

Disease Name	
BIOTINIDASE DEFICIENCY	
<i>MULTIPLE CARBOXYLASE DEFICIENCY, LATE-ONSET; MULTIPLE CARBOXYLASE DEFICIENCY, JUVENILE-ONSET; BTD DEFICIENCY</i>	
Classification:	Organic aciduria
Genetic Information	
Inheritance:	Biotinidase deficiency is inherited as an autosomal recessive condition.
Population Incidence:	The incidence is estimated to be 1 in 60,000 live births.
Ethnic Incidence:	No known population at increased risk.
Gene & Location:	Gene BTD on 3p25
Common Mutation:	None known.
OMIM #	#253260; *609019
Disease Information	
Symptom Onset:	Generally appear in infancy or early childhood.
Symptoms:	The symptoms of biotinidase deficiency are variable with respect to age of onset, frequency, and severity. Signs and symptoms may include seizures, skin rash, hair loss, hypotonia, ataxia, hearing loss, optic nerve atrophy, developmental delay, and metabolic acidosis, which can result in coma and death.
Physical Findings:	Variable, but can include seizures, skin rash, hair loss, hypotonia, ataxia, hearing loss, optic nerve atrophy, and developmental delay.
Treatment:	Acute symptoms will completely disappear with administration of pharmacological doses of biotin, usually 10 mg per day. The hearing loss and vision problems can resolve over time as well. Usually there are no biotinidase-related problems occurring once treatment is started. Life long treatment is recommended for all individuals with biotinidase deficiency.
Natural History without treatment:	Depending on the severity of the defect. Severe or total biotinidase deficiency presents early in life and progresses to neurological signs with rash and if the patient is severely stressed can lead to death from metabolic acidosis. Partial deficiency may have problems in cases of severe metabolic stress, but that is speculative.
Natural History With treatment:	If on biotin supplementation, unlikely to have any problems according to the literature.
Metabolic Information	
Missing Enzyme & Location:	Biotinidase is an enzyme in the serum that recycles biotin, an essential cofactor, from a bound form so that it can be used by the body. In the absence of the enzyme, the body becomes biotin deficient.
NBS profile:	Decreased fluorescence on enzyme assay.
Prenatal testing:	Possible, but not offered clinically.
Miscellaneous Information:	Screening results are affected by blood transfusions. Newborn screening specimen should always be collected prior to a transfusion. Test is not dependent on timing or type of feeding. The enzyme activity may be affected if the sample is delayed in the mail or exposed to high temperatures. Family studies are indicated when an affected newborn is identified.
Credit:	<i>Prepared by Center for Congenital and Inherited Disorders (CIDAC), reviewed by Sara Copeland MD, Iowa Neonatal Metabolic Screening Program.</i>

<p>Sites of Reference:</p>	<p>Biotinidase Deficiency - eMedicine.com www.emedicine.com/PED/topic239.htm</p> <p>Biotinidase Deficiency - Family Support Group www.geocities.com/biotinidasedeficiency/</p> <p>Biotinidase Deficiency - Gene Clinics www.geneclinics.org/profiles/biotin/</p> <p>Biotinidase Deficiency - Nebraska Health & Human Services System www.hhs.state.ne.us/nsp/Newborn5.htm</p> <p>Biotinidase Deficiency - OMIM www3.ncbi.nlm.nih.gov/htbin-post/Omim/dispmim?253260</p> <p>Biotinidase Deficiency - Washington Dept of Health www.doh.wa.gov/EHSPHL/PHL/Newborn/biotinidase.htm</p> <p>Biotinidase Deficiency: A Booklet for Families and Professionals www.ccmckids.org/research/Biotinidase/Biotinidase_Deficiency_Booklet.pdf</p>	
<p>Support Groups:</p>	<p>Biotinidase Family Support Group Contact: Laurie Farmer lfarmer@ora.fda.gov</p>	<p>Save Babies Through Screening Foundation, Inc 4 Manor View Circle Malvern, PA 19355-1622 (888) 454-3383 (610) 993-0545 email@savebabies.org www.savebabies.org/index.htm</p>