MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,FMMD HO	SA00047637	38	F	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 ROCHESTER,MN 55901			
Printed 07/19/2012 16:40				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MTHFR Mutation Detection			REPORTED 07/16/2012 08:56		
MTHFR PCR Specimen		Whole Blood			Y154
MTHFR Mutation: C677T		Heterozygous			Y154
MTHFR Mutation: A1298C		Heterozygous			Y154
MTHFR Interpretation		See Note			Y154

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/A1298C: One copy of each of the MTHFR gene mutations tested, C677T and A1298C, 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/A1298C.
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

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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hybridization probes.
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 Siemens Medical 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:

Y154	ARUP Labs 500 Chipeta Way Salt Lake City, UT 84108	Lab Director: