

1-800-533-1710

PATIENT NAME			PATIENT NU			AGE 58	SEX	ACCESSION #
TESTING, 89611			L3MRNW40	L3MRNW4075932				W4075932
ORDERING PHYSICIAN			CLIENT ORD	CLIENT ORDER #				ACCOUNT #
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04/22/11 11:49 A	04/22/11 1	1:49 A	04/25/11	10:32 A	DATE OF BIRTH:			
DATE TIME	DATE	TIME	DATE	TIME				
Test Client								
Attn: Mayo Liaisons								
200 First Street SW								
Rochester, MN 55905								
507-284-8202								

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LO PERFORM SITE * **TEST REQUESTED REF RANGE**

Rapid DNA Extraction REPORTED: 04/22/11 12:13 P

Comment Genomic DNA was MCR

extracted.

UGT1A1 Sequence, Hyperbilirubinemia

REPORTED: 04/22/11 12:37 P

UGT1A1 **MCR**

Hyperbilirubinemia

Result

The following mutation was identified in the UGT1A1 gene: homozygous

*28 (TA 7/7) (c.-40_-39insTA).

UGT1A1 **MCR**

Hyperbilirubinemia

Interp

Homozygosity for the *28 TA7 promoter repeat polymorphism is consistent with a diagnosis of UGT1A1-dependent unconjugated hyperbilirubinemia and Gilbert syndrome.

Reviewed by Dennis J. O'Kane, Ph.D.

MCR

This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic. It has not been cleared or approved by the U.S. Food and Drug Administration.

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

* Perform Site Legend on last page of report

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME			
TESTING, 89611	Final	04/22/11 11:49 A			

PERFORM SITE *



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TESTING, 89611			L3MRNW4075932			58		W4075932	
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COLLECTION RECEIVED		REPORT PRINTED SPECIMEN INFORMA		TION					
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Attn: Ma	ayo Liaisons								
200 Firs	t Street SW								
Rochest	er, MN 55905								
507-284	-8202								

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TEST REQUESTED LO REF RANGE

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.

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.

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
TESTING, 89611	Final	04/22/11 11:49 A