The Challenging World of Fatty Oxidation Disorders

By Kelly Huber
ckhuber99@gmail.com
with Deb Lee Gould, MEd
deb@fodsupport.org

Fatty Oxidation Disorders (FODs)[1] can be very challenging for families and affected children and adults (FODers). Personally, I know I have lost sleep over it and gained a few gray hairs! Being an FOD family, you are suddenly thrust into being your own Medical Expert, Nurse, Chef, Psychologist, Coach, and Advocate. In a utopian world, we would never wish this disorder on anyone, but if we have to live with it we challenge society to see our world through an FOD lens.

First, it's important to understand how and why an FOD occurs so we can bring the challenges to the surface. Literally, you need a magnifying glass to find the specific FOD that 'lives' in the mitochondria of the cells. These are sometimes called 'silent disorders' because you cannot look at a person and physically see that they have an FOD. You carry two defective genes, one from each parent, that impacts how specific enzymes breakdown or convert fatty acids in the foods we eat into energy. Each child born to two carriers has a 1 in 4 chance of having the disorder and each child/adult can respond to their disorder in their own unique way - there is no ONE blanket presentation for FODs.

When your body depletes its glycogen stores it has no reserves to thrive on. Individuals, especially when undiagnosed and untreated, may present with low blood sugar and could have symptoms of weakness, lethargy, muscle pains, mood changes, stomach & GI pains, chronic illnesses, vomiting, and seizures among other symptoms. These can occur during a common illness or after participating in rigorous activity. Because many of these symptoms are present in other disorders, it makes the search for a diagnosis even more difficult.

The major difference between an affected and unaffected person is that an FODer can quickly and without warning go from being healthy one minute to having liver or organ failure, seizures, coma and death if not treated immediately due to low intake when ill or fasting. This can present anxiety for FOD families who are constantly on guard for signs that their FODer's body has been compromised by a stressor (ie., illness, activity, heat/cold, surgery).

Even though some of the FODs were discovered 35 years ago, many ER doctors still don't know about them, so when a child or adult presents with low blood sugar they may give them a sugary drink and send them home only to have the family bring them back in more of a crisis! By creating awareness, as in this article, I hope more professionals and laypeople think outside the box when a person presents with the previously listed symptoms and not misdiagnose it as fibromyalgia, chronic fatigue, or 'all in your head.' When some professionals can't find a reason for a person's symptoms, and because of the lack of FOD knowledge, several families, moms especially, have been FALSELY accused of munchausen syndrome by proxy – which means they have been accused of making their child sick! Can you imagine how devastating and infuriating that is for a family when they are only trying to HELP their child!

With expanded newborn screening being mandated in every state (@29 disorders, including some FODs) many lives are saved. However, some still fall through the cracks and aren't picked up through screening. Some that have died before a diagnosis are often labeled as SIDS or Reyes Syndrome. And others that are older and undiagnosed often endure years of a medical odyssey searching for an answer. That diagnostic answer can often come via tandem mass spectrometry and dna methods, blood/urine tests, and skin/muscle biopsies.[2] Having Drs aware of what to look for and how to diagnose can alleviate families' stress over all the uncertainty and lead to finally getting the necessary treatment.

One of the greatest challenges an FOD family experiences after a diagnosis is getting the medical support they need in the midst of varying treatment perspectives across the US and world by Genetic and Metabolic Specialists, as well as not having enough Metabolic Specialists to treat children AND adults. Although all agree that avoiding fasting and eating a high carb/lowfat diet is basically a given, some disagree on adding various supplements (ie., MCT oil) and/or drugs (ie., FDA-approved levocarnitine). This variance of treatment adds to the frustration and stress of families.

Some specialists think there are 'mild' cases so they do not recommend the above supplements or drugs. In my experience there is nothing mild about these disorders! Trying to get a specialist to really understand how you/your child responds to NOT having these treatments (ie., always fatigued, muscle aches) is extremely challenging – often to the point of possibly seeking another medical opinion.

