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PATIENT REPORT

Patient Age/Sex: Female

Specimen Collected: 12-Sep-23 13:24

TPMT and NUDT15 | **Received: 12-Sep-23 13:30** **Report/Verified: 14-Sep-23 12:47**

Procedure	Result	Units	Reference Interval
TPMT Predicted Phenotype	Normal		
TPMT2 Specimen	Whole Blood		
TPMT Genotype	*1/*1		
NUDT15 Genotype	*1/*1		
NUDT15 Phenotype	Normal		
TPMT2 Interpretation	See Note f1 i1		
EER TPMT and NUDT15	See Note f2		

Result Footnote

f1: TPMT2 Interpretation

No variant alleles were identified, suggesting a normal metabolizer phenotype and that standard doses of thiopurines are appropriate. See drug labeling and clinical consensus guidelines for more details about dosing.

This result has been reviewed and approved by [redacted]

f2: EER TPMT and NUDT15
Authorized individuals can access the ARUP Enhanced Report using the following link:

[redacted]

Test Information

i1: TPMT2 Interpretation
BACKGROUND INFORMATION: TPMT and NUDT15

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) and nudix hydrolase 15 (NUDT15). Variants in the TPMT and/or NUDT15 genes 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 and NUDT15 variants affect enzyme expression or activity.
VARIANTS TESTED:
(Variants are numbered according to NM_000367 transcript for TPMT and the NM_018283 transcript for NUDT15)

*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

Test Information

i1: TPMT2 Interpretation

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

NUDT15 *2 or *3: rs116855232, c.415C>T
NUDT15*4: rs147390019, c.416G>A

CLINICAL SENSITIVITY: 95 percent.
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Only the targeted TPMT and NUDT15 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.