legal document concerning rare diseases. One of the key documents on EU rare disease policy is the Council Recommendation on an action in the field of rare diseases (2009). In this document, the Council highlighted that "rare diseases are a threat to the health of EU citizens insofar as they are life-threatening or chronically debilitating diseases with a low prevalence and a high level of complexity. Despite their rarity, there are so many different types of rare diseases that millions of people are affected" (Council of the EU 2009: C151/7). The Council further urged member states to adopt a national plan or strategy for rare diseases by the end of 2013. According to EURORDIS -Rare Diseases Europe, a non-profit alliance of 837 rare disease patient organizations from 70 countries, the ultimate goal of a national rare disease plan/strategy is to guarantee rare disease patients’ access to timely and adequate medical and social care. In 2009, only 5 EU Member States had adopted a national rare disease plan (Bulgaria, France, Greece, Portugal and Spain). The situation changed within the next 9 years: by the end of 2018, 25 EU Member States had put in place a national plan or strategy for rare diseases. Malta, Poland and Sweden are still discussing the adoption of a strategy or plan.

In its policy documents on rare diseases, such as the European Commission Communication (2008), EU policy-makers emphasized equity and solidarity. These concepts should be the basis for Member States’ national policies on rare diseases that are to “ensure universal access to high quality healthcare”. In practice, however, there are a number of obstacles to obtain “high quality healthcare” for people with rare diseases in countries which our research has focused on. Money is of course a more common one. The distance to the nearest healthcare center could be another. For instance, with a population of approximately 5.5 million inhabitants, Finland is the third most sparsely populated country in Europe, after Iceland and Norway. Finland’s population is small and unevenly distributed creating a spacial dimension of inequality. Providing the same quality of healthcare to people with rare diseases outside large cities in south-eastern coastal Finland is a challenge.
Heightened medical, genetic, and public health attention to rare diseases in the last decade or so has focused on their “frequency,” “prevalence,” and “treatment.” As mentioned above, rare diseases have also been given special attention by political bodies, such as the Council of the European Union. Emphasizing their “low prevalence” and a high total number of those affected, the Council, however, has focused on mortality prevention as well as the development of new diagnostics and treatments (Council of the EU 2009). Anthropological and sociological scholarship has increasingly addressed rare diseases by writing about patient advocacy groups, patient activism (both in “real life” and in social media), and the issue of orphan drugs. There is also a growing interest in access to and (dis)advantages of newborn screening, especially in the context of health inequalities, kinship, and race, mainly in North America. Drawing on this body of literature, in our research we aim at examining more deeply the ways of coping with rare disorders in daily lives of the afflicted in a comparative perspective. Thus, we are focusing on a number of overlapping areas.

First, in order to analyze daily experiences of living with a rare disease, our research addresses issues related to dietary treatment and access to and utilization of biomedical technologies (e.g. naso-gastric tube and gastrostomy). We ask, for instance, are there any standards of dietary treatment that are utilized in the countries under study? What medicinal products can be reimbursed and how does this work? Is gastrostomy a more “standard” technology or a seldom utilized one? In this context, we also attend to spaces of care, caregivers and care receivers as well as material infrastructures that enable giving and receiving care. Who is, for example, responsible for caring for, or providing care for a child that has a rare metabolic disease? What kind of care can be obtained from state agencies (e.g. school and welfare services)?

Second, as the issue of disability is a recurring topic in the lives of many families whose member suffers from a rare disease, our research examines similarities and differences in ways of obtaining and retaining disability status and access to welfare services by patients with metabolic rare disorders and/or their family members in different countries. We ask, for instance, what are their educational paths and employment possibilities in countries under study.

Third, we examine the role of patient advocacy groups in the development of healthcare policies regarding rare disorders and orphan drugs (such as National Plans for Rare Disorders) and their implementation in Europe, and specifically in Finland, Poland, and Sweden. Do they have a similar impact on policy makers in these respective countries as is the case in the US? Additional questions arise as to the issue of “representation”: patient organizations tend to primarily represent people who have been afflicted with “more popular” rare disorders (e.g. PKU), while only secondarily looking after those afflicted with the “less popular” ones, e.g. LCHADD or GA-1.

