LONG-CHAIN L-3-HYDROXY ACYL-DEHYDROGENASE DEFICIENCY (LCHAD) TRIFUNCTIONAL PROTEIN DEFICIENCY (TFP)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (LCHAD). There is another disorder that can cause a similar positive result on newborn screening. This disorder is very rare and is called trifunctional protein deficiency (TFP).

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.

You will be notified by your primary care provider or the newborn screening program to arrange for additional testing.

What are LCHAD and TFP?

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

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

LCHAD and TFP are genetic disorders that are passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause LCHAD or TFP. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can LCHAD and TFP cause?

LCHAD and TFP are different for each child. Some children with LCHAD or TFP have few health problems, while other children may have very serious complications.

If LCHAD and TFP are 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 and TFP?

LCHAD and TFP can be treated. Treatment is life-long and can include:

- Frequent meals/snacks and a low fat/high carbohydrate diet – a dietitian will help you learn what foods can be eaten
- Medications to help the body make energy and get rid of harmful toxins
- Special approaches to routine illnesses

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

Prompt and careful treatment helps children with LCHAD and TFP live the healthiest lives possible.

Michigan Resources and Support

Michigan Newborn Screening
Nurse Consultant
Toll-free: 1-866-673-9939
newbornscreening@michigan.gov

Michigan Metabolic Coordinating Center
C.S. Mott Children's Hospital, Michigan Medicine:
1-734-764-0579

Children's Special Health Care Services
Toll-free: 1-800-359-3722

MDHHS Michigan Department of Health & Human Services | Michigan Newborn Screening

Phone 1-866-673-9939
www.michigan.gov/newbornscreening
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