



Carnitine Palmitoyl Transferase 2 Deficiency (CPT2) / Carnitine Acylcarnitine Translocase Deficiency (CACT)

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 2 deficiency (CPT2). There is another disorder that can cause a similar positive result on newborn screening. This disorder is very rare and is called carnitine acylcarnitine translocase deficiency (CACT).

A positive newborn screen does not mean your baby has CPT2 or CACT, 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 CPT2 and CACT?

CPT2 and CACT affect an enzyme needed to break down fats in the food we eat, so they can be used for energy and growth. In CPT2 and CACT, the enzyme used to break down fats is missing or not working properly.

A person with CPT2 or CACT 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.

CPT2 and CACT 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 CPT2 or CACT. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can CPT2 and CACT cause?

CPT2 and CACT are different for each child. Some children with CPT2 or CACT have few health problems, while other children may have a more severe form of CPT2 or CACT with serious complications.

If CPT2 and CACT are not treated, a child might develop:

- Low blood sugar
- Breathing problems
- Liver problems
- Heart problems
- Seizures
- Muscle pain and weakness

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

What is the treatment for CPT2 and CACT?

CPT2 and CACT 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.
- Medications to help the body make energy.
- Special approaches to routine illnesses.

Children with CPT2 and CACT should see their regular doctor, a doctor who specializes in CPT2 and CACT, and a dietician.

Prompt and careful treatment helps children with CPT2 and CACT 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