Finally, we are interested in studying the production of knowledge about rare metabolic disorders and their treatment in the Baltic Sea region. Analyzing the emergence of the “rare diseases” category in Europe, in particular France, a French sociologist, Caroline Huyard (2009: 465) highlighted that it was a “by-product of the definition of orphan drugs.” The term itself appeared in the U.S. in the mid-1970s and was appropriated in the US Orphan Drug Act of 1984 and later in Europe. We examine how physicians, geneticists, and policy makers define “rare metabolic disorders” in all three countries. How do they “create” and implement treatment protocols for specific disorders? Medical literature indicates that in the case of some rare metabolic disorders there are no common guidelines and treatment protocols. Treatment guidelines are based on expert recommendations and a few clinical trials; they thus differ between centers and between countries.

As socio-cultural and medical anthropologists, sociologists, and scholars in cultural studies, we cannot develop new medical treatments or technologies that would cure people with rare diseases or facilitate their lives and those of their family on a daily basis. Our methods are more “on the ground” and the results of our work are more modest. By utilizing what is called “ethnography” we talk to and spend time with patients and/or their families, and we interview different stake holders in the field of rare diseases in Finland, Poland, and Sweden. By doing so, we would like to give them both a voice and the opportunity to share their experiences. Furthermore, a comparative study allows us to underline strengths and weaknesses of healthcare and welfare policies and practices in dealing with rare diseases in different countries. Taken seriously, this may eventually help to develop best practices for improving life with a rare disease.

Notes

Two grants were funded by the National Science Center in Poland: “Socio-cultural dimensions of rare diseases: The case of LCHAD deficiency. A comparative study of Poland and Finland” (2016-2019; Grant No. 2015/17/B/HS3/00107), and an ongoing project “An Anthropology of Rare Diseases. A Study of the Baltic Sea Region” (Grant No. 2017/26/E/HS3/00291). Additionally, a project titled “Food, Biomedical Technologies, and Care. The Case of Rare Metabolic Disorders” was carried out at the Helsinki Collegium for Advanced Studies, University of Helsinki in Finland (2018-2019). This project was funded within the EURIAS Fellowship Programme and the European Commission (Marie-Sklodowska-Curie Actions - COFUND Programme - FP7). For more information about our activities and projects see the website of our Rare Disease Social Research Center at: [http://rdsrc.ifispan.pl/en/](http://rdsrc.ifispan.pl/en/)

[Normalised Eating and Dietary Guidelines in LCHAD Deficiency - full published article]

Małgorzata Rajtar, PhD
Associate Professor
Institute of Philosophy and Sociology, Polish Academy of Sciences, Warsaw, Poland/
Rare Disease Social Research Center
mrjatar@ifispan.waw.pl
Our names are Jasmine, Catherine, and Eliana, and we are students from the Human Genetics Program at Sarah Lawrence College. **We are surveying patients with a rare disease diagnosis to better understand how they receive primary care services.**

We are hoping to have our survey (highlighted in the link below) sent to individuals with rare diseases and/or their families. We are focusing on conditions listed [HERE](#) by the National Institute of Health (NIH). We are reaching out to your organization because it was listed as a resource for one or several of these conditions we are researching. Can you please help us by forwarding this to your patient network, adding our link to a newsletter, and/or directing us to someone who can share this?

**Study information ~ DEADLINE Feb 28, 2020:**

- Participation is entirely **voluntary**.
- Responses are completely **anonymous**.
- It should take **5 minutes** or less to complete.
- Individuals with a rare disease OR their parent/legal guardian can fill out this survey.

**SURVEY LINK**

For questions or to request additional information about the study, please email me directly.

