TEXAS Department of State Health Services

Newborn Screening FACT Sheet

Carnitine Acylcarnitine Translocase Deficiency (CACT)

What is Carnitine Acylcarnitine Translocase Deficiency?

Carnitine acylcarnitine translocase deficiency (CACT) is a condition in which the body is unable to break down certain fats. It is considered a fatty acid oxidation condition because people affected by CACT are unable to break down some of the fats they eat into energy the body needs to function. This can cause too many unused fatty acids build up in the body. Detecting the condition early and beginning treatment can help prevent some of the severe outcomes of CACT.

What causes CACT?

When we eat food, enzymes help break it down. Some enzymes help break down fats into their building blocks, called fatty acids. Other enzymes break down these fatty acids. In CACT, the enzyme carnitine acylcarnitine translocase (CAT) is not working correctly.

CAT's job is to help bring fatty acids into the mitochondria. Mitochondria are the energy-making factories of cells. When CAT is not working correctly, fatty acids cannot get inside of the mitochondria. This prevents fatty acids from being broken down for energy. Fatty acids are important sources of energy for the heart, especially when the body runs low on sugar (such as in between meals).

When fatty acids cannot be broken down, it also causes high levels of acid in the blood. Everyone has some acid in the blood, but too much acid can be toxic.

CACT is an autosomal recessive genetic condition. This means that a child must inherit two copies of the non-working gene for CACT, one from each parent, in order to have the condition. The parents of a child with an autosomal recessive condition each carry one copy of the non-working gene, but they typically do not show signs and symptoms of the condition. While having a child with CACT is rare, when both parents are carriers, they can have more than one child with the condition.

What Symptoms or Problems Occur with CACT?

[Symptoms are something out of the ordinary that a parent notices]

There are two main forms of CACT, which differ by their age of onset: CACT in newborns and CACT in children. The signs of CACT are the same for these two forms.

Signs of CACT include:

- sleeping longer or more often
- weak muscle tone (known as hypotonia)
- behavior changes (such as crying for no reason)
- irritability
- poor appetite
- fever / diarrhea / vomiting
- low blood sugar (known as hypoglycemia)
- trouble breathing
- seizures (epilepsy)

Many of these signs may occur when your baby eats foods that his or her body cannot break down. They can be triggered by long periods of time without eating, illnesses, and infections.

If your baby shows any of these signs, be sure to contact your baby's doctor immediately.

What is the Treatment for CACT?

Dietary Treatment - Your baby may need to be on a restricted diet in order to avoid certain foods that his or her body cannot break down. A dietician can help you plan a healthy diet for your baby. Eating often can also help avoid many of the signs mentioned in the Symptoms or Problems section.

Supplements and Medications - Medium Chain Triglyceride oil supplements are a common treatment for CACT.

Your Doctor might also prescribe L-carnitine supplements. L-carnitine is a substance naturally made by the body, but your baby might not make enough of it.

Things to Remember

Even minor illness can lead to a Metabolic Crisis in children with CACT. Call your doctor right away when your child has any of the Symptoms or Problems:

Children need extra fluids and starchy food (such as bread, rice, cereal, noodles) when they're sick in order to prevent a Metabolic Crisis. Sick children with CACT may need to be treated in the hospital to avoid serious health problems.