

Methylenetetrahydrofolate Reductase (MTHFR), DNA Mutation Analysis

CPT Code: 81291

Order Code: 1088

ABN Requirement: No

Synonyms: MTHFR; Methylenetetrahydrofolate reductase

Specimen: Whole Blood

Volume: 5.0 mL

Minimum Volume: 3.0 mL

Container: EDTA (lavender-top) (preferred) EDTA (royal blue-top) Sodium heparin (green-top) ACD solution A (yellow-top) ACD solution B (yellow-top) Lithium heparin (green-top)

Collection:

1. Collect and label sample according to standard protocols.
2. Gently invert tube 10 times immediately after draw. DO NOT SHAKE.
3. Do not centrifuge.

Please Note: This germline genetic test requires physician attestation that patient consent has been received if ordering medical facility is located in AK, DE, FL, GA, IA, MA, MN, NV, NJ, NY, OR, SD, or VT or test is performed in MA.

Special Instructions: Extracted DNA: Please call 1-866-GENE-INFO or 1-866-436-4636 for additional information. Frozen shipping is acceptable.

Transport: Store EDTA whole blood at 2°C to 8°C after collection and ship the same day per packaging instructions included with the provided shipping box.

Stability:

Ambient (15-25°C): 8 days

Refrigerated (2-8°C): 8 days

Frozen (-20°C): 30 days

Methodology: Polymerase Chain Reaction & Detection

Turn Around Time: 4 to 7 days

Reference Range: See Laboratory Report

Clinical Significance: The Methylene tetrahydrofolate Reductase (MTHFR) enzyme plays a major role in homocysteine metabolism and contains several known polymorphisms, of which the most common is C677T. This mutation is reported to reduce MTHFR activity, resulting in hyperhomocysteinemia. This condition is a risk factor for cardiovascular disease, increased risk for arterial and venous thrombosis, and an increased risk for obstetrical complications, e.g., preeclampsia, abruptio placentae, fetal growth retardation, and stillbirth.

Limitations: The purpose of this test is to determine if you have two, one, or no copies of either of two mutations in the MTHFR gene, C677T and A1298C.

The CPT codes provided are based on AMA guidelines and are for informational purposes only. CPT coding is the sole responsibility of the billing party. Please direct any questions regarding coding to the payer being billed.