Thank you so much for your time and help with our study,

**Eliana (Kahan) Spielman**  
Second Year Genetic Counseling Graduate Student – Class of 2020  
Joan H. Marks Graduate Program in Human Genetics  
Sarah Lawrence College  
[ekahan@gm.slc.edu](mailto:ekahan@gm.slc.edu)
Info & Articles of Medical Interest

October 4, 2019, Dr Jerry Vockley from the University of Pittsburgh Children's Hospital spoke for an hour to discuss Fatty Acid Oxidation Disorders: The Other Mitochondrial Energy Diseases! When you click the link it may take a few seconds to load the zoom sound and picture.

When to Consider a Psychoeducational or Neuropsychological Evaluation for Your Child

High Fructose Delivers Low Blow to Liver’s Mitochondria

Get paid for your opinion and benefit the FOD Group at the same time. Patients (14 and older) and Caregivers (family, friends) of any disability, disorder, syndrome, disease or condition are provided an opportunity to voice their opinions through surveys and interviews to improve medical products and services.

Join the community on-line and earn a Dunkin Donuts, Starbucks or CVS gift card. We receive $5 for each qualified signup. Refer others and we will benefit each time. Your information is confidential, and your email/name is never shared. You may be invited to participate in surveys from time to time, where you will earn cash. Click on this link and join today!

Pharmaceutical Update

“The U.S. Food and Drug Administration (FDA) has accepted for review the company’s New Drug Application (NDA) for UX007 (triheptanoin) for the treatment of long-chain fatty acid oxidation disorders (LC-FAOD), a group of genetic disorders in which the body is unable to convert long-chain fatty acids into energy. The FDA has assigned a standard review designation with a Prescription Drug User Fee Act (PDUFA) target date of July 31, 2020.”

Email Eileen at eslawoffices@gmail.com for the entire pdf article
Creating Awareness & Family Fundraisers

Thank you to Michelle Prochaska and Paula Mousighi for ‘manning’ our FOD Awareness table at the 3rd Metabolic Summit for Texas Dietitians in Houston, TX on October 18, 2019.

Michelle Little and Chet Revinski also have summaries about their Awareness activities below ~

FOD Picnic July 2019 ~ Indian Ridge Picnic Area at Battelle Darby Park in Columbus, OH
We had a fun time at a picnic in Columbus, Ohio in July. Three FOD families showed up, with kids in tow. We got a chance to talk about tips and tricks we’ve learned for managing our (or our child’s FODs), concerns we had, local providers and where to go in emergencies. We enjoyed a sense of camaraderie and a feeling of connection. One person put it this way, “I feel so relieved to have met other people who know what I experience. I’ve felt alone a lot in my life, but today has really helped.” We will be planning another FOD picnic in Columbus in the summer of 2020. We hope to have a Regional Meet-Up as well, and to be able to connect with many of the families in the region!

RareMark
I had a conversation with Julie Walters, CEO of biotech company RareMark. The focus of RareMark is to connect patients and families with providers, researchers, and other families. Currently RareMark focuses on eight single-gene mutation disorders (such as Cystic Fibrosis or Sickle Cell Anemia). RareMark hopes to cover a wide variety of genetic disorders someday, to build a strong community that promotes strong relationships between patients and leads providers and researchers to develop new therapies. Mrs. Walters was very interested to learn about Fatty-Acid Oxidation Disorders and the large umbrella of genetic variations FODs cover. She was curious to know if any FOD families would be interested in someday pursuing gene therapy (using technology to alter DNA to reinsert into a patient’s body, so their body can produce DNA without a disorder). She is going to try and meet with Dr. Vockley in the future to discuss the possibility of gene therapy for various metabolic disorders.

My 11-mile kayak paddle was 7/24 starting at 6am. It was covered by the local newspaper and I wanted to make sure the awareness and opportunity to raise more money was positioned properly. My company matched 50% of each donation and I will be doing this continuous at 5% post event. Right now I’m just about at $1,500!

