Patient ID	Patient Name		Birth Date	Gender	Age
SA00055147	SAMPLEREPORT, UGT2		1966-06-10	F	46
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00055147	SA00055147	UNKNOWN, PROVIDER			
Account Information		**Collected**			
C7028846 DLMP Rochester		15 Mar 2013 01:20			

UGT1A1 Sequence, Hyperbilirubinemia

UGT1A1 Sequence, Hyperbilirubinemia

UGT1A1 Hyperbilirubinemia Interp

MCR

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

ADDITIONAL INFORMATION

Bidirectional DNA sequence analysis was used to test for the presence of variants in the promoter, exons, exon-intron boundaries, and 3'-untranslated region of the UGT1A1 gene (transcript NM_000436.2). Some individuals who have a diagnosis of unconjugated hyperbilirubinemia may have a pathogenic variant that is not identified by the methods described above. The presence of a UGT1A1 variant 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 common, benign variants identified for this patient is available from the laboratory upon request.

For information regarding pharmacogenomic genes and their associated drugs, please see the Pharmacogenomic Associations Tables on the Mayo Medical Laboratories webpage, 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. Laboratory developed test.

UGT1A1 Hyperbilirubinemia Result

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This individual was shown to have the following mutation(s) in the UGT1A1 gene: homozygous *28(TA 7/7)(c.–40_-39insTA)

Reviewed by

MCR

Yvonne Philo

UGT, Full Gene Sequencing

MCR

Performed

Received: 15 Mar 2013 01:20 **Reported:** 23 May 2013 10:45

Performing Site Legend

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