

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB: 10/22/1944
Gender: Male
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 01/01/2017 12:34

Methylenetetrahydrofolate Reductase (MTHFR) 2 Variants

ARUP test code 0055655

MTHFR PCR Specimen whole Blood

MTHFR Mutation: c.665C>T **Heterozygous ***

MTHFR Mutation: c.1286A>C Negative

MTHFR Interpretation See Note

Indication for testing: Determine genetic contribution to hyperhomocysteinemia.

Heterozygous MTHFR c.665C>T: One copy of the MTHFR variant c.665C>T (previously designated C677T) was detected; the c.1286A>C (previously designated A1298C) variant was not identified. The common 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 [REDACTED]

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

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 thrombophilia 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).
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.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
MTHFR PCR Specimen	20-057-140509	2/26/2020 9:20:00 AM	2/28/2020 7:57:00 AM	3/1/2020 8:10:00 PM
MTHFR Mutation: c.665C>T	20-057-140509	2/26/2020 9:20:00 AM	2/28/2020 7:57:00 AM	3/1/2020 8:10:00 PM
MTHFR Mutation: c.1286A>C	20-057-140509	2/26/2020 9:20:00 AM	2/28/2020 7:57:00 AM	3/1/2020 8:10:00 PM
MTHFR Interpretation	20-057-140509	2/26/2020 9:20:00 AM	2/28/2020 7:57:00 AM	3/1/2020 8:10:00 PM

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Tracy I. George, MD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 20-057-140509
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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