PARENT INFORMATION about INCONCLUSIVE newborn screening result 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 just by looking at your baby. That is why the blood test is very 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 for a new test.

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 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 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 metabolic disease, so your baby's doctor might consult with or refer you to a metabolic specialist.

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 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 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. If your baby does have biotinidase deficiency, the treatment is very effective.

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

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

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