

## To Request More Information:

Please answer the following questions and mail to Deb at the address below.

I am a:  FOD Family  FOD Familymember  Professional

Check all that apply:

/my child has an FOD

Please add me to your Mailing List Only

### Comments:

\_\_\_\_\_  
Name and/or Professional Title

\_\_\_\_\_  
Address 1

\_\_\_\_\_  
Address 2

\_\_\_\_\_  
Phone [Deb calls new Families in the US and abroad]

\_\_\_\_\_  
Fax

\_\_\_\_\_  
Email

#### Please mail to:

**FOD Family Support Group  
Deb Lee Gould, MEd, Director  
PO Box 54  
Okemos, MI 48805**

The FOD Family Support Group is an all volunteer family-based Support Group and a 501c3 tax-exempt Non-Profit Corporation. **Donations are tax deductible.** Your donation will help us cover copying, postage, website fees, phone calls to US and abroad, and Conference expenses, as well as for Clinical/Research Funds. No donations are used for administrative salaries. If you write a US check, please make it out to the 'FOD Family Support Group' and mail to the address above. We also have a secure PayPal donation link on our homepage. You can designate your donation to the General FOD Fund (daily operations), FOD Clinical Fund, FOD Research Fund or Grief Consults. **THANK YOU from ALL of our Families!**

## For Families Living with a Fatty Oxidation Disorder



### Fatty Oxidation Disorders Family Support Group

www.fodsupport.org



Deb Lee Gould, MEd, Director  
MCAD Parent and Grief Consultant  
PO Box 54  
Okemos, MI 48805

Phone: 517.381.1940

Fax: 866.290.5206

Email: deb@fodsupport.org  
Website: www.fodsupport.org

501c3 tax-exempt Non-Profit Corporation  
Federal Tax ID # 83-0471342

# FOD Family Support Group

## Our History and Mission

The Fatty Oxidation Disorders (FOD) Family Support Group was 'born' in 1991 as a way of dealing with the sudden death of our 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 our son, Kevin, was born and diagnosed with MCAD that we discovered Kristen also had this rare metabolic disorder. Our third child, Brian, is a carrier and not directly affected. To say the least, our Family has been changed forever by this genetic disorder.



*Our hearts  
give immortality to those  
we love in memory...*

Our Mission is clear...to connect and network with FOD Families and Professionals around the world. Through our online newsletters, website, phone calls, and Email/facebook List, we provide emotional support, practical information about living with these disorders, and inform Families of new developments in screening, diagnosis, treatment, and research.

Awareness of these disorders is imperative for early diagnosis and treatment, prompting us to be committed advocates for Expanded Universal and Comprehensive Newborn Screening and longterm follow-up care for FODs and several other metabolic disorders. We do not want other Families to needlessly go through what we experienced.

*From our Family to yours, we want you to know...*

**You are NOT alone  
and  
'We Are All in This Together'**

**Take care...**

**Deb and Dan**

**Kristen, Kevin and Brian Gould**



## FODs Defined

Fatty Oxidation Disorders are genetic metabolic deficiencies in which the body is unable to oxidize (breakdown) fatty acids to make energy because an enzyme is either missing or not working correctly. The main source of energy for the body is a sugar called glucose. Normally when the glucose runs out, fat is broken down into energy. However, that energy is not readily available to a child or adult with an FOD. If undiagnosed and untreated, these disorders can lead to serious complications affecting the liver, heart, eyes, and general muscle development, and possibly death. These disorders are sometimes misdiagnosed as SIDS and Reye's Syndrome.

## Symptoms

There is a wide variation in the 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 illnesses leading to hospitalizations. Please note that **blood sugar levels should not be totally relied on as the only indicator of a possible crisis** - the levels may even look normal. However, some of our children/adults are symptomatic with glucose levels of even 70 or 80mg/dl. Do NOT compare FODs with diabetes! Changes in behavior, irritability, lethargy (sleepy), and blood levels such as liver enzymes, acylcarnitines, ammonia, 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 vomiting or an infection. Other symptoms may include diarrhea, seizures, coma and difficulty breathing.

