



Patient ID <b>SA00062405</b>	Patient Name <b>SAMPLEREPOR, BGLT A</b>	Birth Date <b>2004-04-24</b>	Gender <b>F</b>	Age <b>9</b>
Order Number <b>SA00062405</b>	Client Order Number <b>SA00062405</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>17 Sep 2013 10:23</b>		

## Beta-Glucosidase, Fibroblasts

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MCR



**0.13 nmol/min/mg Prot**

Reference Value  
≥4.85

Low

### Reviewed By

MCR

Daniel Kraft

Received: 17 Sep 2013 11:13

Reported: 17 Sep 2013 13:33

### Interpretation (BGLT)

MCR

In this specimen, the activity of acid beta-glucosidase is reduced. This result indicates that this individual is affected with Gaucher disease. Please be aware that some carriers for Gaucher disease may also have reduced enzyme activity, although this test is not reliable for carrier detection. If clinically indicated, consider further confirmation to include molecular genetics analysis of GBA gene. 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

### Cryopreserve for Biochem Studies

MCR

Cryopreserve for Biochem Studies was cancelled on 09/17/2013 at 11:13; Test not performed.

Received: 17 Sep 2013 11:13

Reported: 17 Sep 2013 11:13

### Fibroblast Culture

MCR

Fibroblast Culture was cancelled on 09/17/2013 at 11:13; Test not performed.

Received: 17 Sep 2013 11:13

Reported: 17 Sep 2013 11:13

### Performing Site Legend

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