

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

Patient ID <b>SA00055167</b>	Patient Name <b>SAMPLEREPOR, UGTK</b>	Birth Date <b>1966-06-10</b>	Gender <b>F</b>	Age <b>46</b>
Order Number <b>SA00055167</b>	Client Order Number <b>SA00055167</b>	Ordering Physician <b>UNKNOWN, PROVIDER</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>15 Mar 2013 01:30</b>		

## UGT1A1 Gene, Known Mutation

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The following familial mutation was NOT detected in the UGT1A1 gene: heterozygous \*28 (TA 6/7) (c.-40\_-39insTA).

### UGT1A1 Gene, Known Mutation Interp

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The above mutation within the UGT1A1 gene was previously identified in an affected member of this family. Since this mutation was not detected in this individual, this suggests that this patient is at no greater risk than someone in the general population for having unconjugated hyperbilirubinemia.

#### ADDITIONAL INFORMATION

Bidirectional DNA sequence analysis was used to test for the presence of a specific variant(s) in the UGT1A1 gene (transcript NM\_000436.2) which was previously identified in a family member of this individual. The presence of a UGT1A1 variant or variants does not necessarily confirm a diagnosis of unconjugated hyperbilirubinemia.

Clinical correlation is recommended. A genetic consultation may be of benefit.

For information regarding pharmacogenomic genes and their associated drugs, please see the Pharmacogenomic Associations Tables on the Mayo Medical Laboratories webpage, [www.mayomedicallaboratories.com](http://www.mayomedicallaboratories.com). This resource also includes information regarding enzyme inhibitors and inducers, as well as potential alternate drug choices. Please note that the information at this link is educational material intended for health care professionals and may not be comprehensive. This educational material is not intended to supersede the care provider's

experience and knowledge of her/his patient to establish a diagnosis or a treatment plan. All medications require careful clinical monitoring. Please contact the laboratory at 1-800-533-1710 for further information about pharmacogenomic testing.

#### CAUTIONS:

Rare sequence variants may be present 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. If results obtained do not match the clinical findings (phenotype), additional testing should be considered.

Samples may contain donor DNA if obtained from patients who received heterologous blood transfusions or allogeneic blood or marrow transplantation. Results from samples obtained under these circumstances may not accurately reflect the recipient's genotype. For individuals who have received blood transfusions, the genotype usually reverts to that of the recipient within 6 weeks. For individuals who have received allogeneic blood or marrow transplantation, a pre-transplant DNA specimen is recommended for testing.

UGT1A1 genetic test results in patients who have undergone liver transplantation may not accurately reflect the patient's UGT1A1 status. Laboratory developed test.

#### Reviewed by

MCR

Jamie Bruflat

### UGT1A1 Known Mutation Sequencing

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Performed

**Received:** 15 Mar 2013 01:30

**Reported:** 28 May 2013 15:16

#### Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905