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**Lab Dept:** Anatomic Pathology

**Test Name:** UDP-GLUCURONOSYL TRANSFERASE 1A1  
(UGT1A1) SEQUENCING, HYPERBILIRUBINEMIA

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**General Information**

**Lab Order Codes:** UGTA

**Synonyms:** GNT1, Phenol/Bilirubin UDP-Glucuronosyltransferase, UDP-Glucuronyltransferase 1, UGT1A1, Uracil Glucuronyl transferase, Uridine Diphosphate Glucosyltransferase 1; Crigler-Najjar Syndrome; Gilbert Syndrome

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

**Test Includes:** UGT1A1 Gene Sequence, Hyperbilirubinemia; UGT Full Gene Sequencing; DNA Extraction

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**Logistics**

**Test Indications:** Useful for identifying individuals who are at risk for hyperbilirubinemia; confirmation of a diagnosis of Gilbert or Crigler-Najjar syndromes; verification of carrier status for Gilbert or Crigler-Najjar syndromes.

**Lab Testing Sections:** Anatomic Pathology – Sendouts

**Referred to:** Mayo Medical Laboratories (MML Test: UGT2)

**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:** Patients who have received a heterologous blood transfusion within the preceding 6 weeks, or who have received a allogeneic blood or marrow transplant, can have inaccurate genetic test results due to the presence of donor DNA.

For bone marrow transplant patients, buccal cells from the **recipient** should be provided to obtain an accurate genotype.

For liver transplant patients, **donor** blood or buccal cells should be provided to obtain an accurate genotype for the recipient patient.

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.

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

1. Patient Information Sheet (Mayo Supply T664):

[http://www.mayomedicallaboratories.com/it-mmfiles/UGT1A1\\_Gene\\_Testing\\_Pt\\_Info\\_Sheet\\_mc1235-165.pdf](http://www.mayomedicallaboratories.com/it-mmfiles/UGT1A1_Gene_Testing_Pt_Info_Sheet_mc1235-165.pdf)

2. [Informed Consent for DNA Testing](#)

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### ***Specimen***

<b>Specimen Type:</b>	Whole Blood
<b>Container:</b>	Lavender top (EDTA) tube
<b>Draw Volume:</b>	Adults: 3 mL , Pediatrics: 1 mL (Minimum: 0.3 mL) blood
<b>Processed Volume:</b>	Same as Draw Volume
<b>Collection:</b>	Routine Venipuncture
<b>Special Processing:</b>	Lab Staff: Do Not Centrifuge. Specimen should remain in the original collection container. Store and ship at room temperature. A consent form and patient information sheet (filled out by ordering provider) should be sent with the specimen.  Note: If submitting a microtainer, place inside a larger tube or vial for transport.
<b>Patient Preparation:</b>	N/A
<b>Sample Rejection:</b>	Clotted sample; mislabeled or unlabeled specimens; incorrect specimen type

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### ***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.

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

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.

Absence of a detectable gene mutation or polymorphism does not rule out the possibility that the patient may have a genetic cause for increased unconjugated bilirubin.

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](#) October 2014  
(800) 533-1710

**Updates:**

8/17/2010: ACD is no longer an acceptable specimen type. Addition of Patient information form.  
2/11/2013: CPT update