

Interpretation of Newborn Screening Results for Biotinidase Deficiency:

“<” means “is less than”

“>” means “is greater than”

Final Result	Test Results		Likely Causes
	Biotinidase	Reference Range	
Normal	Normal	> 80.0 U/dL	No indication of a defect in Biotin metabolism.
Borderline risk	Abnormal (decreased)	55.0 – 80.0 U/dL	<ul style="list-style-type: none"> - Variant form of Biotinidase deficiency - Other enzyme defects in red blood cells - Improperly handled sample (heat damage or transit delay) - Impaired liver function, prematurity, severe jaundice
High Risk	Abnormal (decreased)	< 55.0 U/dL	<ul style="list-style-type: none"> - Profound Biotinidase Deficiency - Variant form of Biotinidase deficiency - Other enzyme defects in red blood cells - Improperly handled sample (heat damage or transit delay) - Impaired liver function, prematurity, severe jaundice
No Result	No Result		<ul style="list-style-type: none"> - Transfusion - Possible transfusion - Poor sample quality