MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,FDPD	SA00064657	49	M	SA00064657
Ordering Phys CLIENT,CLIENT				**DOB** 06/27/1964
Client Order # SA00064657	**Account Information**			**Report Notes**
Collected 11/21/2013 06:00	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 11/22/2013 11:22				

Test	Flag	Results	Unit	Reference Value	Perform Site*
DPD Gene Mutation					
RECEIVED: 11/21/2013 15:22 **REPORTED:** 11/22/2013 08:25					
Result		No Mutation Detected			Y096
Interpretation		see message			Y096

DNA testing indicates that this individual is negative for the IVS14+1G>A mutation in the DPD gene. This mutation accounts for approximately 50% of DPD deficiency alleles. This negative result does not rule out the presence of rare DPD mutations that are not detected by this assay. Therefore, the possibility of DPD deficiency and a severe adverse reaction to treatment with pyrimidine-based chemotherapeutic agents (e.g. 5-fluorouracil and capecitabine) cannot be ruled out.

Reviewed By:		see message			Y096

Franklin Quan, Ph.D., ABMG, CGMB.

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 detox-ification of pyrimidine-bases 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 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.

Performing Site Legend on Last Page of Report

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

This test was developed and its performance characteristics have been determined by Quest Diagnostics Nichols Institute, San Juan Capistrano. Performance characteristics refer to the analytical performance of the test.
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-bases 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.

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* Performing Site:

Y096	Quest Diagnostics Nichols Institute 33608 Ortega Highway San Juan Capistrano, CA 92690-6130	Lab Director: