

Patient Name SAMPLEREP, GAL6	Patient ID SA00043805	Age 45	Gender F	Order # SA00043805
Ordering Phys				DOB 06/10/1966
Client Order # SA00043805	Account Information			Report Notes
Collected 02/28/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER, MN 55901			
Printed 02/29/2012 16:14				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Galactosemia Gene Analysis			REPORTED 02/29/2012 15:03		
Specimen		Blood			MCR
Specimen ID		1037932			MCR
Order Date		29 Feb 2012 14:53			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
One copy each of the Q188R and N314D mutations was detected.					
Interpretation					MCR
Biochemical and molecular test results are in agreement. The observed GALT enzyme activity in red blood cells (4.2 U/g Hgb) and the presence of the N314D (Duarte) and Q188R alleles suggest that this individual is affected with Duarte variant galactosemia. If clinically indicated, consider measuring galactose 1 phosphate (MML test ID GAL1P/80337). Some individuals with Duarte variant galactosemia are treated with a low galactose containing diet during infancy.					
Since mutations have been identified, genetic testing of at risk relatives is possible.					
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 instructions for testing patients who have received a bone marrow transplant.					

Performing Site Legend on Last Page of Report

Patient Name SAMPLEREP, GAL6	Collection Date and Time 02/28/2012	Report Status Final
Page 1 of 2		>> Continued on Next Page >>

* Report times for Mayo performed tests are CST/CDT

Patient Name SAMPLEREPORT,GAL6	Patient ID SA00043805	Age 45	Gender F	Order # SA00043805
Ordering Phys				DOB 06/10/1966
Client Order # SA00043805	Account Information			Report Notes
Collected 02/28/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 02/29/2012 16:14				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Laboratory developed test.					
Reviewed By:		Kimberly Anne Schahl			MCR
Release Date		29 Feb 2012 15:00			MCR

* Performing Site:

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

Patient Name SAMPLEREPORT,GAL6	Collection Date and Time 02/28/2012	Report Status Final
Page 2 of 2		** End of Report **

* Report times for Mayo performed tests are CST/CDT