MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,FMMD N	SA00047636	56	M	SA00047636
Ordering Phys				**DOB** 05/16/1956
Client Order # SA00047636	**Account Information**			**Report Notes**
Collected 07/13/2012 09:20	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/03/2012 15:09				

Test	Flag	Results	Unit	Reference Value	Perform Site*
MTHFR Mutation Detection		REPORTED 07/16/2012 08:56			
MTHFR PCR Specimen		Whole Blood			Y006
MTHFR Mutation: C677T		Negative			Y006
MTHFR Mutation: A1298C		Negative			Y006
MTHFR Interpretation		See Note			Y006

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

Negative: Neither of the common MTHFR gene mutations tested, C677T and A1298C, were detected. Other causes of elevated homocysteine levels, coronary heart disease, or venous thrombosis cannot be excluded. This genotype is associated with normal 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 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.

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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:

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