

5,10-Methylenetetrahydrofolate Reductase  
C677T and A1298C Mutations, Blood

Patient ID <b>SA00056231</b>	Patient Name <b>SAMPLEREPOR, MTHP</b>	Birth Date <b>1966-06-10</b>	Gender <b>F</b>	Age <b>46</b>
Order Number <b>SA00056231</b>	Client Order Number <b>SA00056231</b>	Ordering Physician <b>Client, Client</b>	Report Notes	
Account Information <b>C7028846 DLMP Rochester</b>		Collected <b>26 Mar 2013 13:00</b>		

**MTHFR 2 Mutations Analysis, B**

**MTHFR C677T Mutation Analysis, B**

**MTHFR Interpretation**

MCR

This individual DOES have the Methylenetetrahydrofolate reductase (MTHFR) C677T gene mutation on ONE allele (heterozygous mutant). MTHFR C677T carriers are not at increased risk for thrombosis in the absence of hyperhomocysteinemia. In the absence of alternative causes, heterozygous carriers of MTHFR C677T are not at increased risk for hyperhomocysteinemia. Hyperhomocysteinemia is a relatively weak risk factor for both venous thromboembolism and arterial thrombosis. The MTHFR C677T gene mutation test does not detect other causes of hyperhomocysteinemia due to acquired disorders (renal failure, zinc deficiency, leukemia, psoriasis, or antifolate drug therapy). If clinically indicated, suggest Coagulation Consultation 83093 (Thrombophilia Profile) to complete the evaluation for an inherited or acquired thrombosing disorder (i.e., thrombophilia). Consider genetic consultation and

counseling of potentially affected family members regarding laboratory testing.

**ADDITIONAL INFORMATION**

This test is a direct mutation analysis using PCR amplification, signal generation and release by cleavage of sequence specific alleles (Invader Plus Chemistry, Hologic, Madison, WI).

**Methylenetetrahydrofol Reduc Mut, B**

MCR

 Heterozygous  


**Reference Value**  
Negative

**MTHFR Reviewed By**

MCR

Ann Stregre

**MTHFR A1298C Mutation Analysis, B**

**MTHAC Interpretation**

MCR

This individual DOES have the Methylenetetrahydrofolate reductase (MTHAC) A1298C gene mutation on ONE allele (heterozygous mutant). MTHAC A1298C carriers are not at increased risk for thrombosis in the absence of hyperhomocysteinemia. In the absence of alternative causes, heterozygous carriers of MTHAC A1298C are not at increased risk for hyperhomocysteinemia. Hyperhomocysteinemia is a relatively weak risk factor for both venous thromboembolism and arterial thrombosis. The MTHAC A1298C gene mutation test does not detect other causes of hyperhomocysteinemia due to acquired disorders (renal failure, zinc deficiency, leukemia, psoriasis, or antifolate drug therapy). If clinically indicated, suggest Coagulation Consultation 83093 (Thrombophilia Profile) to complete the evaluation for an inherited or acquired thrombosing disorder (i.e., thrombophilia). Consider genetic consultation and counseling of potentially affected family members regarding laboratory testing.

**ADDITIONAL INFORMATION**

This test is a direct mutation analysis using PCR amplification, signal generation and release by cleavage of sequence specific alleles (Invader Plus Chemistry, Hologic, Madison, WI).

**MTHFR A1298C Mutation Analysis, B**

MCR

 Heterozygous  


**Reference Value**  
Negative

**MTHAC Reviewed By**

MCR

Ann Stregre

This test is a direct mutation analysis using PCR amplification, signal generation and release by cleavage of sequence specific alleles (Invader Plus Chemistry, Hologic, Madison, WI).

**Received:** 27 Mar 2013 08:32

**Reported:** 27 Mar 2013 08:36

**Performing Site Legend**

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905