



Patient ID <b>SA00067741</b>	Patient Name <b>SAMPLEREPOR, GPSY-ABN</b>	Birth Date <b>1994-07-04</b>	Gender <b>F</b>	Age <b>19</b>
Order Number <b>SA00067741</b>	Client Order Number <b>SA00067741</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>02 Jun 2014 06:30</b>		

## Glucopsychosine, BS

### Glucopsychosine



155 nmol/L

High

MCR  
Reference Value  
<47

### Reviewed By

Jennifer Peterson

MCR

### Interpretation (GPSY)

MCR

In this sample, the concentration of glucopsychosine (glucosylsphingosine) was elevated. This result is consistent with a diagnosis of Gaucher disease (OMIM 230800). Consider determination of acid beta-glucosidase activity in leukocytes (MML test ID: BGL/Beta-Glucosidase, Leukocytes) to confirm this diagnosis. Please contact the Biochemical Genetics consultant on call (1-800-533-1710) if you have any questions.

### ADDITIONAL INFORMATION

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

**Received:** 03 Jun 2014 08:20

**Reported:** 03 Jun 2014 09:10

### Performing Site Legend

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