



Patient ID <b>SA00057814</b>	Patient Name <b>SAMPLEREPORT, BIOTS A</b>	Birth Date <b>1966-06-10</b>	Gender <b>F</b>	Age <b>46</b>
Order Number <b>SA00057814</b>	Client Order Number <b>SA00057814</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>27 May 2013 00:00</b>		

## Biotinidase, S


### Reason For Referral

Not provided

MCR

ordering Biotinidase Deficiency, BTD Full Gene Analysis (MML #89012) to confirm the diagnosis. Free-form biotin supplementation is recommended. For dosage recommendations, please see GeneReviews (<http://www.ncbi.nlm.nih.gov/sites/GeneTests/?db=GeneTests>). Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

### Biotinidase, S

 **1.5 U/L**

Low

MCR

Reference Value  
3.5-13.8

### ADDITIONAL INFORMATION

Colorimetric Enzyme Assay

### Interpretation

In this sample, biotinidase activity is lower than our normal reference range indicating that this individual may be affected with biotinidase deficiency (OMIM #253260). Levels between 1.5 U/L and 3.4 U/L are also seen in milder variants or carriers. If clinically indicated, consider submitting a new blood sample and

MCR

### Reviewed By

Matthew Fatehi

MCR

Received: 28 May 2013 16:43

Reported: 28 May 2013 16:53

### Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905