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PATIENT REPORT

Patient Age/Sex: 38 years Female

Specimen Collected: 30-Jan-24 07:36

NUDT15 Genotyping **|Received: 30-Jan-24 08:22** **Report/Verified: 30-Jan-24 12:36**

Procedure	Result	Units	Reference Interval
NUDT15 Specimen	DNA		
NUDT15 Genotype	***2or*3/*2or*3** *		
NUDT15 Phenotype	**Poor** *		
NUDT15 Interpretation	See Note f1 i1		
EER NUDT15	See Note f2		

Result Footnote

f1: NUDT15 Interpretation

Two no function alleles were identified in the NUDT15 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 NUDT15

[redacted]

Test Information

i1: NUDT15 Interpretation

BACKGROUND INFORMATION: NUDT15 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 nudix hydrolase 15 (NUDT15). Variants in the NUDT15 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: NUDT15 variants affect metabolism of thiopurines and tolerance to the treatment.

VARIANTS TESTED:
(Variants are numbered according to NM_018283 transcript for NUDT15)

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

NUDT15 *2 or *3: rs116855232, c.415C>T

Test Information

i1: NUDT15 Interpretation

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 NUDT15 variants will be detected by this test. 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.