

When Baby Needs a Second Test for Biotinidase Deficiency

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 the 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 an enzyme called biotinidase was very low in your baby's first test, he or she could possibly have a disorder called Biotinidase Deficiency.

What is Biotinidase Deficiency?

Biotinidase deficiency is a genetic disorder that is found in a few babies born each year. When a baby has biotinidase deficiency, he or she cannot use biotin, a vitamin that is found in foods, including breast milk and infant formula. Without biotin, the baby will not grow and develop properly. He or she may have seizures, hearing loss and skin problems.

How will I know if my baby really has Biotinidase Deficiency?

If your baby's newborn screening result showed very low biotinidase, he or she probably has biotinidase deficiency. The newborn screening test will be repeated and additional tests will be done to help the doctors figure out if your baby has biotinidase deficiency. The results of these other tests may take a while 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?

Your baby will probably not have any symptoms at first, but you will need to follow your doctor's instructions very carefully. If your baby seems to be getting sick, call your doctor right away.

How is Biotinidase Deficiency treated?

Treatment for babies with biotinidase deficiency is fairly simple. Biotin pills can be crushed and mixed with the baby's milk each day. Sometimes biotin comes in capsule form. These capsules can be opened and the contents mixed with the baby's milk too. As the baby begins to eat solid foods, the biotin can be mixed into the foods are given to the baby instead of the milk. Once the baby is old enough to swallow pills or capsules, the biotin will not have to be mixed with anything. Babies with biotinidase deficiency will always need to take biotin all of their lives.

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

Don't forget to keep all of your well baby check-ups! Seeing the doctors regularly and giving your baby biotin every day are the best things you can do to help your baby grow and develop.