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PATIENT REPORT

Patient Age/Sex: Female

Specimen Collected: 12-Sep-23 13:26

Dihidropyrimidine Dehydrogenase (DPYD) | **Received: 12-Sep-23 13:30** **Report/Verified: 12-Sep-23 15:13**

Procedure	Result	Units	Reference Interval
EER Dihydropyrimidine Dehydrogenase	See Note [f1]		
DPYD Specimen	Whole Blood		
DPYD Genotype	*1/*1		
DPYD Phenotype	Normal [f2 i1]		

Result Footnote

f1: EER Dihydropyrimidine Dehydrogenase
Authorized individuals can access the ARUP Enhanced Report using the following link:

[redacted]

f2: DPYD Phenotype

Activity Score: 0

Interpretation: This patient is homozygous for the c.1679T>G (*13) variant in the DPYD gene. 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: Avoid use of 5-fluorouracil or 5-fluorouracil prodrug-based regimens. The Clinical Pharmacogenetics Implementation Consortium (CPIC) dosing guidelines for fluoropyrimidines can be found at: https://cpicpgx.org/ and https://www.pharmgkb.org/gene/PA145.

This result has been reviewed and approved by [redacted]

Test Information

i1: DPYD Phenotype

BACKGROUND INFORMATION: Dihydropyrimidine Dehydrogenase (DPYD), 3 Variants
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:
Non-functional alleles and toxicity risk:
*13 (rs55886062, c.1679T>G) - Increased risk
*2A (rs3918290, c.1905+1G>A) - Increased risk
Decreased function allele and toxicity risk:

Test Information

i1: DPYD Phenotype

c.2846A>T (rs67376798) - Increased risk

A result of *1 indicates no variants detected and is predictive of functional alleles and normal enzymatic activity.

CLINICAL SENSITIVITY: Estimated at 31 percent for the DPYD variants analyzed.
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY and SPECIFICITY: 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 non-genetic 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 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.