MAYO CLINIC
Mayo Medical Laboratories

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: July 14, 2009
EFFECTIVE DATE: July 15, 2009

UDP-GLUCURONOSYL TRANSFERASE 1A1 (UGT1A1) GENE, KNOWN MUTATION #89396

USEFUL FOR: Identifying the presence of a *UGT1A1* mutation when the mutation has been identified in a family member (carrier or affected)

PROFILE INFORMATION:

Unit Code	Reporting Name	Available Separately	Always Performed
30987	UGT1A1 Gene, Known Mutation	No	Yes
89437	UGT1A1 Known Mutation Sequencing	No	Yes

ADDITIONAL TESTS:

Unit Code	Reporting Name	Available Separately	Always Performed
81769	Rapid DNA Extraction	No	Yes

TESTING ALGORITHM:
When this test is ordered, DNA extraction will always be performed at an additional charge.

METHODOLOGY: Polymerase Chain Reaction (PCR) Followed by Site-Specific Gene Sequencing Analysis

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS: Draw blood in a lavender-top (EDTA) tube(s), and send 3 mL of EDTA whole blood in original VACUTAINER(S).

NOTE: 1. Bone marrow and liver transplants will interfere with testing.
- For bone marrow transplant patients, buccal cells from the **recipient** should be provided to obtain an accurate genotype.
- For liver transplant patients, **donor** blood or buccal cells should be provided to obtain an accurate genotype for the recipient patient.

2. Transfusions will interfere with testing for up to 4 to 6 weeks. DNA obtained from white cells may not provide useful information for patients who received a recent transfusion of blood that was not leukocyte-reduced. Wait 4 to 6 weeks until transfused cells have left the patient's circulation before drawing the patient's blood specimen for genotype testing.
3. An "Informed Consent for DNA Testing" (Supply T576) is available. See Special Instructions in the on-line test catalog for a copy of the form.

CAUTIONS:

- This test is for individuals who are at risk for a *UGT1A1* mutation that has been previously identified in the family. If the familial mutation is not known, the familial proband should be screened for a *UGT1A1* mutation using #89611, "UDP-Glucuronosyl Transferase 1A1 (*UGT1A1*), Full Gene Sequencing, Hyperbilirubinemia."
- This assay does not rule out the presence of other mutations within this gene. This test can only be used for known mutations occurring in the promoter, exons, exon-intro boundaries, and the region in the distal promoter called the "phenobarbital response enhancer module."
- Blood transfusions or bone marrow transplantation prior to having blood drawn for DNA analysis can generate false results, as DNA in the specimen may be a mix of patient and donor. Donor blood or buccal cells are needed for testing performed on liver transplant recipients.
- An alternative splice site for exon 5 (referred to as exon 5b) has been discovered and described in the literature. This new exon is described to have a decrease in enzymatic activity (compared with exon 5a: previously known as exon 5), but little is known about the frequency of exon 5b or how it impacts hyperbilirubinemia. Currently, we are not testing or sequencing exon 5b; we continue to monitor the literature for new information on exon 5b.
- Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

LIST FEE: $466.50 [Total list Fee = $616.50]

The following test(s) will be added at an additional charge: $150.00 for #81769 "Rapid DNA Extraction"

CPT CODE:

"UDP-Glucuronosyl Transferase 1A1 (*UGT1A1*) Gene, Known Mutation"

83898/Amplification, target, each sequence
83894/Separation by gel electrophoresis
83892/Enzymatic Digestion
83912/Interpretation and Report

"*UGT1A1* Known Mutation Sequencing"

83909/x2 Mutation identification by sequencing, single segment each segment

"Rapid DNA Extraction"

83890/Molecular Isolation or Extraction

ANALYTIC TIME: 7 days

DAY(S) SET-UP: Monday through Friday

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager
Shirley Pokorski, Mayo Medical Laboratories' Technologist Support
Telephone: 800-533-1710

