
Lab Dept: Anatomic Pathology

Test Name: UDP-GLUCURONOSYL TRANSFERASE 1A1
(UGT1A1) SEQUENCING, IRINOTECAN
SENSITIVITY

General Information

Lab Order Codes: UGT1

Synonyms: GNT1, Irinotecan glucuronidation, Irinotecan metabolism, UDP-Glucuronosyltransferase 1, UGT1A1, Uracil Glucuronyl transferase, Uridine Diphosphate Glucosyltransferase

CPT Codes: 81350 – UGT1A1, gene analysis, common variants

Test Includes: UGT1A1 Gene Sequence, Irinotecan Sensitivity; UGT Full Gene Sequencing; DNA Extraction

Logistics

Test Indications: Useful for identifying individuals who are at increased risk of adverse drug reactions with irinotecan and who should be considered for decreased dosing of the drug.

Lab Testing Sections: Anatomic Pathology – Sendouts

Referred to: Mayo Medical Laboratories (MML Test: UGT1)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 7 - 14 days; testing performed Monday - Friday

Special Instructions:

Bone Marrow and liver transplants will interfere with testing.

Multiple drug metabolism genotype tests can be performed on a single specimen after a single extraction.

Transfusions will interfere with testing for up to 4-6 weeks. DNA obtained from white cells may not provide useful information for patients who received a recent transfusion of blood that was not leukocyte-reduced. Wait 4-6 weeks until transfused cells have left the patient's circulation before drawing the patient's blood specimen for genotype testing.

Form required for testing (please send with the specimen or patient to the laboratory):

[Informed Consent for DNA Testing](#)

Specimen

Specimen Type:	Whole Blood
Container:	Lavender top (EDTA) tube
Draw Volume:	3 mL (Minimum: 0.3 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine Venipuncture
Special Processing:	Lab Staff: Do Not Centrifuge. Specimen should remain in the original collection container. Store and ship at room temperature. A consent form (filled out by ordering provider) should be sent with the specimen.
Patient Preparation:	None
Sample Rejection:	Clotted sample; mislabeled or unlabeled specimens; incorrect specimen type

Interpretive

Reference Range:	An interpretive report will be provided.
Critical Values:	N/A

Limitations:

Blood transfusions or bone marrow transplantation prior to having blood drawn for DNA analysis can generate false results as DNA in the specimen may be a mix of patient or donor DNA.

An alternative splice site for exon 5 (referred to as exon 5b) has been discovered and described in the literature. This new exon is described to have a decrease in enzymatic activity (compared with exon 5a: previously known as exon 5), but little is known about the frequency of exon 5b or how it impacts bilirubinemia. Concurrently, Mayo is not testing or sequencing exon 5b and will continue to monitor the literature for new information on exon 5b.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Methodology:

Polymerase Chain Reaction (PCR) Followed by Gene Sequencing

References:

[Mayo Medical Laboratories](#) August 2010
(800) 533-1710

Updates:

2/11/2013: CPT update