MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,GALTK	SA00043809	45	F	SA00043809
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00043809	**Account Information**			**Report Notes**
Collected 02/28/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 02/29/2012 16:15				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Galactosemia Known Mutation			REPORTED 02/29/2012 15:09		
Specimen		Blood			MCR
Specimen ID		1037937			MCR
Order Date		29 Feb 2012 14:55			MCR
Reason For Referral					MCR
Family history of galactosemia. Test for the presence of familial alteration(s) within the GALT gene.					
Method					MCR
DNA sequence analysis was used to test for the presence of the p.Q188R (c.563A>G) and p.N314D (c.940A>G) alterations in exons 6 and 10, respectively, of the GALT gene (GenBank accession number; NM_000155.2). Testing was performed for these specific alterations because they were previously identified in a family member.					
Result					MCR
The following heterozygous sequence change was detected: Exon: 10 DNA change: c.940A>G Amino acid change: p.N314D (Asn314Asp) This sequence change is a variant associated with reduced GALT activity.					
Interpretation					MCR
This result indicates that this individual is a carrier of the Duarte variant and should not be at risk for developing symptoms related to this disorder. However, there is an increased risk to have children with Duarte galactosemia. If appropriate, consider enzymatic and molecular studies of this individual's reproductive partner to further clarify this risk.					
This assay does not rule out the presence of other disease-causing mutations in this gene or other genes associated with metabolic disease. Errors in the diagnosis or pedigree provided to us, including non-paternity, may lead to an erroneous interpretation of test results.					
Since a mutation has been identified, genetic testing of at-risk relatives is possible.					
A genetic consultation may be of benefit.					
A list of common polymorphisms identified for this patient is available upon request.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,GALTK	SA00043809	45	F	SA00043809
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00043809	**Account Information**			**Report Notes**
Collected 02/28/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 02/29/2012 16:15				

Test	Flag	Results	Unit	Reference Value	Perform Site*

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.

Laboratory developed test.

Reviewed By:	Kimberly Anne Schahl	MCR
Release Date	29 Feb 2012 15:06	MCR

* Performing Site:

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

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

* Report times for Mayo performed tests are CST/CDT