

Biotinidase Deficiency

What is biotinidase deficiency?

Biotinidase Deficiency is an inherited metabolic disorder of biotin (Vitamin B complex) recycling that leads to multiple carboxylase deficiencies. The genetic disorder is transmitted as an autosomal recessive disorder.

There are several genetic variants characterized by less severe reduction in the enzyme activity. Although most of these individuals having the disorder are asymptomatic, all should be evaluated, as some will require management and monitoring.

What are the symptoms?

Infants with Biotinidase Deficiency appear normal at birth but develop one or more of the following symptoms after the first few weeks or months of life: ataxia, hypotonia, respiratory problems, seizures, hearing loss, alopecia, developmental delay, skin rash, or metabolic acidosis which can result in coma and death. Individuals with partial deficiency (a variant form) may also be at risk for development of any of the above symptoms, but symptoms are mild and occur only when the child is stressed, such as with a prolonged infection. Early detection is crucial. Treatment is simple, inexpensive, and highly effective.

How do you get it?

Biotinidase deficiency is inherited in an autosomal recessive pattern. As an autosomal recessive disorder, the parents of a child with Biotinidase Deficiency are unaffected. Healthy carriers of the condition have one normal gene and one abnormal gene. With each pregnancy, carrier parents have a 25 percent chance of having a child with two copies of the abnormal gene, resulting in biotinidase deficiency. Carrier parents have a 50 percent chance of having a child who is an unaffected carrier and a 25 percent chance of having an unaffected, non-carrier child. These risks hold true for each pregnancy. All siblings of infants diagnosed with Biotinidase Deficiency should be tested for the deficiency even if they do not exhibit symptoms. Carrier testing for at risk family members by targeted mutation analysis using the panel of common BTD mutations are available. Genetic counseling services should be offered to the family.

How common is it?

Estimated prevalence of Biotinidase Deficiency for profound deficiency (absent enzyme) is 1 in 113,000 births in Missouri.

Estimated prevalence for profound and partial Biotinidase Deficiency is 1 in 60,000 births in Missouri.

The condition occurs in all ethnic groups.

How is it treated?

All individuals with profound Biotinidase Deficiency, even those who have some residual enzymatic activity, should have lifelong treatment with biotin. Biotinidase Deficiency is treated by biotin therapy and should be done in consultation with a pediatric metabolic specialist.

Where can I get services?

Cardinal Glennon Children's Medical Center
St. Louis, MO 1-314-577-5639

Website: <http://pediatrics.slu.edu/index.php?page=medical-genetics-2>

Children's Mercy Hospital
Kansas City, MO 1-816-234-3290

Website: <http://www.childrens-mercy.org/content/view.aspx?id=155>

St. Louis Children's Hospital
St. Louis, MO 1-314-454-6093

Website: <http://www.peds.wustl.edu/genetics/>

University Hospitals and Clinics
Columbia, MO 1-573-882-6991

Website: <http://www.genetics.missouri.edu/>

Related Links

American College of Medical Genetics

<http://www.acmg.net/resources/policies/ACT/condition-analyte-links.html>

Biotinidase Deficiency Family Support Group

<http://biotinidasedeficiency.20m.com/definition.htm>

Genetics Home Reference: Your Guide to Understanding Genetic Counseling

<http://www.ghr.nlm.nih.gov>

Medline Plus (National Library of Medicine and the National Institute of Health)

<http://www.medlineplus.gov>

National Institutes of Health

<http://www.nih.gov>