

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: [REDACTED]/1994
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Dihydropyrimidine Dehydrogenase (DPYD)

ARUP test code 2012166

DPYD Specimen	whole Blood		
DPYD Allele 1	*1		
DPYD Allele 2	*13	*	
DPYD Phenotype	Intermediate	*	
DPYD Interpretation	<p>See Note</p> <p>Activity Score: 1.5</p> <p>Interpretation: The following DPYD allele(s) were detected: *1/*13. This result predicts the intermediate metabolizer phenotype for dihydropyrimidine dehydrogenase (DPD). Because 80 percent of administered 5-fluorouracil (5-FU) is normally inactivated by DPD, a decrease in DPD enzymatic activity may lead to increased concentrations of 5-FU and elevated risk for grade III-IV toxicity.</p> <p>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/.</p> <p>This result has been reviewed and approved by [REDACTED]</p>		

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

BACKGROUND INFORMATION: Dihydropyrimidine Dehydrogenase (DPYD)

CHARACTERISTICS: 5-fluorouracil (5-FU) is the most frequently used chemotherapeutic drug for the treatment of many types of cancer, particularly colorectal adenocarcinoma. Grade III-IV drug toxicity attributed to 5-FU occurs in approximately 16 percent of patients, and may include hematologic, gastrointestinal, and dermatologic complications. In some cases, this toxicity can cause death. When 5-FU is metabolized in the body, approximately 80 percent is catabolized by the dihydropyrimidine dehydrogenase (DPD) enzyme. Variants in the DPYD gene can lead to reduced 5-FU catabolism, resulting in the aforementioned toxicity complications.

INHERITANCE: Autosomal codominant.

CAUSE: DPYD gene mutations.

DPYD Variants Tested:

(Variants are numbered according to NM_000110 transcript)

Nonfunctional alleles and increased toxicity risk:

c.1024G>A (rs183385770)
c.1774C>T (rs59086055)
*13 (c.1679T>G, rs55886062)
*2A (c.1905+1G>A, rs3918290)

Decreased function alleles and increased toxicity risk:

c.557A>G (rs115232898)
c.868A>G (rs146356975)
c.2279C>T (rs112766203)
c.2846A>T (rs67376798)
c.1129-5923C>G (rs75017182)

Functional alleles and normal enzymatic activity:

*1 indicates no variants detected.

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

ANALYTICAL SENSITIVITY and SPECIFICITY: Greater than 99 percent. **LIMITATIONS:** Only the targeted DPYD variants will be detected by this panel. Diagnostic errors can occur due to rare sequence variations. 5-FU drug metabolism, efficacy, and risk for toxicity may be affected by genetic and nongenetic factors that are not evaluated by this test. Genotyping does not replace the need for therapeutic drug monitoring or 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 Dihydropyrimidine Dehydrogenase**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.

[REDACTED]

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VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
DPYD Specimen	25-219-115792	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
DPYD Allele 1	25-219-115792	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
DPYD Allele 2	25-219-115792	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
DPYD Phenotype	25-219-115792	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
DPYD Interpretation	25-219-115792	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
EER Dihydropyrimidine Dehydrogenase	25-219-115792	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