Lab Dept: Anatomic Pathology

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

General Information

Lab Order Codes: UGTFG

Synonyms: UGT1A1 Full Gene Sequencing

CPT Codes: 81479 – Molecular Pathology, Unlisted

Test Includes: UGT Full Gene Sequencing

Logistics

Test Indications: Identifying individuals who are at risk of adverse drug reactions with drugs metabolized by UGT1A1, including irinotecan, atazanavir, nilotinib, pazopanib, and belinostat.

Identifying individuals who are at risk of hyperbilirubinemia.

Follow-up testing for individuals with a suspected UGT1A1 variant, who had negative TA repeat region testing.

Establishing a diagnosis of Gilbert, Crigler-Najjar syndrome type I or type II.

Establishing carrier status for Gilbert, Crigler-Najjar syndrome type I or type II.

Lab Testing Sections: Anatomic Pathology – Sendouts

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

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 7 - 14 days; testing performed Tuesday

Special Instructions: See Limitations:

Forms required for testing (please send with the specimen or patient to the laboratory):
1. Patient Information Sheet (Mayo Supply T664):
2. **Informed Consent for DNA Testing**

**Specimen**

**Specimen Type:** Whole Blood

**Container:** Lavender top (EDTA) tube

**Draw Volume:** Adults: 3 mL, Pediatrics: 1 mL (Minimum: 0.45 mL) blood

**Processed Volume:** Same as Draw Volume

**Collection:** Routine blood collection. Invert several times to mix.

**Special Processing:** 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.

**Patient Preparation:** N/A

**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:** 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 allogeneic blood or marrow transplantation, a pretransplant 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.

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.

**Methodology:** Polymerase Chain Reaction (PCR) Followed by Gene Sequencing