

# Biotinidase deficiency

## What is biotinidase deficiency?

Biotinidase deficiency is an inherited disorder in which the body is unable to reuse and recycle the vitamin biotin. This disorder is classified as a multiple carboxylase deficiency, a group of disorders characterized by impaired activity of certain enzymes that depend on biotin. The signs and symptoms of biotinidase deficiency typically appear within the first few months of life, but the age of onset varies. Children with profound biotinidase deficiency, the more severe form of the condition, often have seizures, weak muscle tone (hypotonia), breathing problems, and delayed development. If left untreated, the disorder can lead to hearing loss, eye abnormalities and loss of vision, problems with movement and balance (ataxia), skin rashes, hair loss (alopecia), and a fungal infection called candidiasis. Immediate treatment and lifelong management with biotin supplements can prevent many of these complications. Partial biotinidase deficiency is a milder form of this condition. Affected children experience hypotonia, skin rashes, and hair loss, but these problems may appear only during illness, infection, or other times of stress.

## How common is biotinidase deficiency?

Profound or partial biotinidase deficiency occurs in approximately 1 in 60,000 newborns

## What genes are related to biotinidase deficiency?

Mutations in the BTD gene cause biotinidase deficiency.

The BTD gene provides instructions for making an enzyme called biotinidase. This enzyme helps the body reuse biotin, a B vitamin found in foods such as liver, egg yolks, and milk. Biotinidase removes biotin that is bound to proteins in food, leaving the vitamin in its free (unbound) state. The body needs free biotin to break down fats, proteins, and carbohydrates effectively. Biotinidase also recycles biotin within the body.

Mutations in the BTD gene reduce or eliminate the activity of biotinidase. Profound biotinidase deficiency results when the activity of biotinidase is reduced to less than 10 percent of normal. Partial biotinidase deficiency occurs when biotinidase activity is reduced to between 10 percent and 30 percent of normal. Without enough of this enzyme, biotin cannot be separated from proteins or recycled normally. As a result, the body is less able to process important nutrients. These defects underlie the potentially serious medical problems associated with biotinidase deficiency.

Read more about the BTD gene.

## How do people inherit biotinidase deficiency?

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## Where can I find information about treatment for biotinidase deficiency?

These resources address the management of biotinidase deficiency and may include treatment providers.

- Gene Reviews: Biotinidase Deficiency
- MedlinePlus Encyclopedia: Pantothenic Acid and Biotin

You might also find information on treatment of biotinidase deficiency in Educational resources and Patient support.

## Where can I find additional information about biotinidase deficiency?

You may find the following resources about biotinidase deficiency helpful. These materials are written for the general public.

- MedlinePlus - Health information (3 links)
- Educational resources - Information pages (7 links)
- Patient support - For patients and families (5 links)

You may also be interested in these resources, which are designed for healthcare professionals and researchers.

- Gene Reviews - Clinical summary

- Gene Tests - DNA tests ordered by healthcare professionals
- ACTion Sheets - Newborn screening follow up (1 link)
- ClinicalTrials.gov - Linking patients to medical research
- PubMed - Recent literature
- Online Books - Medical and science texts
- Scriver's Online Metabolic and Molecular Bases of Inherited Disease (OMMBID): Disorders of Biotin Metabolism
- OMIM - Genetic disorder catalog

## What other names do people use for biotinidase deficiency?

- BIOT
- BTD deficiency
- Carboxylase Deficiency, Multiple, Late-Onset
- Late-onset biotin-responsive multiple carboxylase deficiency
- Late-onset multiple carboxylase deficiency
- Multiple Carboxylase Deficiency, Late-Onset

See How are genetic conditions and genes named? in the Handbook.

## What if I still have specific questions about biotinidase deficiency?

- See How can I find a genetics professional in my area? in the Handbook.
- Ask the Genetic and Rare Diseases Information Center.
- Submit your question to Ask the Geneticist.

## Where can I find general information about genetic conditions?

The Handbook provides basic information about genetics in clear language.

- What does it mean if a disorder seems to run in my family?
- What are the different ways in which a genetic condition can be inherited?
- If a genetic disorder runs in my family, what are the chances that my children will have the condition?
- Why are some genetic conditions more common in particular ethnic groups?

These links provide additional genetics resources that may be useful.

- Genetics and health
- Resources for Patients and Families
- Resources for Health Professionals

## What glossary definitions help with understanding biotinidase deficiency?

alopecia ; ataxia ; autosomal ; autosomal recessive ; biotin ; candidiasis ; carbohydrate ; carboxylase ; cell ; complication ; deficiency ; egg ; enzyme ; gene ; hypotonia ; infection ; muscle tone ; mutation ; newborn screening ; protein ; recessive ; screening ; seizure ; sign ; stress ; symptom ; vitamins

You may find definitions for these and many other terms in the Genetics Home Reference Glossary.

## References

These sources were used to develop the Genetics Home Reference condition summary on biotinidase deficiency.

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See also Understanding Medical Terminology.

## External links

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