Awareness of how to diagnose and treat these disorders is vital, because 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.

## Diagnosis and Treatment

Fatty Oxidation Disorders are autosomal disorders affecting both males and females. Both parents have to be at least a carrier of an abnormal gene (we do have parents with the disorder as well), 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 either 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.

These disorders are screened for and/or diagnosed in a variety of ways at specialized Labs across the country (refer to our website). Blood, urine, skin fibroblasts, amniocytes (from amniotic fluid), and muscle and liver tissue are some of the specimens analyzed. The diagnostic tests often 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 some FODs when the profile is abnormal. The Expanded Newborn Screen is a different test than the diagnostic profile. The bloodspot screen can 'red flag' 50+ different disorders, including many FODs.

**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!**

Treatment for FODs is multidimensional. A major concern is to **avoid fasting**. It is strongly recommended that infants under 6 months be fed around the clock every 3-4 hours. Older children or adults with an FOD eat often throughout their waking hours (i.e., every 3-6 hrs) and can sleep 10-12 hrs, when well. The feeding interval may depend on various factors and their specific circumstances, such as if they are ill versus well, in which case they most likely would eat more often around the clock. Please note that eating timeframes can vary from person to person within the different disorders.

A fasting state, especially while ill, can trigger a 'metabolic crisis' leading to lethargy and hospitalization. If **hospitalized**, it is imperative according to FOD experts, that a **10% dextrose IV is started immediately** following blood chemistry samplings. **Waiting hours for the lab test results before putting in the IV can be fatal** when an FOD child/adult is in crisis.

[**Note:** Experts also recommend avoiding aspirin, oral products that contain salicylates, long term steroid medications, lactated ringers, and fat binding/producing anesthesia medication].

Several snacks and meals of lowfat/high carbohydrate foods such as pasta, cereal, and other high complex carbohydrates (as well as sugar drinks when not well) are recommended throughout the day. Additionally, some Families use MCT Oil (not used for all FODs - i.e., MCAD and SCAD, since the medium chain triglycerides cannot be broken down). Hydrate with caloric drinks such as Gatorade while in heat and during exercise. Infants under 6 months should continue to have at least one night feed/snack as they should not go 10-12 hours without eating. If the FOD child/adult has experienced hypoglycemia (even with frequent meals), in order to help decrease the frequency of low blood sugar in the morning, some find it helpful to mix in 1-3 tablespoons of raw cornstarch (i.e., Argo) to a cold sugar-free liquid or food at night. Be aware that too much cornstarch can be constipating for some and may be hard on teeth. Be sure to brush after eating or drinking it. Also note that cornstarch should not be given to an infant under age one (they lack the necessary pancreatic amylase to digest the cornstarch).

Some physicians may also prescribe Levocarnitine (FDA-approved Carnitor® or an FDA-approved generic form—these are NOT the same as the healthfood store carnitine), especially if a secondary carnitine deficiency is involved. Although carnitine is made within the body, sometimes supplementation is necessary. It is useful in stabilizing blood sugar levels, increasing muscle tone, and removing metabolic waste products from the cells so a buildup of toxins will not occur.

As with any medical condition, discussing treatment options with your physician(s) and FOD specialists is necessary for the well-being of anyone with an FOD. Please do **NOT** make any changes in your child's or your own treatment without first consulting with your physicians.

