Carnitine Palmitoyl Transferase 1 Deficiency *(CPT1)*

MICHIGAN Newborn Screening

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 carnitine palmitoyl transferase 1 deficiency (CPT1).

A positive newborn screen does not mean your baby has CPT1, 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 problems can CPT1 cause?

CPT1 is different for each child. Some children with CPT1 have few health problems, while other children may have a more severe form of CPT1 with serious complications.

If CPT1 is not treated, a child might develop:

- Low blood sugar
- Seizures
- Enlarged liver
- Muscle weakness
- Vomiting
- Poor feeding

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

What is CPT1?

CPT1 affects an enzyme needed to break down fats in the food we eat, so they can be used for energy and growth. In CPT1, the enzyme used to break down fats is missing or not working properly.

A person with CPT1 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.

CPT1 is a genetic disorder that is passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause CPT1. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What is the treatment for CPT1?

CPT1 can be treated. Treatment is life-long and can include:

- Frequent meals/snacks and a low fat/high carbohydrate diet - a dietician will help you learn what foods can be eaten.
- Special approaches to routine illnesses.

Children with CPT1 should see their regular doctor, a doctor who specializes in CPT1, and a dietician.

Prompt and careful treatment helps children with CPT1 live the healthiest lives possible.

Michigan Resources and Support

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

Children's Hospital of Michigan Metabolic Clinic
Toll-free: 1-866-442-4662

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

MDHHS Michigan Department of Health & Human Services
RICK SNYDER, GOVERNOR | NICK LYON, DIRECTOR

Michigan Newborn Screening Follow-up
Phone 1-866-673-9939
www.michigan.gov/newbornscreening
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