ARUP LABORATORIES | aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Jonathan R. Genzen, MD, PhD, Chief Medical Officer

PATIENT REPORT

Patient Age/Sex: 37 years Female

Specimen Collected: 30-Jan-24 07:32

TPMT Genotyping |**Received: 30-Jan-24 08:22** **Report/Verified: 30-Jan-24 10:47**

Procedure	Result	Units	Reference Interval
TPMT Genotype Specimen	Whole Blood		
TPMT Genotype	***3A/*3A** *		
TPMT Predicted Phenotype	**Poor** *		
TPMT Interpretation	See Note f1 i1		
EER TPMT	EERUnavailable f2		

Result Footnote

f1: TPMT Interpretation

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.

This result has been reviewed and approved by [redacted]

f2: EER TPMT

[redacted]

Test Information

i1: TPMT Interpretation

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 methyltrasferase (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

Test Information

i1: TPMT Interpretation

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.