Chet Revinski

[Note from Deb: Thank you Chet for your generous FOD donation!]
FOD Awareness in Australia ~

FOD Supermums Jessica (Mum to Henry and Rosie, LCHAD) and myself (Mum to Leo, MCAD) participated in the Sydney City 2 Surf yesterday! 14km of hard work for our kids, raising money for the Children’s Medical Research Institute (Jeans for Genes). Between us we raised thousands, which will go towards research including gene therapy for metabolic disorders.

This was an amazing experience. Every little ache and pain we experienced is nothing compared to what all our FOD children face. We hope that one day, research like that of the CMRI will give our kids the cure they deserve!

Jessica Seeland

Love Messages

We have had some deaths over this past year in our FOD Family…

Please remember our Families in your thoughts and prayers throughout the year ~ All of our FOD children and adults will ALWAYS be with us in our hearts!

Bentley Powers, age 9, LCHAD

Bentley Lewis Powers, age 9 of Princeton passed away Tuesday, July 9, 2019 at Caldwell Medical Center. Bentley loved his family and always had a hug for everyone. He loved to play Fortnite and Rocket League as well as watching WWE wrestling, especially John Cena. He was a silly boy who loved playing jokes on his family and friends. He loved to dance and sing to his favorite song "Old Town Road." Bentley was a bright young boy who touched every life he encountered. He was his momma’s fighter, his daddy's hero and his papaw's best friend.

He is survived by his parents Cassie Swan Powers of Princeton and Richard Powers of Murfreesboro, TN; three sisters, Kaylynn McKinney of Princeton and Destine Powers and Faith Powers of Murfreesboro, TN; three brothers, Ian McKinney and Scott Whitasel of Princeton and Edward Powers of Bucksport, MA; maternal grandparents Brenda and Michael Swan of Princeton; paternal grandparents Richard Powers of Murfreesboro, TN and Connie Powers of Murfreesboro, TN; three aunts, Carrie Melton and husband Brent of Princeton, Devon Mounts and Jeremy McDonald of Smithland, KY and Amanda Powers of Mufreesboro, TN; two uncles, Eddie Guthrie and wife Mandy of Mufreesboro, TN and Brandin and Lauren Powers of Mufreesboro, TN and four cousins, Derek Lane, Ellie Powers, Emeri Powers, and his best friend, Bailey Lane.

‘Hope is being able to see that there is light despite all the darkness’

~ Desmond Tutu ~

www.fodsupport.org

‘All in This Together’
Chuck Hehmeyer recently updated an article he did for our site years ago - VERY interesting and helpful for any Family that has experienced medical malpractice/negligence (in regard to rare disorders etc). He has worked on several FOD cases over the years.

Nutritional Guidelines for VLCAD

“We now have a software solution to help you enable your employees with ADHD, Dyslexia, learning differences and Autism to work to their strengths! We survey your employees and prescribe individualized supports and accommodations so they can work productively and feel valued. Also I’m about to launch a new podcast - It’s called For All Abilities (like my business) and I will be interviewing and telling stories about people who are highly successful and have adhd, dyslexia, learning differences and high functioning autism. For All Abilities: The Podcast is meant to inform the world of the amazing people out there who are succeeding in HUGE ways with brains who don’t fit into the imaginary norm.”

Betsy Furler, email Betsy for more information.

My name is Marta Campabadal and I work for EURORDIS (European Organization for Rare Diseases), as Manager for RareConnect, the world’s leading rare disease virtual platform.

This is to inform you that we have recently created an online community for Primary Carnitine Deficiency in partnership with a mother that has an affected child. Please share this info.

Primary Carnitine Deficiency Community link

The highlights of the community are:
- It is available in 12 languages
- It offers peer-to-peer support
- A translation system guarantees the translation of all posts and stories
- It represents a meeting point for families that do not speak English or cannot count on any patient associations within their reach

Your organization can also participate in the community. Here's how to:
- Join the community and create a profile
- Propose yourself for the role of moderator
- Add a link to the community on your website
- Send us informative links, pdf, articles... to be added to the Resources section
- Share the link with your members, social networks, newsletter...

