



State of CT
Genetics Newborn Screening Program
Fatty Acid Oxidation Disorders (FAOD) Family Fact Sheet

Fatty acids are a component of fat in the food we eat and from fat in our body tissues. Oxidation is the process that breaks down fatty acids to release energy needed for body functions. Each step of the oxidation process is set in motion by a specific enzyme. Fatty acid oxidation disorders occur when one of these enzymes is missing. 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, the fat source is not readily available to children and adults with an FAOD. If undiagnosed and untreated, these disorders can lead to serious complications affecting the liver, heart, eyes and general muscle development, and possibly death.

Medium-Chain ACYL-CoA Dehydrogenase Deficiency (MCADD) is an example of a fatty acid oxidation disorder. With this disorder the body cannot properly break down fats into sugars to use for energy. When babies and children with MCADD become ill or experience long periods of fasting, blood sugars become dangerously low and they are at risk of having a "metabolic crisis". A metabolic crisis can lead to seizures, failure to breathe, cardiac arrest and death, and/or result in serious brain damage. However, screening can provide a diagnosis before the baby becomes ill. The most important thing is to avoid fasting (going without feeding) your baby for long periods of time and to call your doctor if your baby gets sick.

The following are the Fatty Acid Oxidation Disorders* screened for in CT:

MCADD (Medium-Chain ACYL-CoA Dehydrogenase Deficiency)	Carnitine Palmitoyltransferase Deficiency (CPT I)
LCHADD (3-Hydroxy Long-Chain ACYL-CoA Dehydrogenase Deficiency) or Trifunctional Protein Deficiency	Glutaric Acidemia Type II or Multiple Acyl-Coa Dehydrogenase Deficiency (MADD)
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD)	Carnitine Transporter Deficiency
Carnitine / acylcarnitine translocase Deficiency (CACT OR CAT)	Carnitine Palmitoyltransferase Deficiency II (CPT II)

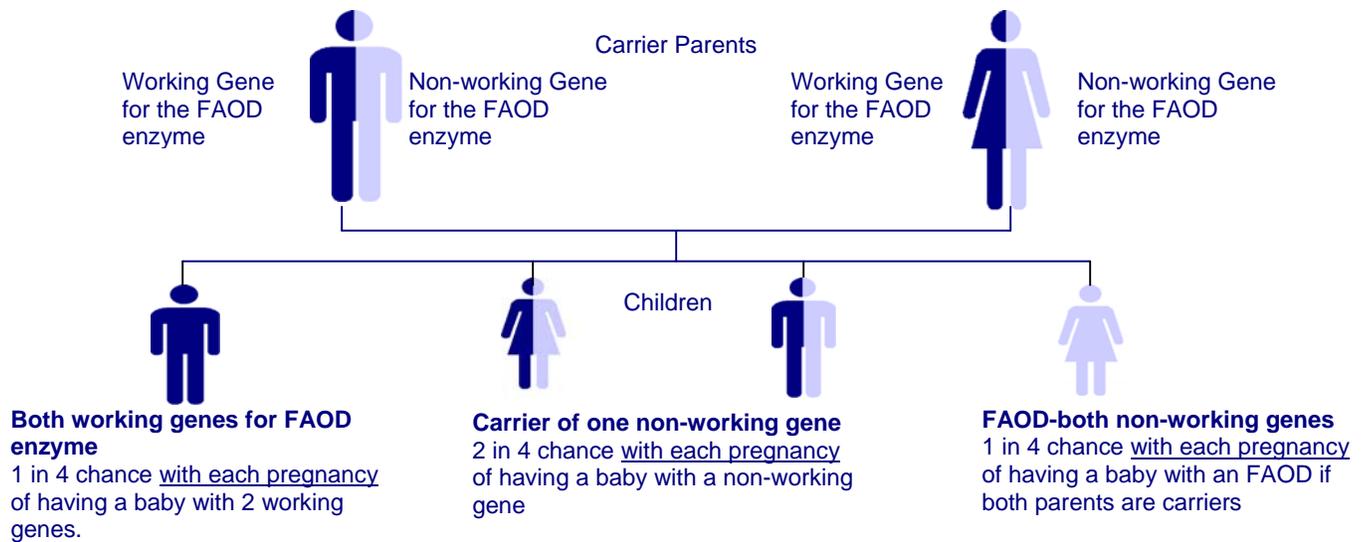
***Specific information on the individual diseases is available in the "Health Professional Fact Sheets" on the Department of Public Health Website. (<http://www.dph.state.ct.us/BCH/NBS/Resources/Resources.htm>)**

How is a baby tested for an FAOD?

Before going home from the facility of birth, a small sample of blood is taken from the baby's heel. The sample is then sent to the State Laboratory to screen for FAOD's. The Family Health Division nurse consultant notifies the baby's primary care provider of any positive screening results. Your baby's doctor will call you if the results are not normal. The doctor will discuss with you the need for more testing. Further testing and follow-up treatment if necessary is done at one of the Genetic Regional Treatment Centers at Yale or the UCONN Health Center.

How does a baby get an FAOD?

FAOD's are inherited conditions. Parents pass the changed gene for the FAOD to their children the same way they pass on other inherited traits. A FAOD can occur if both the mother and the father have a non-working gene for a certain enzyme. As shown in the figure below, a baby can receive a non-working gene from each parent and this will result in a FAOD. If both parents have a non-working gene for an FAOD, they have a one in four chance with each pregnancy of having a baby with an FAOD.



What is the treatment for an FAOD?

Babies with one of these disorders have trouble burning fat for energy. This can lead to drowsiness, poor tone, vomiting, low blood sugar, liver failure, and muscle problems or more serious problems such as coma. Treatment includes avoiding fasting, a special low fat diet, a medication called carnitine and other supplements. A baby with one of these diseases must have regular medical care at a metabolic treatment center.

The most important thing you can do is to avoid fasting (going without feeding) your baby for long periods of time. Be sure the time between feedings does not exceed 4 hours for your newborn and 6 hours for older babies. If your baby becomes sick with fever, vomiting, or diarrhea, (anything that could cause dehydration), be sure to call your baby's doctor right away. Special dietary supplements are often added to the baby's diet to help prevent problems.



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The information provided is offered for general informational and educational purposes only. It is not offered as and does not constitute medical advice. In no way are any of the materials presented meant to be a substitute for professional medical care or attention by a qualified practitioner, nor should they be construed as such. Contact your physician if there are any concerns or questions.