Test Definition: FDPD

Dihydropyrimidine Dehydrogenase (DPD) Gene Mutation Analysis

Reporting Title: DPD Gene Mutation
Performing Location: Quest Diagnostics/Nichols Institute

Specimen Requirements:

Collect 5 mL lavender-top (EDTA) whole blood. Ship ambient immediately.

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

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
Z2293	Result	Alphanumeric		45284-7
Z2294	Interpretation	Alphanumeric		In Process
Z2295	Reviewed By:	Numeric		In Process

CPT Code: 1 × 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 (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

Test Performed By: Quest Diagnostics Nichols Institute
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