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500 Chipeta Way, Salt Lake City, Utah 84108-1221 phone: 801-583-2787, toll free: 800-522-2787 Jonathan R. Genzen, MD, PhD, Chief Medical Officer

Patient Age/Sex: 36 years Female

Specimen	Collected:	5/6/2025	08:17 MDT	
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Dihydropyrimidine Dehydrogenase (DPYD)	Received: 5/6/2025 08	:18 MDT	Report/Verified: 5/7/2025 10:49 MDT
Procedure	Result	Units	Reference Interval
DPYD Specimen	Whole Blood		
DPYD Genotype	Cmpnd Hetero *		
DPYD Allele 1	*2A *		
DPYD Allele 2	*13 *		
DPYD Phenotype	Poor *		
DPYD Interpretation	See Note ^{f1 i1}		
EER Dihydropyrimidine	See Note ^{f2}		
Dehydrogenase			

Result Footnote

f1: DPYD Interpretation

Activity Score: 0

Interpretation: The following DPYD allele(s) were detected: *2A/*13. This result predicts the poor metabolizer phenotype for dihydropyrimidine dehydrogenase (DPD). Because 80 percent of administered 5-fluorouracil (5-FU) is normally inactivated by DPD, the significant reduction of DPD activity may markedly increase concentrations of 5-FU, placing the patient at substantially increased risk for grade III-IV toxicity.

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 EER Dihydropyrimidine Dehydrogenase 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.

Test Information

il: DPYD Interpretation

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.

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Unless otherwise indicated, testing performed at: ARUP Laboratories 500 Chipeta Way, Salt Lake City, UT 84108

500 Chipeta Way, Salt Lake City, UT 84108 Laboratory Director: Jonathan R. Genzen, MD, PhD
 ARUP Accession:
 25-126-900026

 Report Request ID:
 20431780

 Printed:
 5/8/2025 11:49 MDT

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Patient Age/Sex: 36 years Female

Test Information i1: DPYD Interpretation 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.

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