

1-800-533-1710

PATIENT NAME TESTING, GALTMS		PATIENT NUMBER L3MRNG9156452		AGE 20Y	SEX M	ACCESSION # G9156452
ORDERING PHYSICIAN			CLIENT ORDER #		ACCOUNT # LIAISONS	
COLLECTION 09/22/10 10:49 A	RECEIVED 09/22/10 10:49 A	REPORT PRINTED 11/22/10 03:08 P		SPECIMEN INFORMATION DATE OF BIRTH:		
DATE TIME	DATE TIME	DATE TIME				
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
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GALT Gene, Full Gene Analysis

Specimen	Blood	MCR
Specimen ID	1034718	MCR
Order Date	22 Sep 2010 13:01	MCR
Reason For Referral		MCR

Patient sample submitted for biochemical testing shows galactose-1-phosphate uridylyltransferase (GALT) activity of <18.5U/g Hgb. Test for the presence of mutations in the GALT gene.

Method		MCR
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DNA sequence analysis was used to test for the presence of a mutation in all 11 exons of the GALT gene (GenBank accession number; NM_000155.2).

Result		MCR
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The following heterozygous sequence changes were detected:

Exon: 6
 DNA change: c.563A>G (CAG>CGG)
 Amino acid change: p.Q188R (Gln188Arg)
 This sequence change is a known pathogenic mutation.

Exon: 10
 DNA change: c.940A>G (AAC>GAC)
 Amino acid change: p.N314D (Asn314Asp)
 This sequence change is a known variant associated with reduced GALT activity.

Interpretation		MCR
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The reported biochemical test result and results of this molecular analysis are consistent with a diagnosis of Duarte galactosemia. If clinically indicated, consider measuring galactose-1-phosphate (MML test code 80337). Some individuals with Duarte galactosemia are treated with low galactose-containing diet during infancy.

* Perform Site Legend on last page of report

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Since mutations have been identified for this individual, genetic testing of other at-risk family members is possible. A genetic consultation may be of benefit. A list of common polymorphisms identified for this patient is available upon request.

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 allogeneic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant. This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Reviewed By: Heather Lynn Owen
 Release Date: 22 Sep 2010 13:15

MCR
 MCR

* PERFORMING SITE

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.
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