

Overview of Newborn Screening for Biotinidase Deficiency – For Parents

What is newborn screening?

Before babies go home from the nursery, they have a small amount of blood taken from their heel to test for a group of conditions. One of these conditions is **biotinidase deficiency**. Babies who screen positive for biotinidase deficiency need more tests done to confirm they have biotinidase deficiency. **Not all babies with a positive newborn screen will have biotinidase deficiency.**

What is biotinidase deficiency?

Biotinidase deficiency is a condition that changes the way a person's body uses a vitamin called **biotin**. A person with **biotinidase deficiency** has low levels of an **enzyme** (a protein that helps our bodies function) called **biotinidase**. Without biotinidase, a person cannot use the biotin normally found in food. Biotin is important because it helps our bodies make certain fats and carbohydrates (sugars) and break down proteins.

What causes biotinidase deficiency?

Biotinidase deficiency is an **inherited** (passed from parent to child) condition. Everyone inherits two copies of the biotinidase gene (one from our fathers and one from our mothers). Sometimes these genes have changes (also called mutations) that prevent the gene from working correctly.

In order for a person to have biotinidase deficiency, he or she must have two biotinidase gene changes. People with one biotinidase gene change do not have biotinidase deficiency.

What are the symptoms of biotinidase deficiency?

Every child with biotinidase deficiency is different. Most babies with biotinidase deficiency will look normal at birth. Symptoms of biotinidase deficiency can appear shortly after birth, or they may show up later in childhood. Common symptoms of profound (severe) biotinidase deficiency include:

- Skin problems
- Hair loss
- Hearing and/or vision problems
- Seizures
- Weak muscles
- Developmental delay

What is the treatment for biotinidase deficiency?

There is no cure for biotinidase. However, there is a treatment that can help with the symptoms. Children who have biotinidase deficiency will need to take extra biotin for the rest of their lives. With early diagnosis and treatment, most children with biotinidase deficiency will have few symptoms and have a normal life expectancy.

What happens next?

Although there is no cure for biotinidase deficiency, good medical care makes a difference. Children with biotinidase deficiency should see a Metabolic Geneticist (a doctor who specializes in biotinidase deficiency and other related conditions) as well as their pediatrician. Your child's doctor will work with the Metabolic Geneticist to coordinate any treatment, tests, or appointments that your child needs.

Where is Indiana's Metabolic Genetics Clinic?

Indiana's Metabolic Genetics Clinic is located at Riley Hospital for Children in Indianapolis. You can reach the Metabolic Genetics Clinic by calling (317) 274 – 3966.

Where can I get more information about biotinidase deficiency?

- **National Library of Medicine Genetics Home Reference** - <http://ghr.nlm.nih.gov/condition/biotinidase-deficiency>
- **Region 4 Genetics Collaborative** - http://region4genetics.org/family_resources/gc_biotinidase.aspx