

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: ██████████
Gender: Unknown
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/*14

NUDT15 Phenotype Intermediate

NUDT15 Interpretation

See Note

One possible no-function allele was identified in the NUDT15 gene, suggesting an intermediate metabolizer phenotype and susceptibility to dose-related toxicity from standard doses of thiopurine drugs. 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 ██████████

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

Unless otherwise indicated, testing performed at:

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
NUDT15*14: rs777311140, c.80_81insCGGG

METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.

ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 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 nongenetic 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 U.S. 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

Authorized individuals can access the ARUP Enhanced Report with an ARUP Connect account using the following link.

Your local lab can assist you in obtaining the patient report if you don't have a connect account.



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VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
NUDT15 Specimen	25-085-101779	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NUDT15 Genotype	25-085-101779	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NUDT15 Phenotype	25-085-101779	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
NUDT15 Interpretation	25-085-101779	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER NUDT15	25-085-101779	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: