

## **Biotinidase Deficiency**

### **What is biotinidase deficiency?**

Biotinidase deficiency is an inherited condition that affects the way a person's body uses the vitamin, biotin. Biotin is an important vitamin that helps [enzymes](#) called carboxylases make certain fats and carbohydrates and break down proteins. Biotin is essential for proper growth and development. A person with biotinidase deficiency cannot use the bound biotin found in food. This means that the biotin is not available for use. Low levels of biotin may cause seizures, developmental delay, hearing loss and other serious and sometime life threatening illness.

About one baby in 61,000 is born with biotinidase deficiency in the United States. The condition occurs in all ethnic groups.

### **How does biotinidase deficiency affect a child?**

Infants with biotinidase deficiency appear normal at birth, but develop serious symptoms after the first few weeks or months of life. Symptoms include low muscle tone, seizures, developmental delay, loss or absence of hair, hearing loss and optic nerve atrophy. These symptoms can become serious enough to lead to coma and death. With early diagnosis and treatment, all symptoms can be prevented.

### **What causes biotinidase deficiency?**

Biotinidase is a genetic condition caused by changes in the BTD (Biotinidase) [gene](#). The BTD [gene](#) is responsible for making the [enzyme](#) called biotinidase. Biotinidase frees the bound biotin in protein. This free biotin can then help carboxylases make fats, carbohydrates, and break down protein. When there is an alteration in the BTD [gene](#), biotinidase levels go down and the free biotin is too low.

Biotinidase deficiency is inherited in an [autosomal recessive](#) pattern, which means two copies of the BTD [gene](#) must be changed for a person to be affected with biotinidase. Most often, the parents of a child with an autosomal recessive condition are not affected because they are "[carriers](#)", with one copy of the changed BTD [gene](#) and one copy of the normal BTD [gene](#).

When both parents are [carriers](#), there is a one-in-four (or 25 percent) chance that both will pass a changed BTD [gene](#) on to a child, causing the child to be born with the condition. There also is a one-in-four (or 25 percent) chance that they will each pass on a normal BTD [gene](#), and the child will be free of the condition. There is a two-in-four (or 50 percent) chance that a child will inherit a changed BTD [gene](#) from one parent and a normal BTD [gene](#) from the other, making it a [carrier](#) like its parents. These chances are the same in each pregnancy for these parents.

### **Is there a test for biotinidase deficiency?**

Yes. Babies are tested (newborn screening) for biotinidase deficiency before they leave the hospital. The baby's heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to find out if it has less than a normal amount the biotinidase [enzyme](#).

### **Can biotinidase deficiency symptoms be prevented?**

Yes. Generally, taking a daily dose of free (or unbound) biotin can prevent the symptoms of biotinidase deficiency. However, hearing problems may occur in spite of treatment. Treatment should begin as soon as possible following a diagnosis and will continue throughout an

individual's life. Children and adults with biotinidase deficiency require follow-up care at a medical center or clinic that specialize in this condition. In addition, regular blood tests are used to monitor your child's health.

**DISCLAIMER:** The information contained on this page is not intended to replace the advice of a genetic metabolic medical professional.

**Resources:**

Biotinidase Deficiency: A Booklet for Families and Professionals  
[www.ccmckids.org/research/biotinidase/](http://www.ccmckids.org/research/biotinidase/)

MUMS National Parent-to-Parent Network  
Julie J. Gordon  
150 Custer Court  
Green Bay, Wisconsin 54301-1243  
Phone: 1-877-336-5333 (Parents only please)  
Phone: 1-920-336-5333  
Fax: 1-920-339-0995  
E-mail: [mums@netnet.net](mailto:mums@netnet.net)  
[www.netnet.net/mums/](http://www.netnet.net/mums/)

Association for Neuro-Metabolic Disorders  
5223 Brookfield Lane  
Sylvania OH 43560-1809  
Phone: 419-885-1497  
E-mail: [VOLK4OLKS@aol.com](mailto:VOLK4OLKS@aol.com)

**References:**

- GeneTests (Biotinidase Deficiency) <http://www.geneclinics.org>
- Online Mendelian Inheritance in Man (OMIM topics 253260) <http://www.ncbi.nlm.nih.gov/Omim>
- Scriver, C.R. and Kaufman, S (2001) Disorders of Biotin Metabolism. In: Scriver, C.R., Kaufman, S., Eisensmith, E., Woo S.L.C., Vogelstein, B. Childs, B. (eds) The Metabolic and Molecular Bases of Inherited Disease, 8th ed. McGraw-Hill, New York, Ch.156 pg. 3947-3956.
- Wolf, B. (2003) Biotinidase Deficiency: New Direction and Practical Concerns. Current Treatment Options in Neurology 5(4): 321-328.