



Genetics Newborn Screening Program Family Fact Sheet

FATTY ACID OXIDATION DISORDERS (FAOD)

Long Chain Hydroxy ACYL-CoA Dehydrogenase Deficiency (LCHADD)

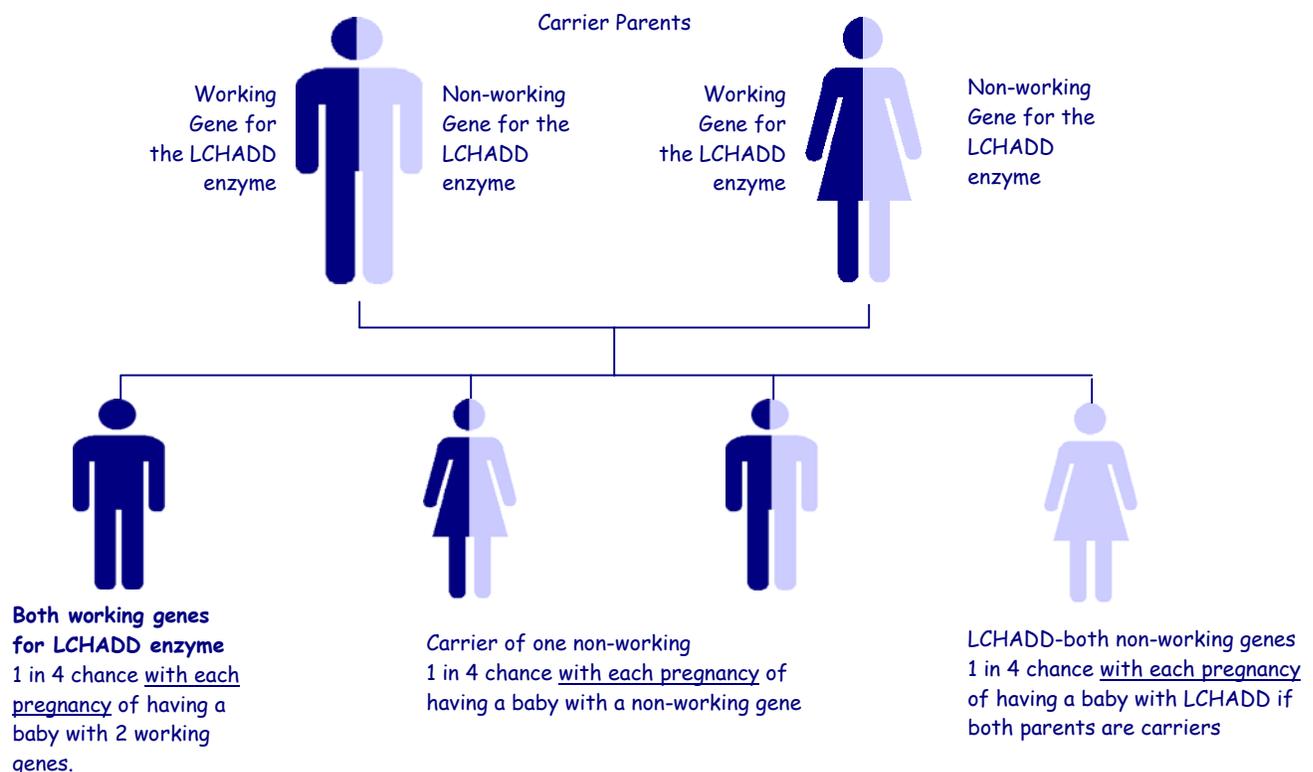
Fatty acids are a component of fat in the food we eat and from fat in our tissues. Oxidation is the process that breaks down fatty acids to release energy needed for body functions. Each step of the oxidation process is set in motion by a specific enzyme. Fatty acid oxidation disorders occur when one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

LCHADD 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 LCHADD become ill or experience long periods of fasting (going without feeding), 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, newborn screening can provide a diagnosis before the baby becomes ill. Early diagnosis leads to early treatment. Parents will learn to seek early medical care, ways to avoid fasting, and when to seek medical care to minimize crisis.

Infants and children with LCHAD require special low fat, high carbohydrate diets and other dietary supplements. 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.

How does a baby get LCHADD?

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



How is a baby tested for LCHADD?

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 LCHADD. 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 LCHADD

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, high carbohydrate diet, a medication called carnitine and other supplements. A baby with LCHADD must have regular medical care at a metabolic treatment center.

Regional Metabolic Treatment Centers

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