MAYO CLINIC LABORATORIES

TEST CHANGE

Notification Date: May 6, 2025
Effective Date: May 8, 2025

UDP-Glucuronosyltransferase 1A1 TA Repeat Genotype, *UGT1A1*, Varies

Test ID: U1A1Q

Explanation: On the effective date the Ordering Guidance, Specimen Required, Cautions, and Times Performed will be updated for this assay.

Current Ordering Guidance	New Ordering Guidance
This test does not detect or report variants other than the *1 (TA6), *28 (TA7), *36 (TA5), and *6 (c.211G>A) alleles. The *37 (TA8) allele cannot be distinguished from *28 (TA7) and will be reported as *28 (TA7) by this methodology. Numerous variants outside of the TA repeat region have been described that impair UGT1A1 activity. Sequencing of the full gene is available for detection of variants outside of the TA repeat region; order UGTFZ / UDP-Glucuronosyltransferase 1A1 (UGT1A1), Full Gene Sequencing, Varies. If Crigler-Najjar syndrome testing is requested, order UGTFZ / UDP-Glucuronosyltransferase 1A1 (UGT1A1), Full Gene Sequencing, Varies. For more information on test ordering, see *UGT1A1* Test-Ordering Algorithm.	This test does not detect or report variants other than the *1 (TA6), *28 (TA7), *36 (TA5), and *6 (c.211G>A) alleles. The *37 (TA8) allele cannot be distinguished from *28 (TA7) and will be reported as *28 (TA7) by this methodology. Numerous variants outside of the TA repeat region have been described that impair UGT1A1 activity. Sequencing of the full gene is available for detection of variants outside of the TA repeat region; order UGTFZ / UDP-Glucuronosyltransferase 1A1 (UGT1A1), Full Gene Sequencing, Varies. If Crigler-Najjar syndrome is suspected, order UGTFZ / UDP-Glucuronosyltransferase 1A1 (UGT1A1), Full Gene Sequencing, Varies. For more information on test ordering, see *UGT1A1* Test-Ordering Algorithm. Multiple genotype tests can be performed on a single specimen after a single extraction. See Multiple Genotype Test List for a list of tests that can be ordered together.

Current Specimen Required	New Specimen Required
Multiple genotype tests can be performed on a single specimen after a single extraction. See Multiple Genotype Test List for a list of tests that can be ordered together. **Submit only 1 of the following specimens:** **Specimen Type: Whole blood** **Container/Tube:** Lavender top (EDTA) **Specimen Volume:** 3 mL **Collection Instructions:** 1. Invert several times to mix blood. 2. Send whole blood specimen in original tube. Do not aliquot.	**Patient Preparation:** A previous hematopoietic stem cell transplant from an allogenic donor will interfere with testing. For information about testing patients who have received a hematopoietic stem cell transplant, call 800-533-1710. **Submit only 1 of the following specimens:** **Specimen Type: Whole blood** **Container/Tube:** Lavender top (EDTA) **Specimen Volume:** 3 mL **Collection Instructions:** 1. Invert several times to mix blood. 2. Send whole blood specimen in original tube. Do not aliquot.

Specimen Stability Information: Ambient (preferred) 9 days/Refrigerated 30 days
Additional Information: To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled if DNA requirements are inadequate.

Specimen Type: Saliva
Patient Preparation: Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.
Supplies: Saliva Swab Collection Kit (T786)
Specimen Volume: 1 Swab
Collection Instructions: Collect and send specimen per kit instructions.
Specimen Stability Information: Ambient 30 days
Additional Information: Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

Specimen Type: Extracted DNA
Container/Tube: 2-mL screw top tube
Specimen Volume: 100 mcL (microliters)
Collection Instructions:
1. The preferred volume is 100 mcL at a concentration of 50 ng/mcL.
2. Provide concentration of DNA and volume on tube.
Specimen Stability Information: Frozen (preferred)/Ambient/Refrigerated
Additional Information: DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days
Additional Information:
1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
2. To ensure minimum volume and concentration of DNA is met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.

Specimen Type: Cord blood
Container/Tube: Lavender top (EDTA)
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube. Do not aliquot.
Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days
Additional Information:
1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
2. To ensure minimum volume and concentration of DNA is met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.
3. While a properly collected cord blood sample may not be at risk for maternal cell contamination, unanticipated complications may occur during collection.
Therefore, maternal cell contamination studies are recommended to ensure the test results reflect that of the patient tested and are available at an additional charge. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type: Saliva
Patient Preparation: Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.
Supplies: Saliva Swab Collection Kit (T786)
Specimen Volume: 1 Swab
Collection Instructions: Collect and send specimen per kit instructions.
Specimen Stability Information: Ambient (preferred) 30 days/Refrigerated 30 days
Additional information: Saliva specimens are acceptable but not recommended. Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

Specimen Type: Extracted DNA
Container/Tube:
Preferred: Screw Cap Micro Tube, 2 mL with skirted conical base
Acceptable: Matrix tube, 1 mL
Collection Instructions:
1. The preferred volume is at least 100 mcL at a concentration of 75 ng/mcL.

	2. Include concentration and volume on tube. **Specimen Stability Information:** Frozen (preferred) 1 year/Ambient/Refrigerated **Additional Information:** DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

Current Cautions	New Cautions
Samples may contain donor DNA if obtained from patients who received non-leukoreduced blood transfusions or allogeneic hematopoietic stem cell 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 hematopoietic stem cell 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. Liver or kidney dysfunction may result in adverse drug reactions with irinotecan independently of thymine-adenine (TA)-repeat variants.	This test is a genotyping test that evaluates 3 common variants in the *UGT1A1* gene only. It is important to note that patients with a negative test may have a rare variant resulting in increased risk of irinotecan and other drug toxicity that is not detected by this test. A sequencing assay is available that can detect rare variants located in the exons of *UGT1A1*; however, it will not detect copy number variations; see UGTFZ / UDP-Glucuronosyltransferase 1A1 (UGT1A1), Full Gene Sequencing, Varies. Additionally, *UGT1A1* sequencing with copy number analysis is available through custom gene ordering; see CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies. Samples may contain donor DNA if obtained from patients who received non-leukoreduced blood transfusions or allogeneic hematopoietic stem cell transplantation. Results from samples obtained under these circumstances may not accurately reflect the recipient's genotype. For individuals who have received non-leukoreduced blood transfusions, the genotype usually reverts to that of the recipient within 6 weeks. For individuals who have received allogeneic hematopoietic stem cell 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. Liver or kidney dysfunction may result in adverse drug reactions with irinotecan independently of thymine-adenine (TA)-repeat variants.

Current Times Performed	New Times Performed
8 a.m.	Varies

Questions

Contact Michelle Raths, Laboratory Resource Coordinator at 800-533-1710.