



Genetics Newborn Screening Program Family Fact Sheet

FATTY ACID OXIDATION DISORDERS (FAOD)

MEDIUM-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (MCADD)

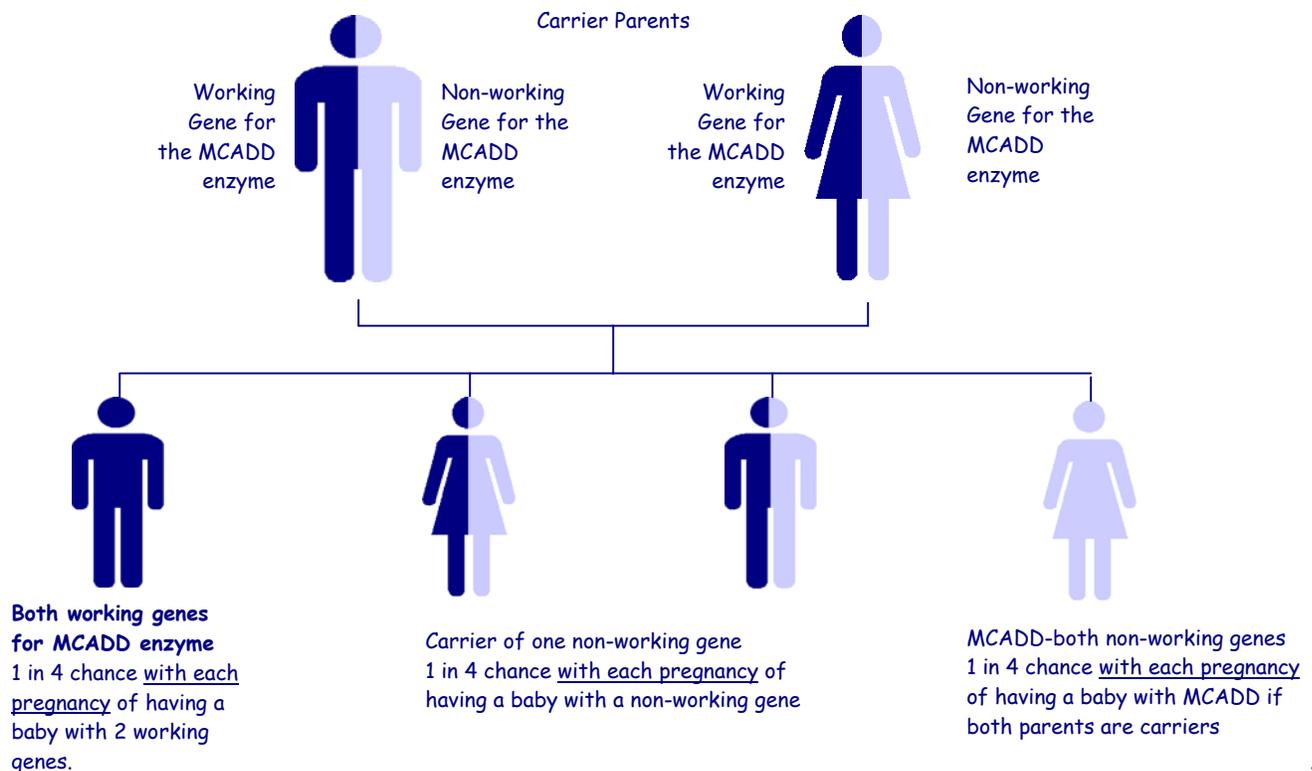
Fatty acids are a component of fat in the food we eat and from fat in our tissues. Oxidation is the process that breaks down fatty acids to release energy needed for body functions. Each step of the oxidation process is set in motion by a specific enzyme. Fatty acid oxidation disorders occur when one of these enzymes is missing.

MCADD is a disorder of fatty acid metabolism. 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, so parents can prevent the fasting periods, and know when to seek early medical care, to prevent crisis.

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 (fever, vomiting, diarrhea) 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.

How does a baby get MCADD?

MCADD is an inherited condition. Parents pass the MCADD genes to their children the same way they pass on other inherited traits. MCADD can occur if both the mother and the father have a non-working gene for the MCADD enzyme. As shown in the figure below, a baby can receive a non-working gene from both parents and have MCADD. If both parents have a non-working gene for MCADD, they have a one in four chance with each pregnancy of having a baby with MCADD.



How is a baby tested for MCADD?

Prior to discharge 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 MCADD. Abnormal screening results are reported to the Department of Public Health (DPH), Genetic Newborn Screening Tracking Unit nurse consultant. If the results are positive, the DPH nurse consultant will notify the baby's primary care physician of the need for a prompt referral to one of the Genetic Regional Treatment Centers at Yale or the UCONN Health Center for confirmation testing and follow-up treatment if necessary.

What is the treatment for MCADD

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 MCADD must have regular medical care at a metabolic treatment center.

Regional Metabolic Treatment Centers

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