

FOD Family Support Group ~ Communication Network

♦ Family Support & Networking  
 ♦ Global Awareness & Advocacy  
 ♦ Newsletters  
 ♦ Conferences  
 ♦ Fundraising for Clinical Training & Research

**FOD**  
All in This Together  
Family Support Group

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Family Support Group**

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**FROM THE EDITOR**

Happy New Year to ALL of our FOD Families! It seems like yesterday that we were wishing that for 2016 ~ time most definitely is flying by. Last year was an exciting time for some of our Families that experienced their very 1st FOD/OAA Metabolic Conference and we hope that many more will begin to start saving so they can attend our **next Conference in July 2018**. Kathy Stagni (OAA) and I will be exploring some new cities in the coming months, along with Eileen Shank (our fabulous MCAD mom event planner), and we will let everyone know which one we decide on. We are **still searching for a major Sponsor** (ie., Metabolic clinic or Hospital, University Medical School etc), so until we finalize our Sponsor, we won't have any Conference or Hotel information. Stay tuned!

This year we hope to increase the number of FOD Families in our Registry ~ We invite all FOD patients to join (ie., Parents of FOD children, as well as our FOD adults 18+years old) to provide **de-identified medical information** to the registry to help everyone in the global FOD community ~ patients, families, researchers, clinicians, and pharmaceutical companies ~ to learn more about Fatty Oxidation Disorders. The **goal is improved diagnoses and medical care, as well as empowerment of patients and families** through knowledge, connections, and support. We are in the process of adding new questions to the Surveys ~ so we'll let everyone know when that process is complete. In the meantime, be sure to read the Press Release on page 15 and help our researchers find answers to help you ~ **join our [FOD Patient Registry](#) and participate in future trials and studies!**

Always remember ~

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

## BOARD OF DIRECTORS

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## EDITORIAL

In an ideal world, there would be no illness and no suffering, and there wouldn't be any mistakes that could forever change the lives of individuals and their families. Yet we all know our world is not immune to illness and suffering and being terribly wrong. The article *'The price of being wrong'* by Ellen Gabler of the Milwaukee Journal Sentinel painfully demonstrates that all too well.

What a beyond terrible tragedy...all because (in my humble opinion) of differences in range limits on a newborn test, a test that has variable ranges across states in this country. If the baby had been born in another state, he and his Family most likely wouldn't be living with the major challenges described in the article. The different ranges may not be the situation in every case – there also may be other factors involved such as not following protocol for taking the bloodspot, or not getting it to the Lab within the prescribed time etc. Yet the outcome of those mistakes can often be horrendous for Families.

Policies need to change so EVERY state can be on the same page as far as how these disorders are screened for AND that the results and follow-up procedures are CLEAR to the Family and their Drs. Unfortunately, like many things in our world, money and politics and lack of common sense often take precedence over the well-being of our most vulnerable.

My hope and wish for 2017 is that our newborns' lives become the priority. Just as many of our grassroot organizations advocated newborn screening in the 1990s+ it's time to let state newborn centers know that they NEED to adopt UNIFORM screening ranges/procedures so that babies do not go undetected! Resources to refer to are the [National Newborn Screening & Global Resource Center](#) and [Baby's First Step](#) ~ let your voices be heard...it WILL save lives! DLG





## FAMILY STORIES

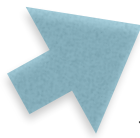
January 6, 2009 my life would change as I gave birth to a baby girl named Kelli. My heart was full and excited with having my first child. However things quickly changed the following day when she was getting placed on a blanket to take her hospital photo. When the photographer went to lay her down she started choking and stopped breathing.

I can remember my sister grabbing her and patting her back trying to get her to come to when a nurse came in and said pull the CODE BLUE! My heart sank as I pulled the code blue and within seconds a team of about 8 to 10 nurses came running in grabbing her and running out. We were then placed in the NICU not knowing what was going on and then brought to the special care unit as she was getting better so we thought.

However, things changed again when she had another episode and was placed back in the NICU. A week later we were discharged to go home but had to go see the pediatrician immediately. When going to see them, we were given a computer printout on the FAOD VLCAD and the doctor telling us he had never heard of it, but to follow the steps.



A month later we were sent to the geneticist where it was confirmed that when my countless four hospital stays



where it was confirmed that when my countless four hospital stays where it was she had VLCAD with the gene mutation of a zebra fish. That is world changed. Kelli would throw up and end up in the hospital times. In just the first year of her life we had already had twenty-four and seeing the hospital as a second home.

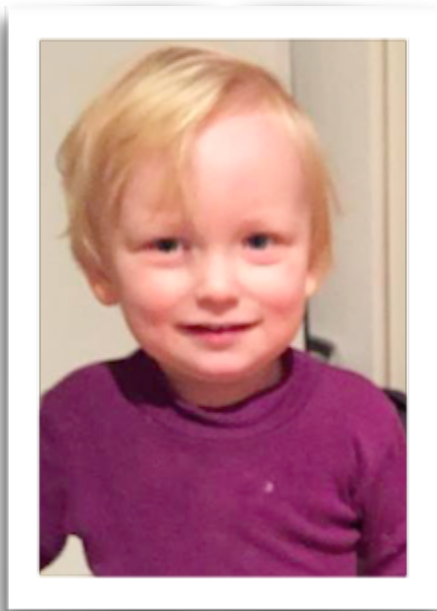
### 'KELLI STRONG' 2016 VLCAD

On Kelli's next hospital stay, the day after she made one, we had been through six IVs and 17 sticks with three in the neck and her cardiologist having to access her veins. We were then sent to a surgeon for a port which was definitely a blessing. Throughout the next few years we would be in and out of the hospital. Kelli at the age of six had sixty-five hospital stays with one of her last stays almost leading to a blood transfusion.

We have had to fight and be a voice for her on numerous occasions, but if we wouldn't have then things would have turned out completely different. Kelli is now about to turn eight years old and I can proudly say that on December 1<sup>st</sup> of 2016 **she made one year hospital free!** This has been a huge milestone for her as it has never happened.

Kelli loves taking care of and playing with her baby brothers, David and Gary, and likes playing with her friends. She has become a huge Alabama fan with a house full of LSU fans, but that doesn't stop her from cheering for them. She loves to play outside especially while roller skating. Her absolute favorite thing to do is anything with crafts. Her favorite movie to watch that she falls asleep to all the time is 'Mary Poppins.' She actually got to walk with Mary Poppins in Disney while holding her hand!. Dancing, singing and putting on fashion shows are also things Kelli loves to do ~ **her VLCAD does not keep her from enjoying her family, friends and life!**

**Jennifer Guillory     Baton Rouge, LA**



My son, Jonas Maximilian. Born 19 May 2015, now 90 cm, 13 kg. Dad, Christian (Norwegian) and mom, Hannah Luise (German), live in Oslo, Norway. MCAD, was diagnosed one week after birth. We had a horrible first 9 months, but then it got better. Several hospitalizations so far, but he's quite healthy on the whole. He is strong-willed, can't stand vegetables or any other health food, loves cars and climbing on things, and can say four words: mommy, daddy, thirst, and pool!

**Hannah Luise Mykland**

## Parent to Parent Suggestions

For those looking for a tasteless version of B2 , you may want to consider [Cyto-B2](#). It mixes best with a warmer liquid (it is a powder) - GA 2ers use it as part of their treatment plan, but some other FODers use it to reduce the fishy smell that can occur with the use of prescription carnitine.

[Rachel Bagosa Quapp](#)

I am a parent of a 2-year-old with VLCAD. I've found my way to make life easier for not only my son, but my husband and I as well, is to modify the foods we love in life. It's little changes here and there, and in the end, is healthier for all of us. Feel free to check out [my website](#) . I've been posting every two weeks or so, but if you have recipes you have no idea how to modify, let me know. I love to experiment. I also include nutrition facts on the recipes.

[Justine Goode](#)



## MEDICAL UPDATE

Professionals: Please contact [Deb](#) if you'd like to write an article/summary for our next issue. Our Families are really interested in learning what



### Autism and Metabolism

Stephen G Kahler, MD

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This essay is based on a talk I intended to give at the 2016 FOD-OAA meeting in Denver. It is based on my own observations and those of Dr. Richard Frye, MD, PhD, who directs our Autism Specialty Clinic. In this clinic we see patients diagnosed with autism and attempt to understand the metabolic/biochemical abnormalities that are present in these patients. Using this approach we then implement treatments designed to improve the underlying biochemical abnormalities, and thereby lessen the autism symptoms. Dr. Frye also directs a research laboratory devoted to studying mitochondrial dysfunction in patients with autism.

Autism is now familiar to most Americans, thanks to increased awareness, increased recognition, and what appears to be increasing prevalence. It is a description of how someone's behavior appears to us, not a specific condition with a single cause, or even a specific condition with several possible causes. The fundamental characteristics of what is called autism include problems with communication, problems with relationships with others, and restricted interests and repetitive behaviors, including insistence on sameness. The onset must be in young childhood or earlier.

Within the broad definition of autism, or what some call "the autisms", are a variety of related conditions, which have less severe problems, that help to make up the "autism spectrum disorders". In Asperger syndrome communication is good; in PDD-NOS (pervasive developmental disorder-not otherwise specified) there is less restriction of interests, and less repetitive behaviors. The onset may be sudden, often accompanied by a febrile event; or the symptoms may appear gradually.

The fact that there are families with more than one child with autism, and families in which different members may have different forms of the autism spectrum disorders, suggests that there is a genetic contribution to the autism spectrum.

Autistic symptoms are associated with changes (mutations) in a large number of specific genes, and regions of chromosomes, or additional entire chromosomes.

Finally, many children with autism have disturbed intestinal function, with slow intestinal transit and abnormal bowel movements (too runny or too firm). Many children have distinct food aversions, or extremely strong preferences, so that meals are one more example of repetitive behavior.

Among the genes and genetic disorders that can be associated with autism are Rett syndrome, Williams syndrome, tuberous sclerosis, Down syndrome, abnormalities of small genetic regions, and abnormalities of a large number of genes associated with brain function, especially genes involved in the formation of synapses (the areas where two nerve cells make a connection—there are billions in a human brain), and genes involved in certain biochemical processes.

Some of the specific genes contribute to processes to create or transport certain molecules in brain cells (neurons) (for example, creatine), to inactivate excess amounts certain harmful molecules that we make or ingest (e.g., ammonia, phenylalanine), and to provide the energy necessary for proper functioning of neurons (the processes that take place in the mitochondria).

Some of the chemical compounds that contribute to autism in some children are propionic acid and its variations. We all make a lot of propionic acid, generally in the form called propionyl-CoA, and we absorb a very large amount, made by our gut microbes (the "gut flora"), which should be processed in the liver to less dangerous substances, and used for energy. It has been known for a few decades that propionic acid and related compounds can interfere with brain function, and that laboratory animals exposed to an increased amount of propionic acid can exhibit autism-like behavior—restricted repertory

of interests, poor interaction with other animals, and poor communication/vocalizations. This sounds similar to what sometimes happens to children with propionic acidemia, in which propionate accumulates because of genetic alterations in body chemistry (metabolism). There is growing evidence that some children with autism may be being exposed to, or are inadequately responding to, high levels of propionic acid made in the gut. The obvious question is would the autism relent if the propionic acid burden was lower?

