

## Complete Information on Biotinidase deficiency

Biotinidase Deficiency (BIOT) is caused by the lack of an enzyme called biotinidase. 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. Children with profound biotinidase deficiency, the more severe form of the condition. Infants with biotinidase deficiency appear normal at birth, but develop critical symptoms after the first weeks or months of life.

Symptoms include hypotonia, ataxia, seizures, developmental delay, alopecia, seborrheic dermatitis, hearing loss and optic nerve atrophy. Metabolic acidosis can result in coma and death. Biotinidase deficiency typically accounts for the so-called late-onset multiple carboxylase deficiency. The early or neonatal onset of multiple carboxylase deficiency is more likely due to another biotin-responsive biochemical abnormality, holocarboxylase synthetase deficiency. 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. Profound biotinidase deficiency typically presents in the first 6 months of life, although presentation in the neonatal period or after the first decade occurs. Approximately 1 in 60,000 newborns are affected by profound (less than 10 percent of normal enzyme activity) or partial biotinidase deficiency. Males and females appear to be affected equally, which is consistent with an autosomal recessive pattern of inheritance. Biotinidase deficiency is treated with free biotin, or biotin that is not bound to protein or other molecules.

Biotinidase deficiency is treated with oral biotin supplementation, which prevents development of the clinical symptoms. It is recommended that parents travel with a letter of treatment guidelines from the patient's physician. Biotin therapy is initiated in prescription doses of 5 mg-20 mg per day. Biotin therapy provides the body with sufficient free biotin for all metabolic needs. Therapy is lifelong, and no dietary restrictions are necessary. The prognosis for individuals diagnosed with biotinidase deficiency is very good, especially for those who were treated before symptoms occurred.

### About the Author

Juliet Cohen writes articles for [health care blog](#). She also writes articles for [hairstyles gallery](#).

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