MAYO CLINIC
Mayo Medical Laboratories

TEST DEFINITION

7/14/2009

Code	Name
89396	UGT1A1 Gene, Known Mutation

ORDER CODE	EFF DATE	TC	TITLE	Checking Normals	Print normals (# coded)	Perform Site *
89396(profile)			UGT1A1 Gene, Known Mutation			
30987	6/29/2009		UGT1A1 Gene, Known Mutation			MCR
			Transport temp : Ambient\Refrig OK\Frozen OK			
			89396 UGT1A1 Gene, Known Mutation			
			30988 UGT1A1 Gene, Known Mutation Interp			
			30989 Reviewed by			
			TEST CODE ALWAYS MESSAGE - [Z30989]			
			Z30989 Bidirectional DNA sequence analysis was used to test for the presence of a specific mutation(s) in the UGT1A1 gene which was previously identified in an affected family member of this individual. The presence of UGT1A1 mutations does not necessarily confirm a diagnosis of unconjugated hyperbilirubinemia. Clinical correlation is recommended. A genetic consultation may be of benefit. CAUTIONS: Rare polymorphisms exist that could lead to false negative or positive results. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Large deletions or rearrangements are not detected by this assay, and these may affect UGT1A1 protein expression, and the ability to conjugate bilirubin.			
89437	6/25/2009		UGT1A1 Known Mutation Sequencing			MCR
			Transport temp : Ambient\Refrig OK\Frozen OK			
			89437 UGT1A1 Known Mutation Sequencing			

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*Performing Site Legend

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

MAYO CLINIC
Mayo Medical Laboratories

TEST DEFINITION

7/14/2009

Code	Name
81769	Rapid DNA Extraction

Order Code	Eff Date	TC	Title	Checking Normals	Print normals (# coded)	Perform Site *
81769	5/23/2007		Rapid DNA Extraction			MCR
			Transport temp : Ambient\Frozen OK\Refrig OK			
		28357	Comment			

*Performing Site Legend

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

MAYO CLINIC
Mayo Medical Laboratories

LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
TESTING, UGTKM		35	M	G9131547

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		LIAISONS

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
07/13/09 10:34 A		07/14/09 01:26 P	DATE OF BIRTH:

Test Client
Attn: Mayo Liaisons
200 First Street SW
Rochester, MN 55905
507-284-8202

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *

UGT1A1 Gene, Known Mutation

UGT1A1 Gene, Known Mutation — MCR

This individual was shown to have the following familial mutation(s) in the UGT1A1 gene:
1043DelA

UGT1A1 Gene, Known Mutation Interp — UG — MCR

The above mutation(s) within the UGT1A1 gene was previously identified in a family member affected with UGT1A1 deficiency. The presence of this mutation(s) is consistent with a diagnosis of unconjugated hyperbilirubinemia in this individual.

Reviewed by — Linnea M. Baudhuin, Ph.D. — MCR

Bidirectional DNA sequence analysis was used to test for the presence of a specific mutation(s) in the UGT1A1 gene which was previously identified in an affected family member of this individual.
The presence of UGT1A1 mutations does not necessarily confirm a diagnosis of unconjugated hyperbilirubinemia.
Clinical correlation is recommended.
A genetic consultation may be of benefit.
CAUTIONS:
Rare polymorphisms exist that could lead to false negative or positive results. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Large deletions or rearrangements are not detected by this assay, and these may affect UGT1A1 protein expression, and the ability to conjugate bilirubin.

Rapid DNA Extraction

** Perform Site Legend on last page of report*

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
TESTING, UGTKM	Final	07/13/09 10:34 A

Specimen receipt and report times are in CST/CDT

REPRINT

MAYO CLINIC
Mayo Medical Laboratories

LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
TESTING, UGTKM		35	M	G9131547

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
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COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
07/13/09 10:34 A	07/13/09 10:34 A	07/14/09 01:26 P	DATE OF BIRTH:

Test Client
Attn: Mayo Liaisons
200 First Street SW
Rochester, MN 55905
507-284-8202

TEST REQUESTED	HI LO		REF RANGE	PERFORM SITE *
Comment		**Genomic DNA was extracted.**		**MCR**
UGT1A1 Known Mutation Sequencing				
UGT1A1 Known Mutation Sequencing		**Performed Sequencing**		**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.

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
TESTING, UGTKM	Final	07/13/09 10:34 A

Specimen receipt and report times are in CST/CDT

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