

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 1/1/1993
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Dihydropyrimidine Dehydrogenase (DPYD), 3 Variants

ARUP test code 2012166

EER Dihydropyrimidine Dehydrogenase

See Note

Authorized individuals can access the ARUP
Enhanced Report using the following link:

[REDACTED]

DPYD Specimen

whole Blood

DPYD Genotype

*1/*1

DPYD Phenotype

Normal

Activity Score: 2

Interpretation: No dihydropyrimidine dehydrogenase (DPYD) gene variants were detected in this individual suggesting the presence of *1 functional alleles. This result predicts the normal metabolizer phenotype for dihydropyrimidine dehydrogenase (DPD) and normal risk for 5-FU toxicity.

Recommendations: 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: 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:

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.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
EER Dihydropyrimidine Dehydrogenase	24-002-118538	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
DPYD Specimen	24-002-118538	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
DPYD Genotype	24-002-118538	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
DPYD Phenotype	24-002-118538	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: 24-002-118538
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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