So we investigate children with autism by looking for abnormalities in a wide variety of body compounds (metabolites), analyze their genes for extra, missing, or altered regions, consider if there are dietary habits and intestinal problems that might be contributing, and consider the role of oxidative stress. We can't change a patient's genes, but we may be able to repair some of the problems caused by gene mutations, and we can certainly try to improve abnormal metabolic processes.

One of the early discoveries about metabolism in children with autism is excessive oxidative stress. This is a quick way of saying there is evidence of damage to tissues due to oxygen. Oxygen is essential for life, but it is also a toxic substance that can damage our tissues. This is similar to what happens in a car—oxygen is essential for burning the fuel (gasoline), but it also damages the car—causing rust spots on metal, causing plastic to change and become brittle. We can measure oxidative stress by looking for increased levels of molecules damaged by oxygen. Damaged amino acids, fatty acids, and nucleic acids (the building blocks of DNA, our genetic material) are found in a significant number of children with autism. The connection between this damage and the autistic behavior isn't clear yet—it's very much like your car not running perfectly, finding rust spots on the bumper, and wondering if there is a connection.

Glutathione is our main anti-oxidant. Many children with autism have inadequate free glutathione. There are ways to improve the situation, and lessen the autism symptoms. Large doses of vitamin B12 (even though the blood level is normal), and N-acetylcysteine (a precursor of glutathione) can be helpful.

Mitochondrial dysfunction in the brain can cause subtle and regional problems, or overwhelming brain damage, seizures and coma. In the muscles there may be weakness or hypotonia. When we find this in a child with autism we are especially likely to investigate mitochondrial function by measuring lactic acid (increased level means mitochondrial impairment), and related substances. Mitochondrial problems sometimes respond to certain common nutrients that we can give. These are collectively called "the mitochondrial cocktail". There are several versions, generally including at a minimum carnitine, riboflavin (vitamin B2), coenzyme Q10 (co-Q), and thiamine (vitamin B1). Like children with primary mitochondrial problems, autistic children with impaired mitochondrial function may benefit from treatments that address this.

Altered immune function is another common aspect in some children with autism. This may be expressed as abnormal intestinal immunity, inflammation, abnormal bowel movements, and intolerance of certain foods because of altered digestion or excessive absorption of toxic substances derived from foods. The foods most commonly involved in this process are proteins present/derived from milk (casein) and wheat (gluten/gliadin). Eliminating these foods from the diet may dramatically improve brain function and lessen autism symptoms. The gut flora includes several hundred different sorts of microbes, all interacting in ways we are barely beginning to understand. If there is excess production of propionate or inability to deal with it, an antibiotic such as metronidazole (Flagyl) may change the numbers of certain microbes, and lessen the production of propionate.

Deficiency of certain nutrients in the brain may also occur. This especially includes the vitamins folic acid and tetrahydrobiopterin, which will lead to altered levels of neurotransmitters, the substances that neurons use to communicate with each other. Serotonin is the neurotransmitter most conspicuously altered in autism (and also depression)—using a low dose of a selective serotonin reuptake inhibitor (SSRI) such as fluoxetine (Prozac) prolongs the action of serotonin, and can lessen symptoms. 95% of the serotonin in the body is made in the intestine, so drugs which influence the bowel may affect serotonin levels.

All of these observations and ideas can be part of understanding autism even in a child who already has a "cause" for autism—Down syndrome, Rett syndrome, propionic acidemia, Angelman syndrome, etc. Of the disorders of most concern to the FOD Family Support Group and the Organic Acidemia Association, for whom I first developed these thoughts, the organic acidemia patients are the more likely to have autistic features. Autism doesn't occur in every patient with propionic acidemia, however, so we must be aware of what other factors might be contributing, and can we improve the situation. Some children with autism with or without a named metabolic error will have a significant improvement if they are only IV feeds (total parenteral nutrition, TPN) for a while. This tells us that something about food or the gut flora is playing a role. Other patients will have a major improvement during and shortly after an illness with fever. A new drug, sulforaphane, derived from broccoli sprouts, may benefit them (and perhaps others) greatly.

To summarize: Autism is a collection of symptoms, not a specific entity, that occurs in a variety of settings; there are numerous nameable genetic "causes" of autism; autism is a systemic disorder which affects the brain as well as other organs; there are numerous biochemical/metabolic problems which can be found in various combinations in children with autism; these problems may be treatable by diet, nutritional support, medications; autism may occur in children with metabolic disorders, especially some of the organic acidemias; treatments that help improve the metabolic situation may improve the autism symptoms as well; and we must keep in mind that if we can't explain and treat all the symptoms of a patient we may be overlooking something that could offer additional benefit.



Imagine you are riding in a smart car; a tiny, pip-squeak of a car with no back seat, barely room for a spare tire or a bag full of groceries and meant to get you from point A to B, nothing more. Now, imagine you somehow are able to drive yourself deep into the middle of the Redwood Forest, the forest of giant trees not seen anywhere else in the world, with this smart car. Look around. Pick a speck of organic material off the moss of one of the tallest trees. Describe that speck's properties, purpose and potentially lifesaving abilities and speculations. That my friends, is the scientific portion of the INFORM conference to all of us lay people! We are talking details that are deep in the forest, sitting on a speck, resting on moss. Tiny.

This deep- in- the- forest journey has one goal... return with answers from those specks, answers that could increase enzyme levels, develop more effective diagnostics or new genetic treatments for those we love and ourselves.

