

UDP-GLUCURONOSYL TRANSFERASE 1A1 (UGT1A1), FULL GENE SEQUENCING, HYPERBILIRUBINEMIA #89611

USEFUL FOR:

- Identifying individuals who are at risk of hyperbilirubinemia
- Confirmation of a diagnosis of Gilbert or Crigler-Najjar syndromes
- Verification of carrier status for Gilbert or Crigler-Najjar syndromes

PROFILE INFORMATION:

Unit Code	Reporting Name	Available Separately	Always Performed
30984	UGT1A1 Sequence, Hyperbilirubinemia	No	Yes
89438	UGT, Full Gene 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 DNA Sequence 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:

- 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.
- Absence of a detectable gene mutation or polymorphism does not rule out the possibility that the patient may have a genetic cause for increased unconjugated bilirubin.
- 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: \$578.90

[Total List Fee = \$728.90]

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*), Full Gene Sequencing, Hyperbilirubinemia"

83900/Amplification, target, multiplex, first two sequences

83901/x3 Amplification, target, multiplex, each additional sequence

83894/Separation by gel electrophoresis

83892/Enzymatic Digestion

83912/Intpretation and Report

"*UGT1A1* Sequence, Hyperbilirubinemia"

83909/x19 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



TEST DEFINITION

7/14/2009

CODE NAME

89611 UGT1A1 SEQUENCE, HYPERBILIRUBINEMIA

ORDER CODE	EFF DATE	TC	TITLE	CHECKING NORMALS	PRINT NORMALS (# CODED)	PERFORM SITE *
89611 (PROFILE)			UGT1A1 SEQUENCE, HYPERBILIRUBINEMIA			
30984	6/29/2009		UGT1A1 SEQUENCE, HYPERBILIRUBINEMIA			MCR
			TRANSPORT TEMP : AMBIENT\REFRIG OK\FROZEN OK			
			89611 UGT1A1 HYPERBILIRUBINEMIA RESULT			
			- - - - -			
			30985 UGT1A1 HYPERBILIRUBINEMIA INTERP			
			- - - - -			
			30986 REVIEWED BY			
			TEST CODE ALWAYS MESSAGE - [Z30986]			
			Z30986 BIDIRECTIONAL DNA SEQUENCE ANALYSIS WAS USED TO TEST FOR THE PRESENCE OF MUTATIONS IN THE PROMOTER, EXONS, EXON-INTRON BOUNDARIES, AND 3'-UNTRANSLATED REGION OF THE UGT1A1 GENE THAT ARE ASSOCIATED WITH THE DIAGNOSIS OF UNCONJUGATED HYPERBILIRUBINEMIA.			
			A SMALL PERCENTAGE OF INDIVIDUALS WHO HAVE A DIAGNOSIS OF UNCONJUGATED HYPERBILIRUBINEMIA MAY HAVE A MUTATION THAT IS NOT IDENTIFIED BY THE METHODS DESCRIBED ABOVE.			
			THE PRESENCE OF UGT1A1 MUTATIONS DOES NOT NECESSARILY CONFIRM A DIAGNOSIS OF UNCONJUGATED HYPERBILIRUBINEMIA.			
			BREAST-FED NEONATES MAY EXPERIENCE A PHYSIOLOGIC UNCONJUGATED HYPERBILIRUBINEMIA AND JAUNDICE FROM DECONJUGATION OF MATERNAL BILIRUBIN-GLUCURONIDES PRESENT IN BREAST MILK. CLINICAL CORRELATION IS RECOMMENDED.			
			A GENETIC CONSULTATION MAY BE OF BENEFIT.			
			A LIST OF VARIATIONS OF UNKNOWN SIGNIFICANCE (VUS) IDENTIFIED FOR THIS PATIENT IS AVAILABLE FROM THE LABORATORY UPON REQUEST.			
			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.

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ORDER CODE	EFF DATE	TC	TITLE	CHECKING NORMALS	PERFORM PRINT NORMALS (# CODED)	SITE *
89611						

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*PERFORMING SITE LEGEND

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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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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

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MCR      MAYO CLINIC DPT OF LAB MED & PATHOLOGY      LAB DIRECTOR:  FRANKLIN R. COCKERILL, III, M.D.
          200 FIRST STREET SW
          ROCHESTER, MN 55905
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LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME TESTING, UGTH		PATIENT NUMBER		AGE 35	SEX M	ACCESSION # G9131546
ORDERING PHYSICIAN		CLIENT ORDER #				ACCOUNT # LIAISONS
COLLECTION 07/13/09 10:33 A	RECEIVED	REPORT PRINTED 07/14/09 01:00 P		SPECIMEN INFORMATION DATE OF BIRTH:		
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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UGT1A1 Sequence, Hyperbilirubinemia**UGT1A1****MCR****Hyperbilirubinemia****Result**

A mutation was NOT detected in the UGT1A1 gene.

UGT1A1**MCR****Hyperbilirubinemia****Interp**

These results decrease the likelihood of UGT1A1 deficiency but do not rule out the diagnosis of unconjugated hyperbilirubinemia associated with UGT1A1 mutations since disease-causing mutations may be present in other regions of the gene.

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

Bidirectional DNA sequence analysis was used to test for the presence of mutations in the promoter, exons, exon-intron boundaries, and 3'-untranslated region of the UGT1A1 gene that are associated with the diagnosis of unconjugated hyperbilirubinemia.

A small percentage of individuals who have a diagnosis of unconjugated hyperbilirubinemia may have a mutation that is not identified by the methods described above.

The presence of UGT1A1 mutations does not necessarily confirm a diagnosis of unconjugated hyperbilirubinemia.

Breast-fed neonates may experience a physiologic unconjugated hyperbilirubinemia and jaundice from deconjugation of maternal bilirubin-glucuronides present in breast milk. Clinical correlation is recommended.

A genetic consultation may be of benefit.

A list of variations of unknown significance (VUS)

* Perform Site Legend on last page of report

PATIENT NAME TESTING, UGTH	ORDER STATUS Final	COLLECTION DATE AND TIME 07/13/09 10:33 A
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Specimen receipt and report times are in CST/CDT

REPRINT

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07/14/2009 1:02PM



LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME TESTING, UGTH		PATIENT NUMBER		AGE 35	SEX M	ACCESSION # G9131546
ORDERING PHYSICIAN		CLIENT ORDER #				ACCOUNT # LIAISONS
COLLECTION 07/13/09 10:33 A	RECEIVED	REPORT PRINTED 07/14/09 01:00 P		SPECIMEN INFORMATION DATE OF BIRTH:		
DATE TIME	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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identified for this patient is available from the laboratory upon request.

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

Comment

Genomic DNA was extracted.

MCR

UGT, Full Gene Sequencing

Sequencing

Performed

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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PATIENT NAME TESTING, UGTH	ORDER STATUS Final	COLLECTION DATE AND TIME 07/13/09 10:33 A
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Specimen receipt and report times are in CST/CDT

REPRINT

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