

When Baby Needs A Second Test for Maple Syrup Urine Disease (MSUD) Elevated VAL or LEU + ILE

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 VAL (valine) or LEU + ILE (leucine + isoleucine) was high in your baby's first test, he or she could possibly have a disorder called Maple Syrup Urine Disease (MSUD).

What is Maple Syrup Urine Disease (MSUD)?

MSUD is a genetic disorder that is found in a few babies born each year. MSUD can be identified when compounds called amino acids are measured in a baby's blood. Amino acids are the "building blocks" of protein. When a baby has MSUD, he or she cannot use some of the amino acids or parts of the protein that are found in foods including breast milk and infant formula. These amino acids form other compounds that 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 MSUD?

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

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

MSUD is treated with a special diet. At first, babies 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.

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

Follow your doctors' instructions carefully! Babies with MSUD 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 one of these disorders.

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.