

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,GALTM	Patient ID SA00043807	Age 45	Gender F	Order # SA00043807
Ordering Phys				DOB 06/10/1966
Client Order # SA00043807	Account Information	1		Report Notes
Collected 02/28/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE			
Printed 02/29/2012 16:14	ROCHESTER,MN 559	901		

Test	Flag	Results	Unit	Reference Value	Perform Site*
	5				
GALT Gene, Full Gene Analysis			REPORTED	02/29/2012 15:05	
Specimen		Blood			MCR
Specimen ID		1037936			MCR
Order Date		29 Feb 2012 14:55			MCR
Reason For Referral					MCR
Biochemical testing shows	-	-	-		
uridyltransferase (GALT) a	-		ior		
the presence of mutations	in the GALT g	gene.			N/GD
Method		6	c		MCR
DNA sequence analysis was		-			
<pre>mutation in all 11 exons o number; NM_000155.2).</pre>	of the GALT ge	ene (GenBank access:	ion		
Result					MCR
The following heterozygous	s segmence cha	anges were detected	•		MCK
Exon: 6	bequence ena	anges were detected	•		
DNA change: c.563A>G					
Amino acid change: p.Q188F	(Gln188Arg)				
This sequence change is a		itation.			
min pequence ondinge in a	Pacific Julia				
Exon: 10					
DNA change: c.940A>G					
Amino acid change: p.N314	D (Asn314Asp)				
This sequence change is a	variant assoc	ciated with reduced			
GALT activity.					
Interpretation					MCR
The biochemical test resul		-			
cells: $4.6~\mathrm{U/g}$ Hgb) and re		_	S		
are consistent with Duarte	galactosemia	a. If clinically			
indicated, consider measur					
test ID GAL1P/80337). Som	ne individuals	s with Duarte			
galactosemia are treated w	ith low galac	tose-containing die	et		
during infancy.					
Since mutations have been	identified o	renetic testing of			
at-risk family members is		genetic testing of			
at-lisk lamily members is	possible.				
A genetic consultation may	be of benefi	t.			
A list of common polymorph is available upon request.		ed for this patient	t		
CAUTIONS:					

Performing Site Legend on Last Page of Report

Patient Name SAMPLEREPORT,GALTM	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

Test results should be interpreted in context of clinical findings, family history, and other laboratory data.



Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,GALTM	Patient ID SA00043807	Age 45	Gender F	Order # SA00043807
Ordering Phys				DOB 06/10/1966
Client Order # SA00043807	Account Information			Report Notes
Collected 02/28/2012	C7028846-DLMP RO 3050 SUPERIOR DRI	VE		
Printed 02/29/2012 16:14	ROCHESTER,MN 559	901		

Test Flag Results Unit Value Site*

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 Release Date Kimberly Anne Schahl 29 Feb 2012 15:03 MCR MCR

* Performing Site:

I MOD IV	layo Clinic Dpt of Lab Med & Pathology	Lab Diversion, Eventsia D. Carlesvill III M.D.
MCR 20	00 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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

^{*} Report times for Mayo performed tests are CST/CDT