

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 biotin bound into 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 BTM (Biotinidase) gene. The BTM 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 BTM 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 BTM 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 BTM gene and one copy of the normal BTM gene.

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

Is There a Test for Biotinidase Deficiency?

Yes. Babies are tested through 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. Approximately half of the states screen newborns for biotinidase deficiency, and Tennessee is one of them.

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 the 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

<http://www.ccmckids.org/research/biotinidase/Biotinidase%20Deficiency%20Booklet%203-12-03.pdf>



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

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

References:

- GeneTests (Biotinidase Deficiency) <http://www.genetests.org>
- Online Mendelian Inheritance in Man (OMIM topics 253260)
<http://www.ncbi.nlm.nih.gov/Omim>
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- Wolf, B. (2003) Biotinidase Deficiency: New Direction and Practical Concerns. Current Treatment Options in Neurology 5(4): 321-328.