

LABORATORY REPORT

Example Client, XYZ123 1234 Warde Road Ann Arbor MI 48108

EXAMPLE, REPORT W

WX0000003827 M 07/08/1978 45 Y

Referral Testing

Collected: 10/20/2023 12:06 Received: 10/20/2023 12:06

Test Name Result Flag Ref-Ranges Units Site

Dihydropyrimidine Dehydrogenase (DPYD), 3 Variants

DPYD Specimen Whole Blood ARRL
DPYD Genotype Cmpnd Hetero AB ARRL
DPYD Phenotype Normal ARRL

This result has been reviewed and approved by Yuan Ji, Ph.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 $% \left(1\right) =\left(1\right) +\left(1\right) =\left(1\right) +\left(1\right) +\left(1\right) =\left(1\right) +\left(1\right)$

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

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

F420000015 WX0000003827 Printed D&T: 11/20/23 15:42 Ordered By: KAJAL SITWALA, MD, PhD WX000000000002365

Kajal V. Sitwala, MD, PhD - Medical Director Form: MM RL1 PAGE 1 OF 2



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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

EERUnavailable

ARRL

Performed By: ARUP Laboratories

500 Chipeta Way

Salt Lake City, UT 84108

Laboratory Director: Jonathan R. Genzen, MD, PhD

CLIA Number: 46D0523979

Reported Date: 10/20/2023 12:07 DPYD3

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

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