

DIHYDROPYRIMIDINE DEHYDROGENASE (DPD) GENE
MUTATION ANALYSIS
Test ID: FDPDG

EXPLANATION: The following new orderable, referred to Quest Diagnostics Nichols Institute, will be available April 17, 2014. This test will replace Test ID FDPD.

REFERRAL LAB CODE: 15538X

METHODOLOGY: Polymerase Chain Reaction (PCR), Single Nucleotide Primer Extension

REFERENCE VALUES:

Dihydropyrimidine dehydrogenase (DPD) is the rate-limiting enzyme in the pathway for the degradation of the pyrimidine bases, uracil and thymine. DPD also catalyzes the detoxification of pyrimidine-based chemotherapeutic agents (e.g. 5-fluorouracil (5-FU) and capecitabine). Decreased DPD activity is associated with severe myelosuppression or even lethal toxicity, in patients treated with standard doses of 5-FU. DPD deficiency is associated with congenital thymine-uraciluria, an autosomal recessive condition characterized by convulsive disorders, microcephaly, and mental retardation. The IVS14+1G>A mutation in the splice-donor site of intron 14 of the DPD gene (located on chromosome 1) accounts for approximately 50% of DPD deficiency alleles.

The IVS14+1G>A mutation is detected by polymerase chain reaction (PCR) amplification of a portion of the DPD gene, followed by a single nucleotide primer extension reaction using fluorescent dideoxynucleotides, and detection of the fluorescent reaction products using an automated, capillary DNA sequencer. Since genetic variation and other problems can affect the accuracy of the direct mutation testing, these results should always be interpreted in light of clinical and familial data.

This test is performed pursuant to a license agreement with Orchid Biosciences Inc.

SPECIMEN REQUIREMENTS:

Draw blood in a purple-top (EDTA), ACD A or B (yellow-top), green-top (sodium heparin) or green-top (lithium heparin) tube(s). Send 5 mL of whole blood ambient in a plastic vial.

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Whole Blood EDTA	Ambient (preferred)	8 days
	Refrigerated	8 days

FEE: \$ 287.50

CPT CODE: 81400

DAY(S) SET UP: Monday, Thursday

ANALYTIC TIME: 6 - 10 days

QUESTIONS: Contact Mary Erath, MML Referrals Supervisor
Telephone: 800-533-1710