FOD FAMILY SUPPORT GROUP
Deb Lee Gould, MEd, Director

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

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 FOD 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.
♥ Three deaths have sometimes been misdiagnosed as Sudden Infant Death Syndrome or Reye’s Syndrome.

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**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 acids, 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.
- Treatment for FODs is multidimensional.
- Avoid fasting. For infants under 6 months, if it is triggered to feed every 2-3 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 it is.
- Most children/adults with an FOD disorder, if ill, etc. ‘red flag’ several FODs, as well as many other metabolic disorders.

**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
- Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
- Short Chain 3-Hydroxyacyl-CoA Dehydrogenase (SCAD) Deficiency
- Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
- Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency
- 2,4 Dienoyl-CoA Reductase Deficiency
- Medium Chain 3-Ketoacyl CoA Thiolase (MCKAT) Deficiency
- Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
- Trifunctional Protein (TFP) Deficiency
- Long Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency
- Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency
- Carnitine Palmitoyl Transferase I & II (CPT I & II) Deficiency
- Carnitine-Acylcarnitine Translocase Deficiency (Translocase)
- Carnitine Transport Defect (Primary Carnitine Deficiency)

**TREATMENT FOR FODs**

- Treatment for FODs is multidimensional.
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**NEWBORN SCREENING ADVOCACY**

- 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 crisis state before diagnosis.
- Why WAIT for a medical crisis to occur – please Promote Expanded Newborn Screening so ALL families can know from the moment their child/children are living with a compromised health condition.

Expanded Newborn Screen test may detect or rule out several FODs, as well as many other metabolic disorders.

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**EXPANDED NEWBORN SCREENING SAVES BABIES’ LIVES!**