While enjoying the view outside my smart car at INFORM 2016, here is what I can share with you that should give you reason to be excited and maybe answer some questions. Some of what is discussed at INFORM could be considered overlap from the FOD conference, so I was careful to focus on topic points that really presented with excitement and new information. Enjoy the view!

#### **Resveratrol and Its Promising Treatment Uses**

Chronic, physical cellular "stress" leads to inflammation and can cause cellular death. Treatments are being explored using anti-inflammatory medications and resveratrol, the highly beneficial compound found in red wine. This treatment combination is in the early stages of use but some promising results are already being seen. Resveratrol, on its own also seems to be a useful tool in cellular protection. Timing and dosing are very critical and have not been worked out quite yet. Some counter effects of resveratrol can occur and those effects aren't completely understood yet. When asked why we don't have all patients taking resveratrol immediately based on the exciting results seen thus far, the answer was given in the following analogy. If each mitochondrial cell is like a battery, resveratrol may provide additional energy and increase the batteries life OR it may actually deplete the battery sooner, pulling its energy quicker and thus shortening its overall useful life. It is for this reason I have not purchased resveratrol at each Costco visit and taken it by handfuls. Waiting for more research would be wise. However, its promising results are by far one of the most simple and least invasive treatments discussed at the conference!

### LCHAD 10 Year Study Follow-Up

A 10 year study using C7 oil (CONCURRENT WITH MCT) showed stability in retinopathy, no increase in CK levels and overall, a more clinically stable presentation. CK levels were lower during times of illness. No side effects were reported. The issue of course, remains whether the benefits were derived purely from C7 or it's combination with MCT oil. More studies will need to occur to determine the exact cause. However, this is clearly a promising path for further research.

### Metabolomics, a Booming Field

Metabolomics is a form of personalized medicine offering a way to assess the metabolic state of the individual. Using multiple methods such as acylcarnitine profile, amino acid profile, organic acid levels and measuring phospholipids, a more useful picture is rendered which helps to show connections between variants and their expression in each affected individual. This approach could be most useful during crisis. Downfalls are the time it takes to receive some of these test results. If it takes 2 weeks for a facility to provide results on a specific test, that information can't be used to develop immediate treatment plans. Conversation developed around which laboratories can provide quicker results and how hospitals could develop quicker turnaround times. However, that takes money and with such a small patient base, it is a challenge. These tests aren't exceptionally costly, so that makes the Metabolomics approach a promising new tool in the physicians' hands.

### Cellular MCAD Study Possible

A cellular MCAD study using only cells collected at the same time as Newborn Screening, during an initial crisis or crisis during an in-patient stay is being considered. This study's purpose would be to understand more clearly why affected, symptomatic individuals may have affected, asymptomatic siblings. Is this a coincidence? Is this just plain luck? Could the lack of symptoms be caused by an additional, undiscovered variant that is effecting the expression of MCAD differently in some patients? Clearly, this could be a very beneficial study and might provide a sneak peek into how to support MCAD affected individuals.

Currently, determining when to collect samples is underway, deciding between collection during stability or times of crisis. Samples at diagnosis and during crisis periods seem to be very helpful. A conversation ensued about how to collect further amounts of samples for studies. Many people shared their perspective. As a parent representative, the perspective I shared was from the patient and their family. First, a relationship built on trust needs to exist. To ask more of these families is a lot. It's really that simple. We and our children go through so much already that it is difficult to subject ourselves to further painful experiences. Second, trying to work with other physicians on our team is important. If a procedure including anesthesia is already occurring, it would be important to co-board and squeeze multiple events under one anesthetic event, rather than make the patient endure a biopsy awake and possibly traumatized. Yes, this may be inconvenient for the physician, but it is the patient and their body that matters most. Third, and finally, I explained the **willingness of our FOD community to provide samples, our eagerness for answers and our cooperative spirit when we are treated as a member of the team and our individual needs are valued.** Several professionals thanked me for this input and said it helped them to see the picture differently. I hope these ideas help us all along our path to answers.

### Unexplained MADD-like Symptoms

Think Riboflavin! For people who have unexplained MADD/GA 2 - like symptoms (MADD symptoms with no genetic findings for MADD), Riboflavin Transporter Deficiencies should be explored. RTDs are easily treated with riboflavin. Ask your doctor before adding any additional supplements, but if you or someone you love falls into this narrow category and are struggling for answers, it is worth asking about how riboflavin may be helpful.

### Cardiac Issues and VLCAD

Luckily, almost all cardiac issues associated with VLCAD are reversible. It was said even as late as age 20, cardiac issues can usually be effectively reversed. Most often, cardiac issues have a negative outcome with an individual who is not diagnosed and receives no treatment. One suggestion for treatment with VLCAD presented was to use extremely high levels of glucose concurrent with insulin. This endocrinologic approach seems to be working well during times of crisis and provides much needed levels of glucose while protecting other vital organs. Exploration continues into this additional form of support for VLCAD patients.



## C7 oil Study Update

Dr Gerard Vockley's C7 study is moving into Phase 3. Thus far, there has been a significant reduction in hypoglycemia with the use of C7 oil. There has not been a change in the occurrence of rhabdomyolysis. However, there has been a reduction in the overall severity of rhabdo episodes. More results will continue to be shared over time. The potential benefits of C7 oil continues to be intriguing and potentially life changing!

## SCADD Overall Commentary

Many families with SCADD mutations struggle with how to manage the knowledge of having SCADD and balancing the existing often unexplained symptoms. This quandary is understandable and is not ignored by many physicians. Here are a few important points that were made during a one-on-one conversation with a well-respected physician and researcher during the INFORM conference, specifically about SCADD.

