

Laboratory Service Report

1-800-533-1710

Patient Name SAMPLEREPORT,FMMD HO	Patient ID SA00047637	Age 38	Gender F	Order # SA00047637
Ordering Phys				DOB 05/16/1974
Client Order # SA00047637	Account Information			Report Notes
Collected 07/13/2012 09:27	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE			
Printed 08/03/2012 15:08	ROCHESTER,MN 55901			

Test	Flag	Results	Unit	Reference Value	Perform Site*
MTHFR Mutation Detection MTHFR PCR Specimen		Whole Blood	REPORTED 0	7/16/2012 08:56	Y00 6
MTHFR Mutation: C677T		Heterozygous			Y00
MTHFR Mutation: A1298C		Heterozygous			6 Y00 6
MTHFR Interpretation		See Note			Y00 6

Indication for testing: Determine genetic contribution to early-onset arteriosclerotic vascular disease or venous thrombosis and/or assess tolerance to antifolate medications.

Compound Heterozygous MTHFR C677T/Al298C: One copy of each of the MTHFR gene mutations tested, C677T and Al298C, was detected. The mutations are rarely found on the same chromosome; thus, this genotype is predicted to be associated with a poor metabolizer phenotype characterized by increased plasma homocysteine levels. This individual is at an increased risk for arteriosclerotic coronary disease and venous thrombosis as well as for toxicity from medications affecting folate metabolism.

BACKGROUND INFORMATION: Methylenetetrahydrofolate Reductase (MTHFR) 2 Mutations

CHARACTERISTICS: Mutations in the MTHFR gene (C677T and A1298C) correlate with reduced enzyme activity; however, only homozygotes for C677T or compound heterozygotes for C677T/A1298C have significantly elevated plasma homocysteine levels and increased risk for premature cardiovascular disease. These individuals may also show toxicity from medications (ie. methotrexate) that affect folate metabolism.

INCIDENCE: US allele frequency of C677T = 0.39 and A1298C = 0.17; homozygosity for C677T is 1-15 percent.

INHERITANCE: Autosomal recessive.

CAUSE: Homozygosity for MTHFR gene mutation C677T or

compound heterozygosity for C677T/Al298C. MUTATIONS TESTED: c.677C>T and c.1298A>C

CLINICAL SENSITIVITY: Undefined. Sensitivity is dependent

upon multiple contributing factors.

METHODS: PCR followed by fluorescent monitoring using

hybridization probes.

Performing Site Legend on Last Page of Report

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Reference Perform Test Flag Results Unit Value Site*

ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent. LIMITATIONS: Only the two MTHFR gene mutations (C677T and A1298C) will be targeted; rare diagnostic errors may occur due to primer site mutations.

This test is performed pursuant to an agreement with Roche Molecular Systems, Inc .

This test is performed pursuant to an agreement with $\operatorname{Siemens}$ $\operatorname{Medical}$ $\operatorname{Solutions}.$

The performance characteristics of this test were validated by ARUP Laboratories. The U.S. Food and Drug Administration (FDA) has not approved or cleared this test. However, FDA approval or clearance is currently not required for clinical use of this test. The results are not intended to be used as the sole means for clinical diagnosis or patient management decisions. ARUP is authorized under Clinical Laboratory Improvement Amendments (CLIA) and by all states to perform high-complexity testing.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online at www.aruplab.com.

* Performing Site:

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Vone	ARUP Laboratories	Lab Director
Y006	500 Chipeta Way Salt Lake City, UT 84108	Lab Director:

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