
Reporting Title: UGT1A1 TA Repeat Genotype, V**Performing Location:** Rochester**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 UGTFG / UDP-Glucuronosyltransferase 1A1 (UGT1A1), Full Gene Sequencing, Varies.

If Crigler-Najjar syndrome testing is requested, order UGTFG / UDP-Glucuronosyltransferase 1A1 (UGT1A1), Full Gene Sequencing, Varies.

For more information on test ordering, see UGT1A1 Test-Ordering Algorithm.

Specimen Requirements:

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.

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: 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

Specimen Minimum Volume:

Blood: 0.4 mL

Saliva, extracted DNA: see Specimen Required

Forms:

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

2. If not ordering electronically, complete, print, and send 1 of the following forms with the specimen:

-Therapeutics Test Request (T831)

-Oncology Test Request (T729)

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
610168	UGT1A1 Genotype	Alphanumeric		93845-6
610169	UGT1A1 Phenotype	Alphanumeric		79718-3
610170	Interpretation	Alphanumeric		69047-9
610171	Additional Information	Alphanumeric		48767-8
610172	Method	Alphanumeric		85069-3
610173	Disclaimer	Alphanumeric		62364-5
610174	Reviewed by	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

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

Reference Values:

An interpretive report will be provided.