

When Baby Needs A Second Test for an Organic Acid Disorder Elevated C3 and C5OH

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 the compounds C3 (propionyl carnitine) and C5OH (3-hydroxy isovaleryl carnitine) were high in your baby's first test, he or she could possibly have an Organic Acid Disorder called Holocarboxylase Synthetase Deficiency (Multiple Carboxylase Deficiency).

What are organic acid disorders?

Organic acid disorders like holocarboxylase synthetase deficiency (HSD) 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 an organic acid disorder, he or she cannot rid the blood of certain waste products that occur during the metabolism of protein. These waste products build up in the baby's blood and can cause the baby to have feeding problems, seizures or even go into a coma.

How will I know if my baby really has HSD?

If your baby's newborn screening result showed very high C3 and C5OH levels, he or she probably has HSD. The newborn screening test will be repeated and additional tests will be done to help the doctors figure out if your baby has HSD. 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?

HSD can cause your baby to get very sick. Even minor illness can be a medical emergency in a baby with HSD. Follow your doctor's instructions very carefully. 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 HSD treated?

A special vitamin called biotin is given daily to babies with HSD.

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

Follow your doctors' instructions carefully! Make sure that you keep all well baby checkups and get your baby's shots on time.