ARUP LABORATORIES
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Tracy I. George, MD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	4/17/1942
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

DPYD Specimen	Whole Blood
DPYD Genotype	Negative
DPYD Phenotype	Normal

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

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

Recommendation: This result does not replace the need for therapeutic drug or clinical monitoring. 5-FU drug metabolism, efficacy and risk for toxicity may be affected by genetic and non-genetic factors not detected by this assay.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Dihydropyrimidine Dehydrogenase (DPYD), 3 Variants

Background information for 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) - Greatly increased risk

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

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

Allele Frequency by Population:

*13: Caucasian - 0.1 percent; Asian - absent; African American - absent

*2A: Caucasian - 0.47-2.2 percent; Asian - absent; African American - absent

c.2846A>T: Caucasians - 1.1 percent; Asian - absent; African American - absent

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.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
DPYD Specimen	18-200-135941	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
DPYD Genotype	18-200-135941	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
DPYD Phenotype	18-200-135941	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