ARUP LABORATORIES | aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Tracy I. George, MD, Chief Medical Officer

Patient Report

Patient Age/Sex: 31 years Female

Specimen Collected: 14-Jun-22 07:12

Methylenetetrahydrofolate Reductase **|Received: 14-Jun-22 07:32** **Report/Verified: 20-Jun-22 11:02**

Procedure	Result	Units	Reference Interval
MTHFR PCR Specimen	Whole Blood		
MTHFR Variant:c.665C>T	Heterozygous		
MTHFR Variant:c.1286A>C	Negative		
MTHFR Interpretation	See Note f1 i1		

Result Footnote

f1: MTHFR Interpretation

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

Indication for testing: Determine genetic contribution to hyperhomocysteinemia.

Heterozygous MTHFR c.665C>T: One copy of the MTHFR c.665C>T variant (previously designated C677T) was detected; the c.1286A>C variant (previously designated A1298C) was not identified. The common c.665C>T variant is present in 12 percent of African Americans, 35 percent of Caucasians, and 50 percent of Hispanic individuals. Although MTHFR enzyme activity may be mildly reduced, this genotype is not predicted to have clinical significance.

This result has been reviewed and approved by Yuan Ji, Ph.D.

Test Information

i1: MTHFR Interpretation

Background Information: Methylenetetrahydrofolate Reductase (MTHFR) 2 Variants
Characteristics: Variants in the MTHFR gene may reduce enzyme activity contributing to hyperhomocysteinemia. Although hyperhomocysteinemia was previously reported to be a risk factor for many conditions, especially venous thrombosis and cardiovascular disease, recent meta-analysis casts doubt on whether lifelong moderate homocysteine elevation has an effect on cardiovascular disease. The American College of Medical Genetics Practice Guidelines indicate that individuals with elevated homocysteine and two copies of the c.665C>T variant have an odds ratio of 1.27 for venous thromboembolism. Thus, they recommend MTHFR genotyping not be ordered as part of a routine evaluation for recurrent pregnancy loss or thromobophilia due to questionable clinical significance.
Incidence: The allele frequency of the c.665C>T variant is 0.35 in European Caucasians, 0.5 in Hispanics, and 0.12 in African Americans.
Inheritance: Autosomal recessive; two copies of the c.665C>T variant may be a contributing factor to hyperhomocysteinemia.
Variants Tested: c.665C>T(p.Ala222Val) and c.1286A>C(p.Glu429Ala). (legacy names C677T and A1298C, respectively).

Test Information

i1: MTHFR Interpretation

Clinical Sensitivity: Undefined; hyperhomocysteinemia is caused by genetic, physiologic and environmental factors. MTHFR variants are only one contributing factor.
Methodology: Polymerase chain reaction (PCR) and fluorescence monitoring.
Analytical Sensitivity & Specificity: 99 percent.
Limitations: Only two MTHFR gene variants (c.665C>T and c.1286A>C) are tested. Diagnostic errors can occur due to rare sequence variations.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.