



5,10-Methylenetetrahydrofolate Reductase
A1298C, Mutation, Blood

Patient ID SA00056234	Patient Name SAMPLEREP, MTHAC	Birth Date 1966-06-10	Gender F	Age 46
Order Number SA00056234	Client Order Number SA00056234	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 26 Mar 2013 13:00		

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 thrombotic 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

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Heterozygous

Reference Value
Negative

MTHAC Reviewed By

MCR

Ann Strege

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