

Biotinidase deficiency (BIOT)

Health Care Professional Fact Sheet

<p>A newborn screening test is a screen and not diagnostic testing. An “abnormal” or “critical” result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose the baby with the condition. Follow-up testing is vital to determine if the baby has the disorder indicated. In the event the condition is diagnosed, timely follow-up testing will result in earlier treatment and better outcomes.</p>	
<p>Disorder Indicated: Biotinidase deficiency (BIOT) is an inherited condition in which the body is unable to reuse and recycle the vitamin biotin. Because the body needs free biotin to break down fats, proteins, and carbohydrates effectively, individuals with BIOT are less able to process important nutrients.</p> <p>Children with BIOT who are identified through newborn screening and begin treatment immediately usually remain healthy with normal development. Some children with mild forms may not need any treatment.</p>	
Incidence	1 in every 60,000 newborns.
Analyte Measured	Biotinidase enzyme activity
Normal Test Results	BIOT > 60 U/dL
Abnormal Test Results	BIOT ≤ 60 U/dL
Critical Test Results	2 samples with abnormal results <i>(Critical results require immediate evaluation and follow-up)</i>
Signs and Symptoms <i>Please note: these findings may not be present in young infants or in milder forms of the disease</i>	<p>There are two main types of biotinidase deficiency (BIOT), differing in the severity of signs: severe “profound biotinidase deficiency” and mild “partial biotinidase deficiency.” Signs of BIOT usually start within a few months after birth. In some cases, the symptoms may not appear until childhood.</p> <p>When a child has biotinidase deficiency, you may see symptoms including:</p> <ul style="list-style-type: none"> • Seizures • Weak muscle tone (hypotonia) • Trouble breathing • Skin rash • Hair loss • Trouble balancing • A fungal infection called candidiasis <p><i>Many of these signs can be triggered by illnesses or infections.</i></p>
Next Steps <i>may</i> include:	<p>Discuss the next steps of evaluation and possible treatment with the regional metabolic consultant</p> <p>Provide parental education (see accompanying sheet)</p> <p>Clinical assessment</p> <p>Serum Biotinidase Assay</p>
Treatment <i>(if indicated)</i>	Change in diet (addition of Biotin supplements)
Additional Resources	<p>VDH Newborn Screening http://vdhlivewell.com/newbornscreening</p> <p>Baby’s First Test www.babysfirsttest.org</p> <p>American College of Medical Genetics (ACMG) ACT Sheets www.ACMG.net</p> <p>Genetics Home Reference https://ghr.nlm.nih.gov/</p> <p>Biotinidase Deficiency Support Group www.biotinidasedeficiency.20m.com/</p>

Educational content adapted from www.babysfirsttest.org

