



Patient ID SA00062398	Patient Name SAMPLEREPORT, BGAT A	Birth Date 1948-08-14	Gender M	Age 65
Order Number SA00062398	Client Order Number SA00062398	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 17 Sep 2013 10:13		

Beta-Galactosidase, Fibroblasts

Beta-Galactosidase, Fibroblasts



0.12 nmol/min/mg Prot

Reference Value
≥7.11

MCR

Low

Interpretation (BGAT)

MCR

In this specimen, the activity of beta-D-galactosidase is 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

MCR

Daniel Kraft

Received: 17 Sep 2013 11:09

Reported: 17 Sep 2013 13:23

Cryopreserve for Biochem Studies

MCR

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

Received: 17 Sep 2013 11:09

Reported: 17 Sep 2013 11:09

Fibroblast Culture

MCR

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

Received: 17 Sep 2013 11:09

Reported: 17 Sep 2013 11:09

Performing Site Legend

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