We want to transform this community into a place that can fulfill the needs as a patient or caregiver, where they can have meaningful connections with other members worldwide. We would really appreciate any help you could offer.

Thank you for reading my message. Should you have any questions, please email me

Mito Foundation ~ Australia

Inside the ADHD mind

Just Like You Dolls

Heathcare Toolbox: Guide to helping children and families cope with illness and injury
Embracing & Living with an atypical FOD...having a lifeline

By: Stacie Poole

FODsupport.org has been a lifeline for our family for many years. When we found ourselves confused, worried, feeling alone or needing guidance, the FOD family has always been readily available at all hours of the day and night, to walk us through moments of stress and join us in life’s celebrations. Truly, an invaluable part of our journey, the families, adults and administrators at FODsupport.org will go down in our family’s history book as a “before and after” moment...what life was like before we connected with FODsupport.org and how much better our lives were once it was a part of our support network.

Even with all the information available to our families through posts and conferences, it may be surprising to learn that many families, like ours, have atypical forms of the FODs we have all come to learn so much about. That additional “rare-ness” can lead to a lack of belonging, even within the generous and loving FOD community.

Our family does not have a classic gene mutation. In fact, no gene responsible for decreased CPT2 activity thus far, has been identified within our family. Yet, within muscle and skin cells, the enzymatic numbers do not lie. Decreased activity has been confirmed repeatedly between multiple samples and across various institutions. The constellation of rare and very rare genetic variants that effect our family in various, impactful ways, also effect CPT2 activity levels, some family members more than others.

It’s been a rocky road. One, that as the uniqueness of our genetic playbook unfolded, made us feel like we fit in less and less, even among our fellow FOD’ers. MCT oil made our child sick. Muscle pain was intense. Stamina and fatigue were real. Doctors struggled to find answers. We struggled to find peace. Eventually, a combination of multiple genetic factors effecting the same pathway was determined to impact many of the necessary proteins and functions of our bodies. By then, leg bracing, occasional wheelchair use, GI impact, migraines and hypoglycemia were a normal part of our family members’ worlds.

It became more and more clear that doctors all over the country had never seen our genetic variants and had not had the chance to fully research these specific spots on the genes they so passionately researched. It was however, fairly certain the gene changes had to be causing our issues based on what the variations did to the genes and their function. We were initially more confused and more alone than before. We would see posts about CPT2’ers and relate to their journey but look at our child and know there was a difference, even with a confirmed CPT2 diagnosis. We were supported, but on a road, alone. Where did that leave us? With whom could we connect? Without an easy, identifiable name to describe our family’s genetic mumbo-jumbo, who would we turn to when we were confused and scared?

Many families within FODSupport.org are like our family. Other disorders can cause secondary FOD impacts similar to the classic gene mutations, but with a presentation and unique set of issues that can further complicate their care. Additional hospitalizations, complex, multi-issue treatment plans and long-term impacts that are slightly different than families working through FODs with classic gene mutations can be a normal part of their world. This can be hard to remember on posts, at conferences or when supporting one another and it can lead to a lack of understanding.
Prior to further genetic testing, NIH had shared with us that there was likely a dominant inheritance pattern to our family's genetic issues. And they were right. Sharing this on FODsupport.org upset some people. We had to work through that. What was accurate for our family wasn’t accurate for those with the classic gene mutation as I had explained, but hearing this information from an FOD family made some people worry that it changed their own futures or children’s futures. Flying in the face of classic gene inheritance patterns, our family was an example of things researchers were still learning. Trying to communicate this was challenging and difficult for some people to understand. It placed us farther away from an important source of support.

Life is nuts. Let’s just say it. It’s true! So much of what we experience is beyond our control, understanding and ability to change. We can no more reach into our own DNA and fix these genetic hiccups than we can lift our house and move it up a hill (although with CRISPR, the ability to make permanent, genetic changes may be coming, sooner than we think! For an example of this topic search “CRISPR and muscular dystrophy, benefits and risks”).

