

Patient ID SA00057737	Patient Name SAMPLEREPOR, UGTI A	Birth Date 1966-06-10	Gender F	Age 46
Order Number SA00057737	Client Order Number SA00057737	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 23 May 2013 11:05		

UGT1A1 Gene Sequence, Irinotecan

UGT1A1 Gene Sequence, Irinotecan

UGT1A1 Irinotecan Result

MCR

This individual was shown to have the following mutation(s) in the UGT1A1 gene: heterozygous *28 (TA 6/7) (c.-40_-39insTA).

UGT1A1 Irinotecan Interp

MCR

Heterozygosity for the *28 TA7 promoter repeat polymorphism is associated with increased risk (approximately 1 in 12) for severe neutropenia when treated with the standard doses of irinotecan. The drug labeling should be consulted for dose recommendations.

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 experience an adverse outcome with a drug metabolized by UGT1A1, may have a variant that is not identified by the methods described above. A list of common, benign variants identified for this patient is available from the laboratory upon request.

Drug metabolism maybe affected not only by variants in the UGT1A1 gene, but also by other drug-drug interactions.

Avoid the concomitant use of strong CYP3A4 inhibitors and inducers. Irinotecan, atazanavir, nilotinib and pazopanib undergo metabolism by CYP3A4 and concomitant use of these drugs with strong CYP3A4 inhibitors and inducers may significantly increase or decrease drug concentrations. For drug-drug interactions, CYP3A4 inhibitors and inducers, and dosing recommendations, see drug labels.

For additional 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 false 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 irinotecan metabolites. 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

Performing Site Legend

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



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UGT, Full Gene Sequencing

MCR

Performed

Received: 28 May 2013 12:58

Reported: 28 May 2013 13:41

QA
Environment

Performing Site Legend

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