The belief is that if the SCADD mutations are solely linked to SCADD and nothing else, there should be no symptoms. (Don't worry there is more to come!) IF SCADD exists WITH symptoms, then those symptoms are unrelated and/or there is something else causing those symptoms, which likely indicates an additional genetic cause.

According to a summary of commentary on the NORD website, it cannot be eliminated that SCADD plays a role in disease, but it is not a disease itself. The physician explained though, that it is also generally the belief that under exceptional circumstances the SCADD patient could have a true metabolic crisis. This would be an exception and a rarity. Changes in lifestyle do not need to occur but if something "weird" were to occur, the patient must be seen. Things should not be dismissed! It was also suggested that an annual metabolic evaluation was still wise.

An additional, important point that was made was that if the SCADD diagnosed individual does have many other symptoms, it is critical that a full evaluation occur. Other FAOD issues could be present and of course, that would be important to identify. Many FAODs are still not always picked up on NBS like GA2, VLCAD, 'mild' LCHAD and CPT2. Although this does not validate many SCADD patients' concerns, it is comforting to know that physicians do recognize SCADD is not completely benign in some cases. Annual exams, contacting your doctor with abnormal symptoms or considering additional evaluations may help to uncover some SCADD patients' full medical pictures. ***[Note from Deb to our Professionals: What might that further evaluation entail – what do our SCAD Families need to ask their Drs? What further tests should/might be done? How can they also get their own METABOLIC Dr to NOT be dismissive and refuse to see them, which many of them are and do, despite showing symptoms, ever since the 'SCAD is not a disease' perspective began years ago? These are very real questions that our Families need answers to!]***

INFORM continues to be dedicated to uncovering and explaining as much of our "Redwood Forest" as possible. Through worldwide research and collaboration, INFORM and its contributors hope to positively impact the medical treatment and understanding of all FAODs. It is with great respect and gratitude that I have reported some of their useful findings. Many conversations occurred within the larger group that could only be described as hope...hope for more answers, effective treatments and even more positive outcomes.

**We will take this "smart car" journey together.** We may need to rent a few buses!





## Clinical Studies, Results and Articles

**Clinical relevance of short-chain acyl-CoA dehydrogenase (SCAD) deficiency: Exploring the role of new variants including the first SCAD-disease- causing allele carrying a synonymous mutation**

[Open access article](#)

### Clinical Trial for VLCAD, LCHAD, TFP and MCAD Adults:

**Purpose**

“The purpose of this study is to learn more about what causes insulin resistance. It has been suggested that proper breakdown of fat into energy (oxidation) in the body is important to allow insulin to keep blood sugar in the normal range. The investigators want to know if having one of the fatty acid oxidation disorders could have an influence on insulin action. Fatty acid oxidation disorders are genetic disorders that inhibit one of the enzymes that converts fat into energy. The investigators will study both normal healthy people and people with a long-chain fatty acid oxidation disorder.”

**Contacts**

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<a href="mailto:martijul@ohsu.edu">martijul@ohsu.edu</a>	<a href="mailto:gillingm@ohsu.edu">gillingm@ohsu.edu</a>

\*\*\*Please Note: This information was extracted from a longer news release dated Nov 30, 2016:

### 78-week data from the Phase 2 study of UX007 (triheptanoin) in patients with long-chain fatty acid oxidation disorder (LC-FAOD)



#### Patient Assistance Programs through NORD ~ help with medications, clinical trials, etc

If your **Physician** needs more information about the brandname **Levocarnitine - Carnitor®**, dosages, or has other questions, please have him/her contact Sigma-Tau Pharmaceuticals, Inc.. at **1-800-447-0169** or [sigmatauinfo@sigmatau.com](mailto:sigmatauinfo@sigmatau.com).

The liquid and tablet **generic drug brand** for Levocarnitine was approved for distribution by the FDA several years ago. Please note that the generic drug form by **HiTech/Akorn**(as well as the brand name Carnitor®) needs a Prescription from the Dr for the **oral solution** and **tablet**.

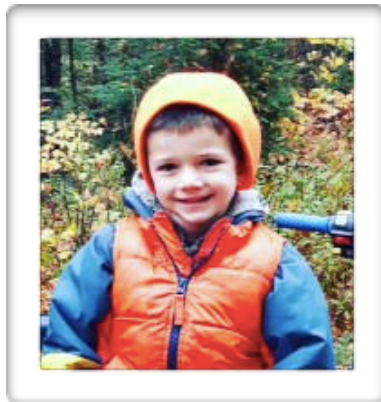
**There has been a major shortage of levocarnitine due to Rising Pharmaceuticals discontinuing its production of the generic drug. Many of our FOD Families have been scrambling to find other ways to obtain their prescriptions. I contacted HiTech/Akorn and their Customer Service rep said that production should be back up to normal by mid-January. So that's good news for ALL!**

# Kids Korner

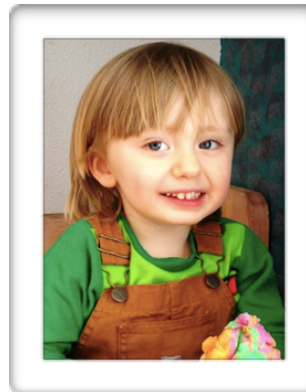
*Our Wonderful kids/adults living with an FOD!  
Please send pics/info to [Deb](#)*



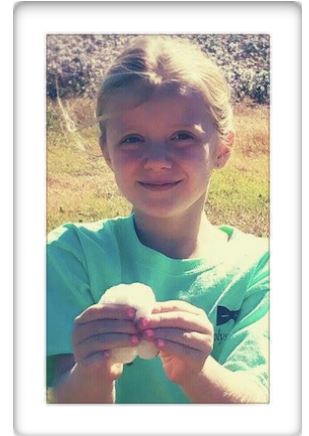
Isabelle ~ Unclassified FOD age 18  
Texas and her service dog, Nani



