

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

Patient ID SA00055175	Patient Name SAMPLEREPOR, UGTKO	Birth Date 1966-06-10	Gender F	Age 46
Order Number SA00055175	Client Order Number SA00055175	Ordering Physician UNKNOWN, PROVIDER	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 15 Mar 2013 01:34		

UGT1A1 Gene, Known Mutation, Saliva

UGT1A1 Gene, Known Mutation

MCR

The following UGT1A1 variant was identified: homozygous *28 (TA 7/7), nucleotide c.-40_-39insTA.

Reviewed by

MCR

Jamie Bruflat

UGT1A1 Gene, Known Mutation Interp

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The *28 TA7 promoter repeat polymorphism was previously identified in a family member affected with UGT1A1 deficiency. This individual was determined to be homozygous for the *28 TA7 promoter repeat polymorphism, which 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 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. 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.

UGT1A1 Known Mutation Sequencing

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Performed

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Performing Site Legend

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