
Reporting Title: TPMT and NUDT15 Genotype, V**Performing Location:** Rochester**Ordering Guidance:**

For thiopurine methyltransferase (TPMT) enzyme activity testing, order TPMT3 / Thiopurine Methyltransferase Activity Profile, Erythrocytes; however, this test should also be ordered because TPMT enzyme activity testing cannot detect variants in NUDT15, which also impact thiopurine metabolism.

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

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:
 - Neurology Specialty Testing Client Test Request (T732)
 - Gastroenterology and Hepatology Test Request (T728)
 - Therapeutics Test Request (T831)

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
610159	TPMT Genotype	Alphanumeric		41048-0
610160	TPMT Phenotype	Alphanumeric		79713-4
610161	NUDT15 Genotype	Alphanumeric		93194-9
610162	NUDT15 Phenotype	Alphanumeric		93195-6
610163	Interpretation	Alphanumeric		69047-9
610164	Additional Information	Alphanumeric		48767-8
610165	Method	Alphanumeric		85069-3
610166	Disclaimer	Alphanumeric		62364-5
610167	Reviewed by	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

0034U

Reference Values:

An interpretive report will be provided.