

FOD FAMILY SUPPORT GROUP

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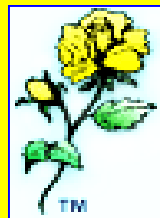


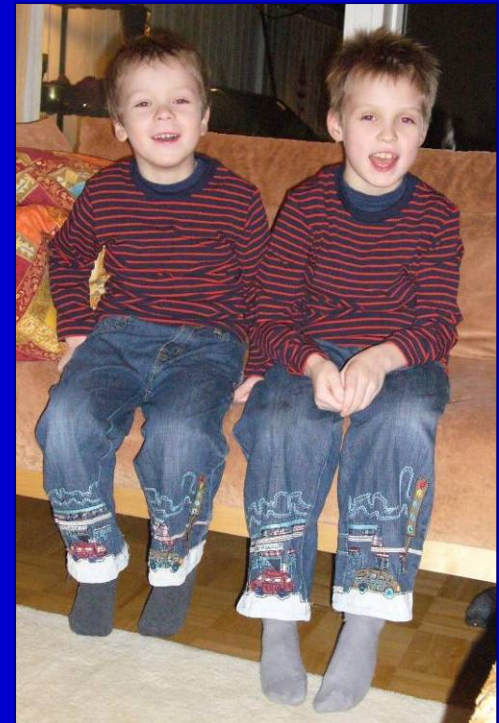
MISSION

- ♥ CREATE AWARENESS & EDUCATE the Public & Professionals about Fatty Oxidation Disorders (FODs)
- ♥ PROVIDE EMOTIONAL, PRACTICAL & GRIEF SUPPORT to Families around the world via the FOD website, Email List & Phone Networking
- ♥ ADVOCATE UNIVERSAL & COMPREHENSIVE NEWBORN SCREENING for ALL Babies

- ♥ INFORM FAMILIES OF NEW DEVELOPMENTS in Screening, Diagnosis, Treatment & Research
- ♥ RAISE FUNDS for CLINICAL TRAINING of New Metabolic Professionals & for FOD RESEARCH Endeavors

'WE ARE ALL IN THIS TOGETHER'





FODs DEFINED

- **Fatty Oxidation Disorders are genetic metabolic deficiencies** in which the **body is unable to break down fatty acids to make energy** because an enzyme is either missing or not working correctly. The main source of energy for the body is glucose and normally when the glucose runs out, fat is broken down into energy. However, that energy is not readily available to children and adults with an FOD.
- **When diagnosed and treated AT BIRTH the prognosis for most of the FODs (i.e., MCAD etc) is excellent.** Most can make adjustments to diet/meds when necessary during times of extra activity and illness and **lead a full life**. However, **if undiagnosed and untreated**, these disorders can **lead to serious complications** affecting the liver, heart, eyes and general muscle development, and possibly death.

- Fatty Oxidation Disorders are **autosomal recessive disorders** affecting both males and females. Both parents are carriers of an abnormal gene, in that when two abnormal genes unite the child will have an FOD. There is a 25% chance that each child will have an FOD and a 75% chance of being a carrier or not having the disorder at all.
- **If one child is diagnosed with an FOD, their siblings should also be tested, even if they are asymptomatic.**
- **One CAN live with an FOD!** There may be various challenges along the way for some individuals with FODs (i.e., especially if diagnosed late, after a severe crisis, etc) but **KNOWING** you have an FOD and **how to treat** on a daily basis and in an emergency is the **KEY!**

SYMPTOMS

- There is a **wide variation in presentation of FODs, even within the same family**. Not every individual responds to a disorder in the same way. Some may be without major symptoms, yet others may have chronic bouts of low blood sugar or illness leading to many hospitalizations, especially during the 1st years of life.
- Please note that **blood glucose levels should not be TOTALLY relied on as the ONLY indicator of a possible 'crisis.'** Do NOT compare FODs to diabetes!
- Changes in behavior, irritability, lethargy, and blood levels such as ammonia, liver enzyme and ck (cpk) should also be monitored. An emergency situation exists when a 'metabolic crisis' occurs. A crisis is often preceded by a period of fasting, possibly due to an ear infection or flu.

