MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,BGAW	SA00048853	46	F	SA00048853
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00048853	**Account Information**			**Report Notes**
Collected 09/04/2012 13:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 10/17/2012 15:31				

Test	Flag	Results	Unit	Reference Value	Perform Site*
			REPORTED 09/05/2012 14:36		
Beta-Galactosidase, B	L	4.9	nmol/h/mL	>=5.0	MCR
Interpretation					MCR
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), mucolipidosis 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.					
Fluorometric Enzyme Assay					
Reason for Referral		Not provided			MCR
Reviewed By		Marie Quade			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

Patient Name	Collection Date and Time	Report Status
SAMPLEREPORT,BGAW	09/04/2012 13:00	Final
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* Report times for Mayo performed tests are CST/CDT