

Patient Name SAMPLEREPORT,BGLT A	Patient ID SA00062405	Age 9	Gender F	Order # SA00062405
Ordering Phys CLIENT,CLIENT				DOB 04/24/2004
Client Order # SA00062405	Account Information			Report Notes
Collected 09/17/2013 10:23	C7028846-DLMP Rochester SDSC 2 - Client Support			
Printed 10/14/2013 13:52	Rochester, MN 55901			

Test	Flag	Results	Unit	Reference Value	Perform Site*
Beta-Glucosidase, Fibroblasts	L	0.13	nmol/min/mg Prot	>=4.85	MCR

RECEIVED: 09/17/2013 11:13 REPORTED: 09/17/2013 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.

Fluorometric Enzyme Assay

Reviewed By

Daniel Kraft

MCR

Cryopreserve for Biochem Studies

MCR

RECEIVED: 09/17/2013 11:13 REPORTED: 09/17/2013 11:13

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

Fibroblast Culture

MCR

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

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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Patient Name SAMPLEREPORT,BGLT A	Collection Date and Time 09/17/2013 10:23	Report Status Final
Page 1 of 1		** End of Report **

* Report times for Mayo performed tests are CST/CDT