



MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT

NOTIFICATION DATE: March 10, 2014

EFFECTIVE DATE: March 11, 2014

THIOPURINE METHYLTRANSFERASE (TPMT) GENOTYPING, BLOOD Test ID: GTPMT

USEFUL FOR: Predicting potential for toxicity to thiopurine drugs (6-mercaptopurine, 6-thioguanine, and azathioprine)

METHODOLOGY: Polymerase Chain Reaction (PCR) 5'-Nuclease End-Point Allelic Discrimination Analysis

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS:

Multiple whole blood EDTA genotype tests can be performed on a single specimen after a single extraction. See Multiple Whole Blood EDTA Genotype Tests in Special Instructions for a list of tests that can be ordered together.

Container/Tube: Lavender top (EDTA)

Specimen Volume: 3 mL

Collection Instructions: Send specimen in original tube.

Forms: **New York Clients-Informed consent is required.** Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Whole Blood EDTA	Ambient (preferred)	
	Refrigerated	

CAUTIONS:

Patients who have received a heterologous blood transfusion within the preceding 6 weeks, or who have received an allogeneic blood or marrow transplant, can have inaccurate genetic test results due to presence of donor DNA.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If genotype results obtained do not match the clinical findings, additional testing should be considered (TPMT / Thiopurine Methyltransferase [TPMT], Erythrocytes).

The results do not rule out the possibility that a patient harbors another variation that can impact drug response or drug side effects. These genotyping procedures will not distinguish between heterozygous *3A from the rare *3B/ *3C, which has a frequency of 1:120,890. This rare genotype is associated with low enzyme activity. Enzyme activity evaluation is necessary to definitively identify this rare genotype (TPMT / Thiopurine Methyltransferase [TPMT], Erythrocytes).

This PCR-based assay tests for the presence of *TPMT*2* (c.238G>C), *TPMT*3A* (c.460G>A, c.719A>G), *TPMT*3B* (c.460G>A), and *TPMT*3C* (c.719A>G). This test will not detect all *TPMT* genetic variants. A negative result does not rule out the possibility of toxicity if thiopurines are used, since multiple factors (eg, other genetic factors, drug-drug interactions) are known to play a role. Coprescription of allopurinol might inhibit TPMT activity. Drugs that have been shown to inhibit TPMT activity include: naproxen, ibuprofen, ketoprofen, furosemide, sulfasalazine, mesalamine, olsalazine, mefenamic acid, thiazide diuretics, and benzoic acid inhibitors.

CPT CODE: 81401

DAY(S) SET UP: Monday through Friday **ANALYTIC TIME:** 1 day

NOTE: The following referral test code will become obsolete effective April 10th, 2014.

Test Name	Test ID	Referral Lab Code	Referral Lab
Prometheus TPMT Genetics	FPRTG	3300	Prometheus

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or
Brandon DeBoom, MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710