

# Biotinidase Deficiency Fact Sheet for Parents

All babies born in North Carolina are screened at birth for certain medical conditions that can be treated to prevent serious illness. This is called newborn screening. The newborn screening result showed that your baby might have Biotinidase Deficiency (bye-o-tin-uh-dase). Your baby will be referred to a specialist for more testing to know for sure.

*There may not be signs of Biotinidase Deficiency in some newborns, although it may show up later.*

## What is Biotinidase Deficiency?

Biotinidase deficiency is a rare inherited disorder that affects your child's ability to break down certain foods to create energy. A child with biotinidase deficiency cannot properly use the vitamin biotin, which is essential for many enzymes that create energy in the body.

Early treatment can prevent symptoms of biotinidase deficiency.

## What are the symptoms of Biotinidase Deficiency?

Individuals with biotinidase deficiency may have a range of symptoms, or signs of the disease. Newborns with biotinidase deficiency often have no symptoms at birth. Symptoms usually appear after the first few weeks or months of life. Symptoms of untreated individuals may include:

- Lack of energy (lethargy)
- Poor feeding
- Decreased muscle tone (hypotonia)

Untreated children with biotinidase deficiency may have the following additional symptoms:

- Seizures
- Hair loss (alopecia)
- Skin rash
- Rapid breathing (hyperventilation)
- Temporary stoppage of breathing (apnea)

## What happens next?

First, your baby's doctor will examine your baby for symptoms. Next, the doctor will order more testing to determine whether your baby has biotinidase deficiency. Your doctor may recommend that you give your baby biotin while waiting for the results of this test.

If your baby is confirmed to have biotinidase deficiency, your family will be referred to a specialist to help with your child's care. The specialist will explain your child's treatment options.

## How is Biotinidase Deficiency treated?

Children with biotinidase deficiency need to take a daily biotin supplement for their entire life. The goal of this treatment is to provide enough biotin to restore normal enzyme function.

## Where do I go for more information?

Use your smart phone's camera to scan the QR codes below.



**Baby's First Test:** <https://www.babysfirsttest.org/newborn-screening/conditions/biotinidase-deficiency>



**Video:** <https://www.youtube.com/watch?v=cIRS9vJWEpA>



**Medline Plus:** <https://medlineplus.gov/download/genetics/condition/biotinidase-deficiency.pdf>



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