

What is Biotinidase Deficiency?

Biotinidase Deficiency is a rare disorder in which the body cannot process the vitamin biotin in a normal manner.

Biotinidase is an important enzyme in the body. When the body lacks biotinidase, signs and symptoms of the deficiency occur, such as: hair loss; developmental delay; poor muscle tone, skin rash, poor balance, redness of the eye, drowsiness, seizures, and coma.

What causes Biotinidase Deficiency?

It is inherited from both parents. It cannot be “caught” like a cold and cannot be “given” to another child.

How is Biotinidase Deficiency found?

Shortly after birth, several drops of blood are taken from a baby’s heel. The dried blood sample is sent to the State Department of Health and Senior Services’ Inborn Errors of Metabolism Laboratory, where screening tests are done for biotinidase deficiency and several other conditions. These tests are all part of the State’s Newborn Screening

Program. If there is any abnormality, the baby’s doctor and parents are notified.

What does the lab look for?

The lab tests the baby’s blood for the enzyme biotinidase. People with biotinidase deficiency do not have enough, or almost no, biotinidase.

Does an abnormal screening test mean that my baby has Biotinidase Deficiency?

Screening tests always need to be confirmed by additional testing and medical evaluation.

IF YOU ARE ASKED TO HAVE YOUR
BABY RE-TESTED, ACT QUICKLY AND
FOLLOW YOUR DOCTOR’S ADVICE

What will my baby’s doctor do?

The doctor may refer you and your baby to a metabolic/genetic specialist. The doctor and specialist will determine whether your baby has biotinidase deficiency by doing one or more of the following:

- Get a complete medical history and do a physical exam;
- Look for symptoms;
- Get another blood sample to confirm the screening results and determine the diagnosis. Parental blood samples may be required to better understand your baby’s results.

How is Biotinidase Deficiency treated?

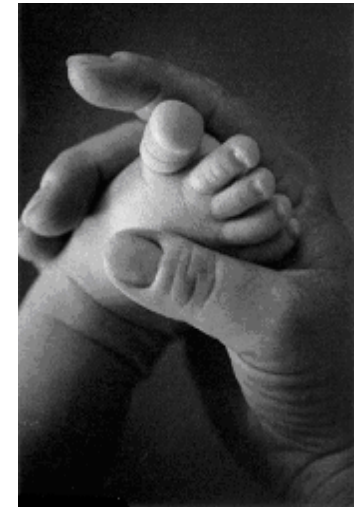
Children with biotinidase deficiency need to take the vitamin biotin by mouth every day for life. Biotin capsules must be prescribed by a doctor—they cannot be bought “over the counter”. Infants who are treated before symptoms appear are not expected to develop health problems from the disorder.

For more information, please contact:

The New Jersey Department of Health
and Senior Services

Newborn Screening and Genetic
Services at (609) 292-1582

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**Important Information
For Parents**