Briar ~ MCAD age 3  
Ontario Canada



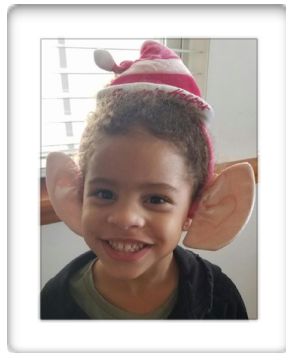
Koltin ~ SCAD age 4  
Oregon



Abby ~ MCAD/Type 1 Diabetes age 6  
Georgia



Dominic ~ GA 2 age 3  
Pennsylvania



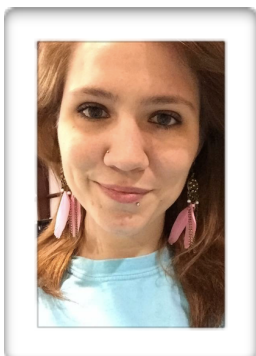
Rory ~ LCHAD age 3  
Florida



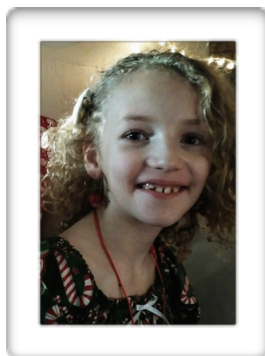
Sophia MCAD age 5 months  
Tennessee



Raylan ~ VLCAD age 3  
Kentucky



Danielle ~ MCAD age 25  
Mississippi



Melody VLCAD age 8  
California



Kade MCAD age 2.5 yrs  
Missouri



Madeline VLCAD age 8  
Texas

# Love Messages



*Please remember our Families in your thoughts and prayers throughout the year...*



Please send thoughts and prayers to Ronnie French and Chelsie French as they walk through this journey with their newest baby, 3 mos old Sophie (VLCAD), June 10, 2016



We have had some child and adult deaths over this past year in our FOD Family...

*All of our FOD children and adults will ALWAYS be with us in our hearts!*

*'Give light, and the darkness will disappear of itself'*

*~ Desiderius Erasmus ~*



## SPECIAL FAMILY ARTICLE

*from those that live in the*

*FOD/Metabolic/Mito world!*

**Note from Deb:** In 'light' of the deaths that some in our FOD Family have experienced this year and from years ago, I wanted to reprint an article I wrote back in 2001 in memory of our daughter, Kristen, who died suddenly in July 1985 from Undiagnosed MCAD, and also inspired by Ryan, one of our FOD kids that had recently died. With the advancement of expanded newborn screening, we are saving more lives by identifying, diagnosing and treating these disorders sooner. However, I wanted to address the concept of 'Hope' that can be transformed over the course of receiving the diagnosis, going through illnesses and hospitalizations, etc that occur throughout the lifetime of a child/adult with an FOD...and for some Families that have tragically experienced a death before a firm diagnosis or after a severe metabolic crisis. I realize that this topic may be uncomfortable for some to read, yet unfortunately many of our Families are living this reality. I am writing this in HONOR of ALL of our Families whether their child/ren or adult child is here with us or is in eternal life.





*T*here is ALWAYS hope...it just may not be or look like we feel and think it SHOULD be!

Many of my articles are fueled by my conversations with parents, just as the ['Find the Light in the Darkness'](#) article was ~ where a professional wasn't going to tell a new FOD Family about our Group because she felt our newsletter was 'too depressing' for the family to read. In other words, this one professional was basically telling this family that it's HOPELESS to find any kind of 'good' support out there because there isn't any at this time! She was making decisions for this family ASSUMING they wouldn't want to read about LIFE and REALITY and all because it was too depressing for HER! Of course, there's support ~ it just doesn't 'fit' what others perceive as supportive ~ which to them may mean, "Let's not talk about death." Trying to protect families FROM reality as well as DENYING them their right to decide for themselves what is supportive is wrong.

Just as wrong is when someone tells a family that, "It's no big deal, the treatment is very simple. We have this all under control. No need to overreact." Yes, some of the treatments vary in their amount of restrictions etc, but what that statement does is dismiss or minimize the seriousness of the disorder, possibly leading families into denial or thinking that they REALLY don't have to be concerned or do anything for treatment. You may think that doesn't happen ~ but it has and it does...some with serious consequences.

Hearing that message from a medical authority, on another subconscious level, also seems to magically 'absolve' parents/grandparents for having passed on a defective gene. That may sound like a bunch of psychobabble but let me tell you, I've spoken to enough families over the last 11 years to hear 'guilt' even when the words aren't actually spoken!

None of us wants to think that we are responsible for our child's disorder. Of course, when you put things in context and realize that **NO ONE knows for sure which genes will be passed on and in which combination, it's totally unrealistic to think we're responsible for knowing that we are carriers** (since diagnosis is often AFTER newborn screening, an episode or a death and BEFORE we know we're carriers!) or even when we DO know, we need to remember that there is MORE of a chance that EACH child WON'T have a disorder (75%) versus having it (25%).

Waiting for some outside entity to tell us it's no big deal or to absolve us of guilt or fear is NOT what HOPE is about. However, dealing with those issues of guilt and fear/stress (discussed in other Healing articles on the website) is a highly PERSONAL PROCESS that takes time and a lot of internal work and it CAN impact how you view hope for the future.

Sometimes that can be done by yourself, but in some cases it may help to talk with a counseling professional if they are causing concerns in your life. This is getting away from my main point but in a strange sort of way, this type of message almost skews hope at the other end of the continuum ~ implying there's hope for your child and future children, because "It's no big deal!"

