

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

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 2/20/1990  
**Gender:** Female  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** 00/00/0000 00:00

**NUDT15 Genotyping**

ARUP test code 3017373

NUDT15 Specimen	whole Blood
NUDT15 Genotype	<b>*2or*3/*2or*3</b> *
NUDT15 Phenotype	<b>Poor</b> *
NUDT15 Interpretation	See Note

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.

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: 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

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 allogeneic 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 NUDT15

See Note

[REDACTED]

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
NUDT15 Specimen	24-052-121556	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NUDT15 Genotype	24-052-121556	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NUDT15 Phenotype	24-052-121556	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NUDT15 Interpretation	24-052-121556	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER NUDT15	24-052-121556	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: 24-052-121556  
Patient Identifiers: 01234567890ABCD, 012345  
Visit Number (FIN): 01234567890ABCD  
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