What we can do is continue to remember a few things...

1. Researchers have barely scraped the surface when it comes to genetics, their purposes, defects and ways to help them perform better. One doctor said to us “only the Creator knows what’s wrong with your child”. Not very comforting when your 18-month-old is sick and other members of your family are struggling, but he too, was right. We always said our little one was the “canary in the coal mine”, those issues leading to a greater understanding of what other family members had struggled with since birth. Trouble is, human genetics is super complex. Genes are one thing, but did you know a strong or weak promoter can impact whether a gene under or over expresses itself? Did you know the area around and in between the gene itself is like the Grand Canyon? Many researchers have proven the “Grand Canyon’s” worth and continue to work on this very important area in spite of the once held theory that this area was just “junk”. Now, it is understood to clearly be a vital part of our overall genetic picture. Not much junk in the human body, it seems. (Except for too much holiday food!)

2. Not every FOD person is the same. I won’t speak for the classic gene community, but I can say that within the non-classic gene community, variation is King. Some people struggle more with hypoglycemia than rhabdo. Some have debilitating muscle pain. Some have up and down days without rhyme or rhythm, based on physical exertion, sleep, overall GI function, illness and growth spurts, or even the temperature of the day, like our kiddos.

3. The future is bright, even for Rare Families, like ourselves. Classic gene mutations within the FOD community are gaining increasing numbers of clinical trials, studies to help understand and solve everything from massive cardiac involvement to rhabdomyolysis. Research in classic genes and their impacts are critical to improving outcomes, quality of life and overall treatment plans for the entire FOD community. Valuable information gained from classic gene research can also help doctors treat the rare variant person, increasing treatment options and improving the ability to create effective treatment plans based on comparable symptoms to the classic mutation community.

Even within our rare, small slice of the genetic pie, variation exists. Embracing genetic diversity within the FOD community is critical to offering families the much-needed support and guidance they so long for and solutions they so desperately need.

Thank you for the love and support you’ve offered our family. We hope anyone struggling in the desert of an atypical FOD diagnosis will reach out, ask for help, say what you need, and let our community rally around you. Together, we can continue to forge this road, breaking down barriers to knowledge, embracing our variations (pun intended), and supporting one another through life’s ups and downs.

Here’s to a bright 2020 full of joy, good health and medical breakthroughs! From our family to yours, we wish EACH AND EVERY ONE OF YOU a beautiful, safe, HAPPIEST of New Years!
More Lifeline Support...

~ Facebook Groups for FOD Families ~

- Main FOD Group for ALL FODs
- LCHAD WARRIERS
- Long and Very Long chain FOD food group
- GA 2/MADD Families
- Carnitine Deficiency (Primary and Secondary)
- MCAD Deficiency
- Raising Rare and Beautiful Children with CPT 2 Deficiency
- Fatty Acid Oxidation Disorder (FAOD)
- LCHAD Poland
- Parents of VLCADD Kids
- Adults with FODs
- Metabolic Support UK
- MCADD Families UK
- The Metabolic Foundation - UK
- MCAD Norge
- Ethan James Wyne MCADD Organization
- Mitoaction

Genetic Mistakes, Understanding and Living with Fatty Acid Oxidation Disorders, by Rosemary Forrest and Nicole Baugh, is published by Nova Science Publishers (ISBN#978-1-53612-244-2) and is on Amazon.

Rosemary is a CPT 2 grandma!

NEEDED for JULY 2020 NEWSLETTER ~

KidsKorner Pictures, Family Stories, Special Articles, Reach for the Stars, and Professional Articles etc

Please think about sharing ALL of the above for upcoming issues ~ for ALL Submissions please email t Deb

Pictures ~ please include their name, age, disorder, and state/country and that you give me permission to print in the Newsletter

We are also looking for Families or Professionals to do their own videos that tells their Stories or creates FOD Awareness in other ways...such as sharing how they prepared for a school IEP, what is necessary to have ready before a hospitalization, how has their disorder changed as they have grown into adulthood etc. Then we can upload it to our FOD youtube channel! So email me if interested.

www.fodsupport.org

‘All in This Together’
Thank you to all that have done their own ‘Facebook Birthday Fundraisers or In Memory of Donations’ to benefit the FOD Group ~ all the donations are greatly appreciated and will assist us in either our General costs, Networking and Educational Services & Programs, Grief Support, or other areas of the nonprofit that need funding!