There's hope all right. It's just NOT because it's "No big deal!"

When a family hears a diagnosis of a rare disorder, the initial hope may be for a cure. However, as more information is gained and realizing that there is no cure at this time, that hope may be readjusted to being able to treat effectively. And depending on the situation, that hope may need further 'readjustment' as time goes by and experiences change.

But what happens to hope when you hear that your child is going to die because there is no effective treatment so "Just go home and enjoy your child while you can." **Statements like this have been made to some of our families** ~ talk about blasting all hope (according to the 'normal' definition/use of the word) for this family into the stratosphere! That professional or any person for that matter may be stating a 'cold fact' based on what few cases have been seen of a very severe form of an FOD, but HOW that 'fact' of inevitable or probable death is conveyed has a HUGE IMPACT on how a family perceives hope!

Having experienced various transformations of hope myself (when we were given Kevin's MCAD diagnosis I/we IMMEDIATELY thought he was going to die suddenly as Kristen had, until we gained more information), I try to compassionately convey to parents that have just been given that heart-wrenching prognosis that hope can be perceived in different ways, for not only the benefit of their child, but for their own present and future coping and dealing with their child's (or other loved ones) condition, as well as death, whenever that may be.

Parents often feel so helpless AND hopeless when they hear a prognosis like that ~ especially when it's given in such a detached and many times unemotional way. Parents have told me that they feel as if the doctors and others have "given up" on the child and family. Now, before you jump all over me about Drs sometimes being detached and cold, I realize that SOME may present that way to really 'protect' themselves from feeling too much and getting attached ~ if they get too attached they might not be able to do the kind of work they do. I understand that thinking ~ however, that still doesn't mean that the content of a devastating message can't be given in a more compassionate and caring way. 'Bedside manner' DOES make a difference!

I'm here to say that despite what the eventual and inevitable 'outcome' MAY be for some, HOPE IS STILL POSSIBLE, yet in a different way. It's not a matter of debating whether knowing ahead of time is 'better or easier' than a sudden death ~ that isn't the point. The point is if you can work through your fear, anxiety, guilt, and other emotions of grief instead of focusing ONLY on the 'inevitable outcome' that was devastatingly placed before you, you can take each precious moment in the PRESENT and cherish it NOW, so those memories will be with you to help you in the FUTURE.

You can also try to make your child as comfortable as possible during this time ~ families have sometimes been 'advised' (directly and indirectly) from having certain procedures done (i.e. gtube) because "it won't do any good" ~ but if it makes your child more comfortable instead of fighting for every ounce of energy or strength for days or weeks or years, than it WILL DO SOME GOOD! If you can embrace your circumstances and your child from THAT perspective, it opens you up for channeling some of your heartache in a more HOPEFUL way...and benefiting you and your child in the process. Hope CAN BE transforming for all involved!

Intellectually acknowledging that yes, a death may happen soon, LOOKS like all hope is gone, but it really isn't. Saying that there's absolutely NO hope is a false statement to me. There may be no hope for an absolute physical cure of some disease, but that doesn't mean that ALL hope in ALL realms is nonexistent!

When you perceive hope 'wholistically' you become more open to other meanings for yourself. By not just 'staying up in your head,' and experiencing what is happening with your mind, body, AND spirit ~ you will SEE how hope can be transformed from the darkest of darkness to a new kind of 'lightness.' It certainly may not look or feel that way the moment you hear a shattering prognosis, but over time, it IS possible if you BELIEVE it's possible. I KNOW firsthand it's possible!

Hope comes in many 'colors.' I happen to be drawn to 'yellow' (as evident by our pamphlet, card, and 'my rose'). For me, it brings me that light and strength when the 'shadings' of hope may not look so bright. Every time I see or think of a small yellow rose, memories of Kristen flood my being and in a strange sort of way I draw energy and HOPE from that. As odd as it may sound, THAT is what the true power of 'mystery' and spiritual connection is all about and it CAN have a transforming effect on the rest of your life, as well as your family's life!

On that note, I'll end this as I began ~ There is ALWAYS hope...it just may not be or look like we feel and think it SHOULD be!

Allow YOUR more enlightened vision of HOPE to transform your own lives.

Deb Lee Gould, Director July 21, 2001 Kristen's 16th 'anniversary' [July 2001 FOD Communication Network Newsletter]

## WELCOME TO NEW BABIES



Rosalie (LCHAD) was born on 31st August 2016 at 2kg, 6 weeks early (due to rapid acute fatty liver of pregnancy and obstetric cholestasis, both linked to LCHAD). Sister to Henry (3, LCHAD) and mum Jessica and father Tim. Rosie also has LCHAD. Doing amazingly well and a perfect way to complete our family (Sydney, Australia)

On December 13th 2016 at 8:01am Paton & Martha Martha (and 4 year old Big Brother Charlie) welcomed Alexander Joseph who weighed 8lbs 13oz and was 21.5 inches long. At 4 days old (thanks to newborn screening) he was diagnosed with MCADD and is doing great!