Some may have been prescribed levocarnitine for years and then the Dr discontinues it because he/she doesn't believe it is effective. This puts a huge amount of stress on the family as they have seen the benefits of the drug as far as mood, overall health, less muscle pains, and increased energy. It would be like telling someone with a thyroid deficiency they cannot have their medicine anymore. Some of the FODs also require other medical foods, supplements, or services such as speech/physical therapy that can cause challenges as well. Medical foods (ie., metabolic formulas) are often not covered by insurance so families can be burdened by large out-of-pocket expenses. Some families have accrued many other costs due to frequent hospitalizations, uncovered supplement/medical foods and equipment, hiring extra home care, loss of their own job etc, that they have declared bankruptcy or have lost their homes. Even though our health system is currently going through changes, I believe we can all agree that the high costs and other issues need to be addressed!

Over the years, researchers have become more successful at identifying the disorders, but creating more treatment options is still in its infancy. According to the NIH, in the US a rare disorder is one that impacts fewer than 1 in 200,000 and there are @ 6800 rare diseases[3]. Unfortunately because FODs are rare disorders there is not enough research money and clinical trials around the country – the results of which could help discover new or more effective FOD treatments. In the meantime, families struggle with what their specialist knows or doesn't know. Again, I hope this article creates more awareness of where Drs and others can gain further knowledge of diagnosis and treatment of FODs so everyone is on the same page for the benefit of the child or adult!

Along with the challenges at home, families also deal with school, athletic and work environments. Trying to communicate the seriousness of the disorder but in the context of allowing the child to be

'normal' among the other kids can be a major challenge. Be aware that one can LOOK quite healthy on the outside, but when asked to run the mile in PE class, some FODers may not be able to accomplish that due to not having the 'fuel' available in their cells to make their muscles work. Having to limit or sit out activities or being singled out as 'different' etc, can also lead to bullying experiences, which is another serious issue for everyone involved.

For schools, it's important to have all paperwork up-to-date AND fully complied with. Having special dietary/drug treatments may involve filing a 504 plan for school, which mandates what needs to be done for the child while in school or participating in a sport. Parents also need to provide the Dr's medical letter and emergency protocol so it's on file in case of an emergency.

One of the fears of parents is allowing another adult to 'be in charge' of their child while away from home and them possibly not recognizing the early signs of metabolic crisis. FODers are bright, capable, responsible, good students and workers, but when the FOD comes 'knocking on their door' during certain activities or when one doesn't eat enough or coming down with a cold/fever, they may be wiped out and unable to concentrate (ADHD may be an issue too), their head becomes fuzzy, vision is blurred, muscles start to be compromised and metabolic breakdown occurs.

So PROACTIVE and CLEAR communication with the school and coaches and work colleagues is VITAL. Listening to the CHILD/ADULT is important too – sometimes their speaking up when not feeling well is dismissed as "You're just trying to get out of running" or "Suck it up" as they often say in sports. Not listening could land the person in the ER and it could take them days or weeks to fully recover!

One of my biggest challenges while raising three FOD children is that the schools cannot accommodate the dietary restrictions that are necessary to keep FODers healthy. School meals are often filled with processed or high fat and high sugar foods – although that is slowly changing in some districts. Thus many parents become the Dietitian and make their own lunches/snacks/drinks for their child so they have enough to fuel them throughout the day.

When families have chronically struggled with schools or daycares, they often choose to home school. That way they can monitor the diet and other needs more carefully. But again, it adds another role to a parent's growing list – as teacher. In some ways, that can decrease the stress a bit because they are not constantly fighting with the school. But it all comes down to who and how to best accommodate an FODer's needs.

Living in a fast-paced and competitive society can also be challenging for some with an FOD. Trying to keep up with others can become frustrating and disappointing and lead to other issues, such as low self-esteem. This is NOT always the case, however, but being aware of how those living with a chronic disorder can be stigmatized or made to feel 'less than' others can have a huge impact on psychological and emotional development. Parents are all too aware of that fact.