- Other symptoms may include vomiting, diarrhea, seizures, coma and difficulty breathing.
- **Awareness of how to diagnose and treat these disorders is vital** ~ during a metabolic crisis, an **undiagnosed** individual may experience excessive buildup of fat in the liver, heart and kidneys, along with some brain swelling...**all of which can lead to death.**
- Those deaths have sometimes been misdiagnosed as **Sudden Infant Death Syndrome or Reye's Syndrome.**



Kristen, Undiagnosed MCAD

Oct 6, 1983 ~ July 21, 1985



Candice, Undiagnosed MCAD

Feb 21, 1991 ~ Nov 8, 1993



DIAGNOSIS of FODs

- These disorders are diagnosed in a variety of ways at **specialized labs across the country** (refer to Medical Info page on www.fodsupport.org).
- Blood, urine, skin fibroblasts, amniocytes (from amniotic fluid) and muscle and liver tissue are some of the specimens analyzed.
- The diagnostic tests include an acylcarnitine profile, urine organic acid analysis, carnitine levels and enzyme assays in fibroblasts. The acylcarnitine profile with whole blood on a 'PKU card' (filtered paper card) is the most direct approach for diagnosis of most of the FODs.
- Molecular DNA testing is also available for several FODs. It is often recommended to test the newborn screen card with whole blood by analyzing the acylcarnitine profile and then the DNA mutation, if the profile is abnormal.

- The expanded Expanded Newborn Screen test may detect or 'red flag' several FODs, as well as many other metabolic disorders.
- However, the **Expanded Newborn Screen is DIFFERENT than the Diagnostic Acylcarnitine Profile test** that is used to specifically diagnose FODs.
- If there is a positive screen, further testing will be suggested (i.e., plasma carnitine levels, urine organic acids, DNA testing, acylcarnitine profile), as well as types of specimens needed (i.e., skin or muscle biopsy, amniocytes, postmortem tissues) to further assist in the diagnostic evaluation of FODs.



SPECIFIC FODs

- Carnitine Transport Defect (Primary Carnitine Deficiency)
- Carnitine-Acylcarnitine Translocase Deficiency (Translocase)
- Carnitine Palmitoyl Transferase I & II (CPT I & II) Deficiency
- Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency
- Long Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency
- Trifunctional Protein (TFP) Deficiency
- Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
- Medium Chain 3-Ketoacyl coA Thiolase (MCKAT) Deficiency
- 2,4 Dienoyl-CoA Reductase Deficiency
- Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency
- Short Chain L-3-Hydroxyacyl-CoA Dehydrogenase (SCHAD) Deficiency - now called 3-Hydroxyacyl-CoA Dehydrogenase (HADH) Deficiency
- Electron Transfer Flavoprotein (ETF) or ETF Dehydrogenase Deficiency (GAII & MADD)
- 3-Hydroxy-3 Methylglutaryl-CoA Lyase (HMG) Deficiency
- Unclassified FODs

TREATMENT FOR FODs

- **Treatment for FODs is multidimensional.**
- **Avoid Fasting.** For **infants under 6 months**, it is suggested to feed every 3-4 hrs around the clock. As children grow, and depending on various factors, most can eventually sleep 10-12 hrs. Of course, these times aren't 'written in stone' and can vary based on the specific disorder, if ill, etc.
- Most children/adults with an FOD **eat often throughout their waking hours (i.e., @every 3-6 hrs)** ~ and more often if ill. If well, they can sleep up to 10-12 hrs at night.
- A fasting state, especially while ill, can trigger a 'metabolic crisis.' If hospitalized, it is **imperative** that a **10% dextrose IV (5% is NOT enough) is started immediately** ~ **waiting hours for lab results can be fatal** when an FOD child/adult is in crisis.

- A high carb/lowfat diet is recommended throughout the day. Additionally, some use **MCT Oil** for fats that can be utilized (not used for all FODs, especially MCAD and SCAD, since the medium chain triglycerides cannot be broken down). Hydrate with caloric drinks (i.e., gatorade) in heat and during exercise.
- Some physicians prescribe the **FDA-approved drug Carnitor®** or the generic drug Levocarnitine as an L-carnitine supplement for several FODs, **especially if carnitine deficiency is a secondary disorder.**
- Although carnitine is made within the body, sometimes supplementation is necessary. L-carnitine helps the body's metabolism run smoothly, is useful in preventing low blood sugar, and assists in removing metabolic wastes from the cells so a buildup of toxins will not occur.

NEWBORN SCREENING ADVOCACY

- Imagine if **ALL babies** across this country/world were **UNIVERSALLY, UNIFORMLY, and COMPREHENSIVELY screened** as newborns ~
- Families would not have to experience the **heartache of a child's death** due to lack of diagnosis **or live with major medical complications** because the baby/child went into a severe crisis before diagnosis!
- **Why WAIT for a medical crisis to occur** ~ please Promote Expanded Newborn Screening so ALL families can know from BIRTH what they and their child/children are living with!

Labs that perform Expanded NBS and other Diagnostic testing:
[more Labs are listed on www.fodsupport.org, Medical Info page]

Institute of Metabolic Disease: Dallas, TX
(NBS phone) 214.4.BAYLOR (Diagnostics phone) 214.820.4533

Mayo Medical Lab: Rochester, MN
(NBS and Diagnostics phone) 800.533.1710



**Support Expanded
Newborn Screening**





EXPANDED NEWBORN SCREENING SAVES BABIES' LIVES!