MAYO CLINIC
Mayo Medical Laboratories

Test Definition: FDPDM

Dihydropyrimidine Dehydrogenase (DPD) Gene Mutation Analysis
NYS Clients Only

Reporting Title: DPD Gene Mutation
Performing Location: Quest SJC, Interface

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

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
Z3757	DPD Gene Mutation	Alphanumeric	mL	45284-7

CPT Code Information:

81400

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 (eg. 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 the 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.

Test Performed by: Quest Diagnostics Nichols Institute
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