

PARENT INFORMATION about POSITIVE Newborn Screening Results for BIOTINIDASE DEFICIENCY

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

The results of your baby's screening test were "presumptive positive" for a disorder called Biotinidase deficiency (BYE-o-TIN-I-days). These results are from a screening test. A new blood sample will be collected from your baby for a different test. This is done to help find out if your baby has biotinidase deficiency or not.

What does a positive screen mean?

A small number of all babies are identified as positive on screening. These babies are more likely to have biotinidase deficiency. With further testing, some are found not to have the disease.

What is the next step?

Be sure to follow your baby's doctor's instructions for getting a new blood sample for further testing. Biotinidase deficiency is a rare disease, so your baby's doctor might consult with or refer you to a metabolic specialist.

What is biotinidase deficiency?

It is a disease that causes the body to have a problem using the vitamin biotin. In order for the body to use biotin, it must be broken down. Babies who are missing the enzyme biotinidase need more biotin than is normally found in their food. The effects of not having enough biotin could include nerve and brain damage, seizures and mental retardation.

Finding out early if your baby has Biotinidase Deficiency means steps can be taken to prevent these effects. The treatment is easy 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 has a new blood sample collected without delay. Also, you should report to your baby's doctor immediately, if your baby shows any signs of illness such as fever, vomiting, or diarrhea.

If treatment is ordered, it is important to follow the doctor's instructions.

Now is also a good time to be sure your baby is covered by your health insurance.

Remember this could be a false alarm. If your baby does have biotinidase deficiency, the treatment is very effective.

For more information about biotinidase deficiency, talk to your baby's doctor. You can contact the Nebraska Newborn Screening Program at (402) 471-0374 for information on Metabolic Specialists in Nebraska.

Internet information is available at <http://www.dhhs.ne.gov/nsp/> and at <http://ghr.nlm.nih.gov/condition/biotinidase-deficiency>

