

# **Test Definition: TPNUQ**

Thiopurine Methyltransferase (TPMT) and Nudix Hydrolase (NUDT15) Genotyping, Varies

# **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 <u>Multiple Genotype Test List</u> 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:

Invert several times to mix blood.
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:

The preferred volume is 100 mcL at a concentration of 50 ng/mcL.
Provide concentration of DNA and volume on tube.

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

# Forms:

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:

 <u>Informed Consent for Genetic Testing</u> (T576)
 <u>Informed Consent for Genetic Testing-Spanish</u> (T826)

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

 <u>Neurology Specialty Testing Client Test Request</u> (T732)

-Gastroenterology and Hepatology Test Request (T728)



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#### -<u>Therapeutics Test Request</u> (T831)

Specimen Type	Temperature	Time	Special Container	
Varies	Varies			

### **Result Codes:**

Result ID	Reporting Name	Туре	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<sup>®</sup> and CPT codes are provided by the performing laboratory.

### Supplemental Report:

No

# **CPT Code Information:**

0034U

# **Reference Values:**

An interpretive report will be provided.