

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB: 2/20/1992
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

TPMT Genotyping

ARUP test code 3017372

TPMT Genotype Specimen whole Blood

TPMT Genotype ***3B/*3B** *

TPMT Predicted Phenotype **Poor** *

TPMT Interpretation See Note

Two no function alleles were identified in the TPMT gene, suggesting a poor metabolizer phenotype and susceptibility to dose-related toxicity from standard doses of thiopurine drugs. A substantial dose reduction of thiopurine drugs may be required. See drug labeling and clinical consensus guidelines for more details about dosing.

Recommendation: Guidelines for genotype - based dosing are published by the Clinical Pharmacogenetics Implementation Consortium(CPIC) and can be found at: <https://cpicpgx.org/> and <https://www.pharmgkb.org/>.

This result has been reviewed and approved by [REDACTED]

H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: TPMT Genotyping

CHARACTERISTICS: Thiopurine drug therapy is used for autoimmune diseases, inflammatory bowel disease, acute lymphoblastic leukemia, and to prevent rejection after solid organ transplant. The inactivation of thiopurine drugs is catalyzed in part by thiopurine methyltransferase (TPMT). Variants in the TPMT gene are associated with an accumulation of cytotoxic metabolites leading to increased risk of drug-related toxicity with standard doses of thiopurine drugs. These effects on thiopurine catabolism can be additive.

INHERITANCE: Autosomal codominant.

CAUSE: TPMT variants affect enzyme activity.

VARIANTS TESTED:

(Variants are numbered according to NM_000367 transcript for TPMT)

*1: Indicative of no detected targeted variants and an assumption of functional allele.

TPMT*2: rs1800462, c.238G>C

TPMT*3A: rs1800460, c.460G>A; rs1142345, c.719A>G

TPMT*3B: rs1800460, c.460G>A

TPMT*3C: rs1142345, c.719A>G

TPMT*4: rs1800584, c.626-1G>A

CLINICAL SENSITIVITY: 95 percent.

METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.

ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.

LIMITATIONS: Only the targeted TPMT variants will be detected by this test. Because the complex TPMT*3A allele contains the variants found in the *3B and *3C alleles, this test cannot distinguish the 3A/Negative genotype (intermediate enzyme activity) from the rare *3B/*3C genotype (no or low enzyme activity). Genotyping may reflect donor status in patients who have received allogenic stem cell or bone marrow transplants within 2 weeks of specimen collection. Actual enzyme activity and expression and risk for adverse reactions to thiopurines may be affected by additional genetic and non-genetic factors not evaluated by this test. Diagnostic errors can occur due to rare sequence variations. Genotyping does not replace the need for therapeutic drug monitoring and clinical observation.

Please note the information contained in this report does not contain medication recommendations, and should not be interpreted as recommending any specific medications. Any dosage adjustments or other changes to medications should be evaluated in consultation with a medical provider.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

EER TPMT

See Note

[REDACTED]

H=High, L=Low, *=Abnormal, C=Critical

VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
TPMT Genotype Specimen	24-052-121307	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
TPMT Genotype	24-052-121307	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
TPMT Predicted Phenotype	24-052-121307	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
TPMT Interpretation	24-052-121307	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER TPMT	24-052-121307	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 24-052-121307
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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