## Lab Dept: Anatomic Pathology

## Test Name: MTHFR GENOTYPE

### *General Information*

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| **Lab Order Codes:** | MTHFR |
| **Synonyms:** | Hyperhomocysteinemia; Methylenetetrahydrofolate reductase genotype; MTHFR Mutation Analysis |
| **CPT Codes:** | 81291 – MTHFR gene analysis, common variants |
| **Test Includes:** | MTHFR mutation is reported as present or absent. Heterozygosity or homozygosity is also determined. |

### *Logistics*

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| **Test Indications:** | Determination of the presence of the thermolabile C677T mutation of the methylenetetrahydrofolate reductase (MTHFR) gene. This mutation results in MTHFR deficiency, which can cause an accumulation of homocysteine in plasma (hyperhomocysteinemia). Hyperhomocysteinemia has been identified as an independent risk factor in the development of premature vascular disease. |
| **Lab Testing Sections:** | Anatomic Pathology - Sendouts |
| **Referred to:** | Fairview University Medical Center (Test code: MTHFRD) |
| **Phone Numbers:** | MIN: 612-813-6280<br>STP: 651-220-6550 |
| **Test Availability:** | Daily, 24 hours |
| **Turnaround Time:** | Results are reported within 7 days |
| **Special Instructions:** | Obtain special tube from the laboratory. See Container. Include patient history relating to hyperhomocysteinemia. A signed informed consent in the patient's medical record is required; the consent should not be sent to the laboratory. |

### *Specimen*

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| **Specimen Type:** | Whole blood |

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| **Container:** | Yellow top (ACD A) tube (Available from the laboratory)<br>Alternate tubes: Yellow top (ACD B) or Lavender (EDTA) |
| **Draw Volume:** | 10 mL (Minimum: 5 mL) blood |
| **Processed Volume:** | Same as Draw Volume |
| **Collection:** | Routine venipuncture. Invert specimen gently to mix. |
| **Special Processing:** | Lab Staff: **Do not** centrifuge. Specimen should remain in original collection container. Ship and store specimen at room temperature. Specimen must be less than 5 days old. |
| **Patient Preparation:** | None |
| **Sample Rejection:** | Improper specimen type; improper tube used; stored at incorrect temperature; clotted specimen; frozen specimen; specimen older than 5 days old; mislabeled or unlabeled specimens. |

## *Interpretive*

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| **Reference Range:** | Results are reported as "present" or "absent" for the mutation. Heterozygosity or homozygosity is also determined. |
| **Critical Values:** | N/A |
| **Limitations:** | Patients are only tested ONCE; if duplicate sample is collected, testing is canceled, credited and original results and collection date reported. |
| **Methodology:** | Amplification of DNA with restriction digest, detection by gel electrophoresis |
| **References:** | MHealth Fairview Reference Lab (December 2023) |
| **Updates:** | 2/7/2013: CPT update<br>12/7/2023: Correction of typos, updated website address |