

When Baby Needs A Second Test for a Fatty Acid Disorder Elevated C16 with Elevated C18:1

A small sample of your baby's blood was collected soon after birth and sent to the DHEC laboratory for testing. This testing is called Newborn Screening. In SC, newborns are tested for several genetic and chemical disorders. Some times, a second test is needed to help your doctor decide if your baby has one of these disorders. In many cases, the second test will be normal. However, if your baby does have one of the newborn screening disorders, early treatment will give him or her the best chance to grow up healthy.

Because compounds called C16 (palmitoyl carnitine) and C18:1 (oleyl carnitine) were high in your baby's first test, he or she could possibly have a Fatty Acid Disorder called Carnitine Palmitoyltransferase II Deficiency (CPT II) or Carnitine/Acylcarnitine Translocase Deficiency (CACT).

What are fatty acid disorders?

Fatty acid disorders like carnitine palmitoyltransferase II deficiency (CPT II) and carnitine/acylcarnitine translocase deficiency (CACT) are genetic disorders that are found in a few babies born each year. Some of these disorders can be identified when compounds called acyl carnitines are measured in a baby's blood. When a baby has a fatty acid disorder, he or she cannot use fat for energy. Most of the time our bodies use a sugar called glucose to give energy to the body's systems. Some of the ways our bodies get glucose are:

- ▶▶ By eating foods that have glucose as a part of their make-up (like breast milk and formula for babies)
- ▶▶ By changing stored fats and proteins in our bodies into glucose

If we become sick and are not able to eat very much for a while, we turn our fat stores into glucose for energy until we are able to eat again. Babies with a fatty acid disorder are not able to turn fat into glucose. So when their bodies run out of glucose, they get sick very fast. They may have seizures, be very hard to wake up, or have problems breathing. Babies with a fatty acid disorder may get sick when they have an illness that makes them not want to eat. Almost any childhood illness can cause a baby to eat less than usual, like a cold, an ear infection or a virus. Stomach viruses that cause vomiting and/or diarrhea are a real concern in babies with a fatty acid disorder.

How will I know if my baby really has CPT II or CACT?

If your baby's newborn screening result showed very high C16 and C18:1 levels, he or she probably has CPT II or CACT. The newborn screening test will be repeated and additional tests will be done to help the doctors figure out if your baby has CPT II or CACT. Usually the results of these tests take a few days to come back. You will also be referred to a doctor who specializes in these kinds of disorders.

What do I need to do until I know the final results?

CPT II or CACT can cause your baby to get very sick. Even minor illness can be a medical emergency in a baby with CPT II or CACT. Follow your doctor's instructions very carefully. You will need to make sure your baby eats at least every four hours including through the night. If your baby is hard to wake up, refuses to eat, or seems to be getting sick, your doctor may have you call 911 for emergency help.

How is CPT II or CACT treated?

CPT II and CACT are treated by making sure that the baby doesn't go a long time between feedings. Sometimes the baby will have to be woken up during the night to eat. The baby's diet may have to be lower in fat. A special kind of fat called MCT Oil may be used to help the baby get enough calories. A dietitian will help the family learn which foods the baby can eat.

What else should I do to keep my baby as healthy as possible?

Follow your doctors' instructions carefully! Babies with CPT II or CACT can get sick very fast when they are not able to eat their usual diet or when they have any illness. Almost any childhood illness can cause a baby to eat less than usual, like a cold, an ear infection or a virus. Stomach viruses that cause vomiting and/or diarrhea are a real concern in babies with CPT II or CACT.

Treating illnesses before they become serious and following your baby's diet plan carefully are the best things you can do to help your baby grow and develop.