## RESOURCES

[The Complete Guide to Disability Claims, Insurance and Benefits](#)

[Parent and Educator Resource Guide to Section 504 in Public Elementary and Secondary Schools](#)



# Press Release!

[ From Deb: PatientCrossroads, which helped us begin to build our [FOD Registry](#), was acquired by AltaVoice, and now AltaVoice was just acquired by Invitae. The 'PIN' (Patient Insights Networks) pictures below are graphics for the data collected so far in our FOD Registry. **Please Register so we may help researchers save LIVES!**]

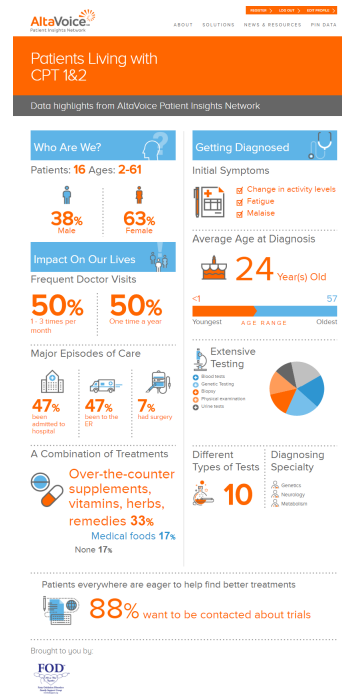
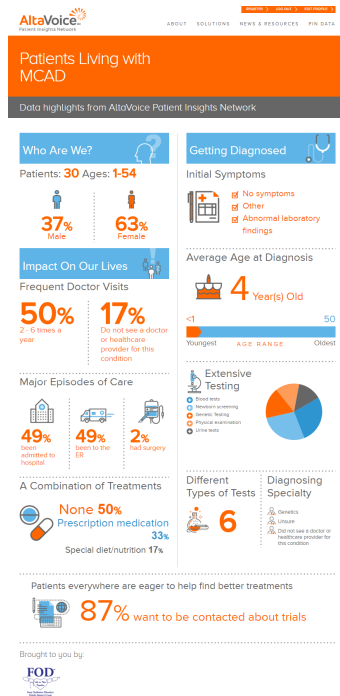
## [INVITAE ACQUIRES PATIENT-CENTERED DATA COMPANY ALTAVOICE, CREATING NEW OFFERINGS TO ADVANCE RESEARCH AND ACCESS TO CARE FOR PATIENTS WITH INHERITED AND RARE DISEASES](#)

[Invitae](#) Corporation's (NYSE: NVTX) mission is to bring comprehensive genetic information into mainstream medical practice to improve the quality of healthcare for billions of people. Invitae's goal is to aggregate most of the world's genetic tests into a single service with higher quality, faster turnaround time, and lower price than many single-gene and panel tests today. The company currently provides a diagnostic service comprising hundreds of genes for a variety of genetic disorders associated with oncology, cardiology, neurology, pediatrics, and other rare disease areas. Additionally, the company has created a Genome Network to connect patients, clinicians, advocacy organizations, researchers, and drug developers to accelerate the understanding, diagnosis, and treatment of hereditary disease.

Invitae's acquisition of patient-centered data company AltaVoice (formerly PatientCrossroads), complemented by several other unique partnerships, expands [Invitae's Genome Network](#), designed to connect patients, clinicians, advocacy organizations, researchers, and therapeutic developers through Patient Insights Networks<sup>SM</sup> (PINs - see FOD Pins below), which **enable organizations to more efficiently build engaged, research-ready patient communities, recruit for trials, educate, and track patient outcomes.**

Invitae and AltaVoice are of the belief that there is strength in numbers when it comes to advocacy. By working together to bring advocacy efforts to the forefront of hereditary diagnoses and treatments, we believe our patients will feel more supported and educated when it comes to their genetics and treatment plan.

Invitae and AltaVoice believe that individuals own and control their genetic and medical information. While we believe that such information is most valuable when voluntarily shared to improve healthcare and advance science and medicine, we will not share any patient's genetic information without their consent. **With this acquisition, patients will continue to own, manage and direct the use of their own genetic data.**





## FOD GROUP FINANCES

### 2015 FOD Group 990 tax return

2016 FOD tax return will be posted by May 2017

The bulk of Expenses are for monthly phone, website fees, supplies, Conferences, 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 Bracelet and Ribbon Sales, Cafepress.com, iGive, Goodsearch, and any donation that isn't specifically designated for the other Funds. Once the Research and Clinical Funds reach a substantial amount (@\$50,000) we will be able to offer grants to clinicians and researchers in the US. No FOD money is used for salaries - we are an ALL Volunteer organization.

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

## THANK YOU DONATIONS SINCE JULY 2016

**Family Donations:** Nicole Lackenby. Deb Bennett. Crystal & Jack Branta. Arnie Messing. Elaine Cavagnaro. Melanie Adams. Diane Schroppe. Amarilis Sanchez. Diane McCarthy. Kendra & Michael Grasseschi. Barb Hollowick. Anonymous. Neva & Ned Asplundh. Kelley V.

**Tshirts, Bracelets, Ribbons, CafePress, GoodSearch browsing, MissionFish/eBay selling, iGive or Amazonsmile.org shopping etc:** Savannah Speer. Amazonsmile.com shoppers. Robert Morales. Toni Ray. eBay iGiving.

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.

**Professional Donations :** Rare Patient Voice. United Way/Truist donation. KPMG donation. Maple City Savings Bank. JM Family Enterprises, Inc/David Bastian. Sandra Der/Fairway Gorge Dragon Flyers. Irene S Scully Fund.

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

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**Mailing lists:** [Erika Wallace](#)

**Website Designer:** [Mary Lingle](#)

**Newsletter consulting:** Brian Gould

**Email/website consultants:** [Mark Heinz](#)

**Website slide shows & Graphic arts:** [Keith Widmann](#)

**FOD/OAA Event Planning:** [Eileen Shank](#)

**Newsletter formatting:** [Sean](#) and [Elizabeth](#) Weitner



### Families and Professionals...

Please send all Submissions to [Deb](#) by **June 15, 2017** for the July 2017 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!

***'My mission in life is not merely to survive, but to thrive; and to do so with some passion, some compassion, some humor, and some style.'***

*~ Maya Angelou ~*

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