

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 2/20/1991
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 *1/*1

NUDT15 Phenotype Normal

NUDT15 Interpretation See Note

No variant alleles were identified in the NUDT15 gene, 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.

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	24-052-121411	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NUDT15 Genotype	24-052-121411	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NUDT15 Phenotype	24-052-121411	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NUDT15 Interpretation	24-052-121411	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER NUDT15	24-052-121411	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: 24-052-121411
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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