MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,GAL6	SA00043804	45	F	SA00043804
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00043804	**Account Information**			**Report Notes**
Collected 02/28/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 02/29/2012 16:12				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Galactosemia Gene Analysis		REPORTED 02/29/2012 15:01			
Specimen		Blood			MCR
Specimen ID		1037931			MCR
Order Date		29 Feb 2012 14:45			MCR
Reason For Referral					MCR
Biochemical testing shows galactose 1 phosphate uridyltransferase (GALT) activity of <18.5 U/g Hgb. Test for the presence of mutation(s) in the GALT gene.					
Method					MCR
A real-time PCR-based assay was utilized to examine DNA for six alterations: Q188R, S135L, L195P, K285N, N314D (Duarte) and L218L (Los Angeles).					
Result					MCR
None of the listed mutations were detected.					
Interpretation					MCR
Biochemical and molecular test results for this patient are inconsistent. The observed GALT enzyme activity in red blood cells (9.8 U/g Hgb) is most consistent with positive carrier status for classic galactosemia. However, no mutation was identified by molecular analysis. Approximately 30% of carriers for classic galactosemia are expected to have an undetected mutation by this assay.					
Full gene sequencing is available to evaluate for mutation(s) in the GALT gene not detected by this six mutation panel. If this test is desired, please contact the Molecular Genetics Laboratory to have this test added on to the existing sample or submit a new sample for testing (MML test ID GALTM/88877).					
A genetic consultation may be of benefit.					
CAUTIONS: Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.					
Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.					
Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for					

Performing Site Legend on Last Page of Report

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Test	Flag	Results	Unit	Reference Value	Perform Site*
instructions for testing patients who have received a bone marrow transplant.					
Laboratory developed test.					
Reviewed By:		Kimberly Anne Schahl			MCR
Release Date		29 Feb 2012 14:59			MCR

* Performing Site:

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905	Lab Director: