

UDP-Glucuronosyl Transferase 1A1 TA Repeat Genotype, UGT1A1, Varies

Test ID: U1A1Q

Useful for:

Identifying individuals who are at increased risk of adverse drug reactions with drugs that are metabolized by UGT1A1; especially irinotecan but also atazanavir, nilotinib, pazopanib, and belinostat

Identifying individuals with Gilbert syndrome due to the presence of homozygous *UGT1A1**6 (c.211G>A, based on NM_000463.2) allele, TA7, homozygous TA8, or compound heterozygous *6, TA7 or TA8

Identifying individuals who are carriers of Gilbert syndrome due to the presence of heterozygous TA7 or TA8

Methods:

Real-Time Polymerase Chain Reaction (PCR) with Allelic Discrimination Analysis

Advisory:

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 also available for detection of variants outside of the TA repeat region; order UGTFG / UDP-Glucuronosyl Transferase 1A1 (UGT1A1), Full Gene Sequencing.

Reference Values:

An interpretive report will be provided.

Specimen Stability Information:

Specimen Type	Temperature	Time
Varies	Varies	

Specimen Requirements:

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

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 specimen in original tube.

Specimen Stability Information: Ambient (preferred) 9 days/Refrigerated 30 days

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

Specimen Type: 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. Include concentration and volume on tube.

Specimen Stability Information: Frozen (preferred)/Ambient/Refrigerated

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 renal dysfunction may result in adverse drug reactions with irinotecan independently of thymine-adenine (TA)-repeat variants.

CPT Code:

81350-*UGT1A1* (UDP glucuronosyltransferase 1 family, polypeptide A1) (eg, irinotecan metabolism), gene analysis, common variants (eg, *28, *36, *37)

Analytic Time: 3 days (not reported on Saturday or Sunday)

Day(s) Setup: Monday through Friday

Questions:

Contact your Laboratory Technologist Resource Coordinator Heather Flynn Gilmer at 800-533-1710.