### **Early Screening, Diagnosis and Treatment will prevent needless deaths**

#### **Disorders of Fatty acid Metabolism**

Medium Chain acyl coA Dehydrogenase Deficiency (MCAD), Medium Chain 3-Ketoacyl coA Thiolase Deficiency (MCKAT), Medium/Short Chain L-3 Hydroxy acyl coA Dehydrogenase Deficiency (M/SCHAD), Long Chain L-3 Hydroxy acyl coA Dehydrogenase Deficiency (LCHAD), Very Long Chain acyl coA Dehydrogenase Deficiency (VLCAD), Short Chain acyl coA Dehydrogenase Deficiency (SCAD), 3-Hydroxy acyl coA Dehydrogenase Deficiency (HADH, formerly called SCHAD), Short Chain 3-Ketoacyl coA Thiolase Deficiency (SKAT, 3-ketothiolase), Electron Transfer Flavoprotein or Electron Transfer Flavoprotein ubiquinone-oxidoreductase Deficiency (ETF and ETF-QO, also known as GAI1 or MADD), Carnitine Palmitoyltransferase Deficiency (CPT I & II), Trifunctional Protein Deficiency (TFP), Carnitine Acylcarnitine Translocase Deficiency (CACT), Carnitine Transport Defect (Primary Carnitine Deficiency), 2,4-Dienoyl coA Reductase Deficiency, 3-Hydroxy 3-Methylglutaryl Deficiency (HMG), Unclassified FODs

## Additional Metabolic Family Support Resources

### Organic Acidemia Association (OAA)

Kathy Stagni, Director  
9040 Duluth Street  
Golden Valley, MN 55427  
763.559.1797 phone  
<http://www.oaanews.org>  
kstagni@oaanews.org

### United Mitochondrial Disease Foundation (UMDF)

8085 Saltzberg Road, Suite 201  
Pittsburgh, PA 15239  
412.793.8077 or toll free 888.317.8633 phone  
412.793.6477 fax  
<http://www.umdf.org>  
info@umdf.org

### Children Living with Inherited Metabolic Disease (CLIMB)

Climb Building  
176 Nantwich Road  
Crewe CW2 6BG  
United Kingdom  
0800 652 3181 phone  
0845 241 2174 fax  
<http://www.climb.org.uk>  
Info.svcs@climb.org.uk

Expanded NBS & FOD Diagnostic Testing:  
[more Labs are listed on [www.fodsupport.org](http://www.fodsupport.org), Medical Info page]

### Mayo Medical Laboratories (Rochester, MN)

Mayo Clinic Biochemical Genetics Laboratory  
800.533.1710 phone for NBS testing, Diagnostic testing, and Consultation  
507.284.1759 fax  
<http://www.mayoclinic.org/departments-centers/laboratory-medicine-pathology/minnesota/laboratory-genetics/overview/biochemical-genetics-laboratory>

### National Newborn Screening and Global Resource Center (NNSGRC) (Austin, TX)

512-921-1400 phone  
<http://genes-r-us.uthscsa.edu/>

### Sigma-Tau Pharmaceuticals, Inc.

Makers of Carnitor®  
9841 Washingtonian Blvd, Suite 500  
Gaithersburg, MD 20878  
800.447.0169 phone  
<http://www.sigmatau.com>  
sigmatauinfo@sigmatau.com

### FOD Family Support Group Board of Directors

Deb Lee Gould, MEd  
FOD Parent, Director, and President

Daniel R Gould, PhD  
FOD Parent and Treasurer

Mary Lingle  
FOD Parent, Webmaster, and Secretary

### Honorary Directors

Kevin Gould, MCAD Adult  
Brian Gould, MCAD Sibling

### Board of Medical Advisors

Charles R Roe, MD  
Consultant  
Dallas, TX

Mark Korson, MD  
Genetic Metabolic Center for Education  
Salem, MA

Jerry Vockley, MD, PhD  
Children's Hospital of Pittsburgh  
Pittsburgh, PA

Stephen G Kahler, MD  
University of Arkansas for Medical Sciences  
Little Rock, AR

Gary Siskin, MD  
Albany Medical College  
Albany, NY

Andrew Morris, PhD, FRCPCH  
Royal Manchester Children's Hospital  
Manchester, UK

Arnold Strauss, MD  
Cincinnati Children's Research Foundation  
Cincinnati, OH

Barb Marriage, PhD, RD  
Ross Metabolics/Abbott Nutrition  
Columbus, OH

Janice Fletcher, MD, FRACP, FRCPA  
Women and Children's Hospital  
North Adelaide, Australia

James Gibson, MD, PhD  
Specially for Children  
Austin, TX



**You are not alone...**