

LABORATORY REPORT

QC ACCOUNT (WARDE) 300 W. TEXTILE ANN ARBOR MI 48108

EXAMPLE, REPORT W

WX0000003827 M 07/08/1978 46 Y

Referral Testing

Collected: 07/16/2024 14:35 Received: 07/16/2024 14:35

Test Name Result Flag Ref-Ranges Units Site

Dihydropyrimidine Dehydrogenase (DPYD), 3 Variants

DPYD Specimen Whole Blood ARRL
DPYD Genotype *1/*1 ARRL
DPYD Phenotype Normal ARRL
DPYD Interpretation See Note ARRL

This result has been reviewed and approved by Philip Bernard, $\mathrm{M.D.}$

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 $$\operatorname{functional}$$ 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.



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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.

EER Dihydropyrimidine Dehydrogenase

See Note

ARRL

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

https://c11-erpt.aruplab.com/?t=062406aA32Z6Hc232

Performed By: ARUP Laboratories

500 Chipeta Way

Salt Lake City, UT 84108

Laboratory Director: Jonathan R. Genzen, MD, PhD

CLIA Number: 46D0523979

Reported Date: 07/16/2024 14:35 DPYDV

Performing Site:

ARRL: ARUP REFERENCE LAB 500 Chipeta Way Salt Lake City UT 841081221

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

G316000288 WX0000003827 Printed D&T: 07/16/24 14:36 Ordered By: CLIENT CLIENT WX00000000000002516

Kajal V. Sitwala, MD, PhD - Medical Director Form: MM RL1 PAGE 2 OF 2