Some of the Families that created facebook Birthday fundraisers or In Memory Of Donations since our last Newsletter included: Laura & Jesse Rocha and Jacqui Robinson Kemp. Memory donations for Matthew Koch, Dominic Ray and Tom Carmody, Sr. All of our current donations from the last 6 months are posted on our last page!

If I missed anyone please let me know. Facebook sends the funds 30-60 after the end of the fundraiser so be sure to let me know when your Fundraiser ended and how much was raised. I will look for that in my automatic deposits ~ HOWEVER facebook never sends me names so I don’t have any idea which Fundraiser it was from - so please let me know!

And it doesn’t matter if nothing was raised monetarily…you raised AWARENESS and that is what’s important!

Also THANK YOU to the Committee members for the INFORM Network and the FOD Registry for all your work on FOD efforts~

Michelle Little, Dave Perritt and Brittany Leigh Pridal
Lindsay Johnston, Christy Perez and Brittany Leigh Pridal

Mailing lists: Erika Wallace
Website Designers: Mary Lingle, Jamie Payne
Newsletter consulting: Brian Gould
Email/website consultants: Mark Heinz
Website slide shows & Graphic arts: Keith Widmann
FOD/OAA Conference Event Planning: Eileen Shank
Website slide shows & Graphic arts: Keith Widmann
Newsletter formatting: Deb
FOD GROUP FINANCES

2019 FOD Group Tax Return will be up on the site by May 2020

The bulk of Expenses are for monthly phone, website fees, supplies, MeetUps/Seminars, Insurances, and for our Grief Consultation office (rent, advertising, etc) to offer pro bono grief support to local Bereaved Parents & Families (and also via Skype/phone to FOD Families around the world). We also donate FOD funds from undesignated donations to various FOD related entities (ie., for NBS issues, outreach) to support their efforts.

All Undesignated and Grief Consult donations are deposited into the General Fund or Gen Trust Fund, as are Awareness Item Sales, Cafepress.com, iGive, amazonsmile, etc and any donation that isn’t specifically designated. Online links are available for outside Research and Clinical clinics/individual researchers if you’d like to directly donate to their FOD efforts. No FOD money is used for salaries - we are an ALL Volunteer organization.

Additionally, we have a 1yr & 3yr certificate and long-term stocks/bonds earning interest and dividends for future FOD endeavors and programs.

THANK YOU [DONATIONS SINCE JULY 2019]


Some of the above donors have purchased Tshirts, Bracelets, Ribbons, CafePress, or used GoodSearch browsing, MissionFish/eBay selling, iGive or AmazonSmile.org shopping etc.

Thank you to all that have bought products from companies on the Internet that support the amazonsmile, iGive, GoodSearch and GoodShop, and Cafepress.com programs of donating a certain percentage to Groups like ours. All of those links are on our website.


We greatly appreciate donations to help with daily costs, website fees, insurances, supplies, Regional MeetUp/Seminar costs, phone calls around the world, rent for the Grief Consult office, and raising funds for future Programs & Services and long-term investments.

ALL donations go toward FOD efforts & programs.

US checks made payable to the ‘FOD Group’ mailed to: FOD Group PO Box 54 Okemos, MI 48805

Families and Professionals...

Please send all Submissions to Deb by June 15, 2020 for the July 2020 Newsletter. We are always looking for Family Stories, Professional Research and Clinical summaries, New Babies and KidsKorner pics etc. Also keep spreading the word about FODs and expanded Newborn Screening ~ it could save a life!

‘Write it on your heart that every day is the best day in the year’
~ Ralph Waldo Emerson ~