Quest Diagnostics

Report Status: Final

Patient Information	Specimen Information	Client Information

COMMENTS:

Test Name	In Range	Out Of Range	Reference Range	Lab
METHYLENETETRAHYDROFOLATE REDUCTASE (MTHFR), DNA	POSITIVE			
RESULT: POSITIVE FOR ONE COPY OF THE A1298C VARIANT				
INTERPRETATION	See Below			

INTERPRETATION:This individual is heterozygous for the A1298C variant and negative (normal) for the C677T variant in the MTHFR gene. This result is not associated with a significantly increased risk for coronary artery disease, venous thromboembolism, or adverse pregnancy outcome.

Laboratory testing supervised and results monitored by Franklin Quan, Ph.D., FACMG, HCLD, CGMB.

Reduced methylenetetrahydrofolate reductase (MTHFR) enzyme activity is a genetic risk factor for hyperhomocysteinemia, especially when present with low serum folate levels. Two common variants in the MTHFR gene result in reduced enzyme activity. The "thermolabile" variant C677T [NM 005957.3: c.665C>T (p.A222V)] and A1298C [c. 1286A>C (p.E429A)] occur frequently in the general population.

Mild to moderate hyperhomocysteinemia has been identified as a risk factor for coronary artery disease and venous thromboembolism. Hyperhomocysteinemia is multifactorial, involving a combination of genetic, physiologic and environmental factors. Recent studies do not support the previously described association of increased risk for coronary artery disease and venous thromboembolism with mild hyperhomocysteinemia caused by reduced MTHFR activity. Therefore, the utility of MTHFR variant testing is uncertain and is not recommended by The American College of Medical Genetics and Genomics (ACMG) or the American Congress of Obstetricians and Gynecologists (ACOG) in the evaluation of venous thromboembolism or adverse pregnancy outcome.

Modest positive association has also been found between the "thermolabile" variant of the MTHFR gene and many other medical complications, such as recurrent pregnancy loss, risk of offspring with neural tube defects, neuropsychiatric disease, and chemotherapy toxicity. Increased risk of coronary artery disease, venous thromboembolism and increased plasma homocysteine can be caused by a variety of genetic and non-genetic factors not screened for by this assay. If indicated by personal or family history of thromboembolism, consider additional testing such as plasma homocysteine levels, factor V Leiden and prothrombin gene mutations.

The C677T and A1298C variants are detected by amplification of the selected regions of the MTHFR gene by polymerase chain reaction (PCR) and fluorescent probes hybridization to the targeted regions, followed by melting curve analysis

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with a real time PCR system. Although rare, false positive or false negative results may occur. All results should be interpreted in context of clinical findings, relevant history, and other laboratory data. Health care providers, please contact your local Quest Diagnostic's genetic counselor or call 866-GENEINFO (866-436-3463) for assistance with interpretation of these results.

For additional information, please refer to http://education.QuestDiagnostics.com/faq/FAQ66 (This link is being provided for informational/educational purposes only.)

This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics Nichols Institute San Juan Capistrano. It has not been cleared or approved by the U.S. Food and Drug Administration. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

PERFORMING SITE:

SPECIMEN: