

PARENT INFORMATION for inconclusive newborn screening result for BIOTINIDASE DEFICIENCY

When your baby was born a small sample of blood was taken from your baby's heel to test for certain diseases. These diseases usually cannot be found simply by looking at your baby, so that is why the blood test is so important.

The results of your baby's screening test were inconclusive for a disorder called Biotinidase deficiency (BYE-o-TIN-I-days). It is important to remember this is only a screening test. A new blood sample will be collected from your baby and a new test will be done to help make a diagnosis.

What does an inconclusive screen mean?

A small percent of all babies screened are identified as inconclusive on screening. Most are later found not to have the disorder. This is just a "screening" test so further testing should be done to find out if your child has this disorder or not. Keep in mind that the new test could show that your baby does not have Biotinidase Deficiency.

What is the next step?

Be sure to follow your baby's doctor's instructions for getting a new blood sample for further testing, as soon as you receive them. In the meantime, before the results of the additional test are available, your doctor will talk with you about your baby's current health. Biotinidase deficiency is a rare metabolic disease, so your baby's doctor might consult with or refer you to a pediatric metabolic specialist.

What is biotinidase deficiency?

It is an inherited disease. It causes a problem in the use of the vitamin biotin. The body uses biotin in many ways. In order for the body to be able to use biotin, it must be broken down first. Babies who are missing the enzyme biotinidase need more biotin than is normally found in the diet. The effects of not having enough usable biotin could include nerve and brain damage, seizures and mental retardation.

Finding out early if your baby has Biotinidase Deficiency means you and your baby's doctor can take steps to prevent these effects! The treatment is simple and includes giving a prescribed dose of the vitamin biotin.

What should I do?

The most important thing you can do is to be sure that your baby goes in for a new blood sample to be collected and tested as soon as possible. Secondly, report to your baby's doctor immediately, if your baby should show any signs of illness such as fever, vomiting, or diarrhea.

If treatment is ordered, it is very important to follow the doctor's instructions. Now is also a good time to be sure your baby is covered by your health insurance provider.

Remember this could be a false alarm, but if it turns out your baby does have biotinidase deficiency, the treatment is very effective and simple.

For more information talk with your baby's doctor or you can contact the Nebraska Newborn Screening Program at (402) 471-0374 for information on Pediatric Metabolic Specialists available in Nebraska.