

When Baby Needs A Second Test for an Organic Acid Disorder Elevated C5

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 a compound called C5 (isovaleryl carnitine) was high in your baby's first test, he or she could possibly have an Organic Acid Disorder called Isovaleric Acidemia (IVA) or 2-Methylbutyryl co-A Dehydrogenase Deficiency (2-MBCD). Because 2-MBCD is very rare, this handout will focus on IVA.

What are organic acid disorders?

Organic acid disorders like isovaleric acidemia (IVA) 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 IVA?

If your baby's newborn screening result showed very high C5 levels, he or she probably has IVA. The newborn screening test will be repeated and additional tests will be done to help the doctors figure out if your baby has IVA. 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?

IVA can cause your baby to get very sick. Even minor illness can be a medical emergency in a baby with IVA. 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 IVA treated?

Babies with IVA are treated with a special diet. At first, they must be fed a special formula. When they begin to eat solids, the protein in their diet will need to be controlled. They will still need to drink their special formula to make sure they get everything they need to grow properly. A dietitian will help the family learn which foods the baby can eat. Supplements called glycine and carnitine are also used.

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

Follow your doctors' instructions carefully! Babies with IVA 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 IVA.

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.