

## Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency LCHAD (long-chain 3-hi-drok-cee ay-sill-co-A dee-hi-drodge-in-ace de-fish-in-sea)

### What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called LCHAD (ell-chad). There is another very rare disorder called trifunctional protein deficiency (TFP) that can cause this problem too.

**A positive newborn screen does not mean your baby has LCHAD or TFP, but it does mean your baby needs more testing to know for sure.**

Your baby's doctor will help arrange for more testing by specialists in disorders like LCHAD and TFP.

### What is LCHAD?

LCHAD affects an enzyme needed to break down fats in the food we eat so they can be used for energy and growth. In LCHAD, the enzyme used to break down fats is missing or not working properly.

A person with LCHAD doesn't have enough enzyme to break down fat into energy. Using stored fat for energy is especially important in between meals when the baby is not getting new energy from eating food.

LCHAD is a disorder that is passed on, or inherited, from a child's mother and father. Because LCHAD is a genetic disease, family members are at risk of having LCHAD too, even if no one in the family has had it before.

### What problems can LCHAD cause?

LCHAD is different for each child. Some children with LCHAD have fewer health problems, while other children may have very serious complications.

If LCHAD is not treated, a child might develop:

- Low blood sugar
- Sleepiness
- Vomiting
- Behavior changes (such as crying for no reason)
- Seizures
- Coma

It is very important to follow the doctor's instructions for testing and treatment.

### What is the treatment for LCHAD?

LCHAD can be treated. The treatment is life-long. Treatment for children with LCHAD can include:

- Frequent meals/snacks and a low fat, high carb diet - a dietician will help you learn what foods can be eaten
- Medications to help the body make energy and get rid of harmful toxins
- Careful treatment of routine illness

Children with LCHAD should see their regular doctor, a doctor who specializes in LCHAD, and a dietician.

Children with LCHAD and TFP can benefit from prompt and careful treatment.

### Resources for parents

#### Genetics Home Reference

<http://ghr.nlm.nih.gov>

#### Fatty Acid Oxidation Support

[www.fodsupport.org](http://www.fodsupport.org)

#### MN Children & Youth with Special Health Needs

(651) 201-3650 or (800) 728-5420

#### MN Newborn Screening Program

[www.health.state.mn.us/newbornscreening](http://www.health.state.mn.us/newbornscreening)