



Patient ID <b>SA00062401</b>	Patient Name <b>SAMPLEREPORT, BGLR A</b>	Birth Date <b>1997-04-09</b>	Gender <b>F</b>	Age <b>16</b>
Order Number <b>SA00062401</b>	Client Order Number <b>SA00062401</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>17 Sep 2013 10:18</b>		

## Beta-Glucuronidase, Fibroblasts

### Beta-Glucuronidase, Fibroblasts

MCR



0.06 nmol/min/mg Prot

Reference Value  
≥ 2.33

Low

### Reviewed By

MCR

Daniel Kraft

Received: 17 Sep 2013 11:11

Reported: 17 Sep 2013 13:27

### Interpretation (BGLR)

MCR

In this specimen, the activity of beta-glucuronidase is significantly reduced. This result indicates that this individual is affected with Sly disease (Mucopolysaccharidosis type VII). In rare instances this enzyme may be low due to a pseudodeficiency mutation. If clinically indicated, consider further confirmation to include molecular genetic analysis of the GUSB 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 11:11; Test not performed.

Received: 17 Sep 2013 11:11

Reported: 17 Sep 2013 11:11

### Fibroblast Culture

MCR

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

Received: 17 Sep 2013 11:11

Reported: 17 Sep 2013 11:11

### Performing Site Legend

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