



Patient ID SA00048853	Patient Name SAMPLEREPOR, BGAW	Birth Date 1966-06-10	Gender F	Age 46
Order Number SA00048853	Client Order Number SA00048853	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 04 Sep 2012 13:00		

Beta-Galactosidase, B

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4.9 nmol/h/mL

Low

MCR

Reference Value
≥5.0

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

Interpretation

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

Reason for Referral

MCR

Not provided

Reviewed By

MCR

Marie Quade

Received: 05 Sep 2012 14:26

Reported: 05 Sep 2012 14:36

Performing Site Legend

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