


Patient ID <b>SA00049337</b>	Patient Name <b>TESTINGRNV, BGAABNORM</b>	Birth Date <b>1987-08-10</b>	Gender <b>M</b>	Age <b>25</b>
Order Number <b>SA00049337</b>	Client Order Number <b>SA00049337</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>26 Sep 2012 00:00</b>		

## Beta-Galactosidase, Leukocytes

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 **0.23 nmol/min/mg**  
**Low**

**MCR**  
**Reference Value**  
≥1.56

### Interpretation (BGA)

In this specimen, the activity of beta-D-galactosidase is significantly reduced. This result indicates this individual is affected with GM1 gangliosidosis (OMIM #230500, 230600, or 230650) or mucopolysaccharidosis type IVB (Morquio B; OMIM #253010). Please note that this enzyme is also decreased in galactosialidosis (OMIM #256540), mucopolipidosis II/III (I-cell disease; #252500 or 252600), and in rare instances where asymptomatic individuals carry a pseudodeficiency mutation in the GLB1 gene (Gort L. Clin Genet 2007; 72: 109-11). If clinically

indicated, consider follow-up testing which may include molecular genetic analysis of the GLB1 gene for GM1 gangliosidosis and Morquio type B. A genetics consultation is recommended. We need clinical information to provide further interpretation or to discuss the best follow up testing strategy. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

### ADDITIONAL INFORMATION

Fluorometric Enzyme Assay

### Reviewed By

JANICE HELGESON

**MCR**

**Received:** 27 Sep 2012 14:29

**Reported:** 27 Sep 2012 14:36

### Performing Site Legend

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