It takes a lot more planning and understanding from others as families strive to enrich the lives of a person with an FOD. Most parents encourage their children and tell them that they can do anything in life that they put their mind and effort to. It is no different for an FOD parent, except we have to juggle

various realities and help them balance the social fun stuff in life with the not-so-fun stuff ~ the reality of dealing with taking medications, possibly having hospitalizations, etc. However, not all FODers are as restricted in activity or diet – some are able to fully participate in sports and leisure activities, but they do so by learning how their bodies respond to exercise and by keeping power drinks/snacks available throughout the activity. Even children within the same family can differ in their levels of participation and their presentation when ill. So it's important to treat each FODer as an individual and tailor their treatment and activities to fit THEIR needs so as to enhance and not jeopardize their overall physical/emotional/social health and wellbeing.

Living with an FOD is about making CHOICES - It is very much a give and take relationship with the world and somehow by creating FOD awareness, hopefully we can get others to understand FODs so that the individual and family journeys and challenges don't have to feel so isolating. It may be idealistic, but if every household in America had a family member with a chronic disorder, maybe our society wouldn't be as competitive and we could get back to simpler times and maybe we would all be more loving and compassionate…it would be a wonderful reality!

We can't force anyone to understand our challenges and yet we keep striving to raise awareness and help educate others and by doing so, recognize how amazing FOD individuals truly are. FODers are incredibly remarkable people as they learn to live with their rare genetic metabolic disorder and come out ready to tackle each day proving to the world that they can handle any and all of life's challenges. It is the most extraordinary mystery I have witnessed every day in my own family. Of course, I wish and pray each day for a cure, but for today I am thankful for the walking miracles.

References:

1. http://fodsupport.org/description.htm

2. http://fodsupport.org/diagnostic-approach.htm

3. http://www.rarediseases.org/rare-disease-information

Kelly's Bio: Kelly and her husband have three wonderful children with a rare fatty oxidation disorder. She and her husband grew up in Ohio and currently reside in South Carolina. Kelly has her bachelor's in Business Administration and is a full-time working mom in the Aerospace Industry.

The state newborn screening program played a large role in identifying that Kelly's children have a rare genetic condition. She has served on a Consumer Task Force that promotes awareness about newborn screening and rare conditions and continues to travel to hospitals to educate parents. Kelly has also written several articles on FODs.

Besides being an advocate for fatty oxidation disorders, Kelly and her husband play a very active role in their children's lives and community by coaching baseball, robotics and dance teams. She and her family also have a love for books, music, fishing, swimming, watersports, beach time, and food which is so essential to FODs well-being.

Deb's Bio: Deb Lee Gould, MEd, is a bereaved parent since 1985 and a Grief Consultant in Okemos, MI. She is also Co-Founder and Director of an all-volunteer 501c3 nonprofit and international Family Support Group for rare Fatty Oxidation Disorders (www.fodsupport.org), as well as an ordained non-denominational spiritual counselor.

The FOD (Fatty Oxidation Disorders) Family Support Group™ was 'born' in 1991 as a way of dealing with the sudden death of their 21-month old daughter, Kristen, in 1985 from undiagnosed MCAD. Initially, she was 'diagnosed' with Reye's Syndrome and it was not until one year later, when their son, Kevin, was born and diagnosed with MCAD that she discovered Kristen also had this rare metabolic disorder. Their third child, Brian, is a carrier of MCAD.

Along with running the Group, Deb offers pro bono one-on-one grief support (face-to-face or by zoom) to parents and other adult family members and friends living with the death of a child of any age and from any cause. Support is offered early in grief or many years down the road. Deb received her Masters in Counseling in 1993 with a specialization in Grief Counseling. No fees are charged for her support services, however, donations are always appreciated and will benefit the FOD Group.