Lab Dept: Anatomic Pathology

Test Name: MTHFR GENOTYPE

General Information

Lab Order Codes: MTHFR

Synonyms: Hyperhomocysteinemia; Methylene tetrahydrofolate 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 homzygosity is also determined.

Logistics

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

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

Specimen Type: Whole blood
**Container:**
Yellow top (ACD A) tube (Available from the laboratory)
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

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**Interpretive**

**Reference Range:**
Results are reported as “present” or “absent” for the mutation. Heterozygosity or homozygosity is also determined.

**Critical Values:**
N/A

**Limitations:**
N/A

**Methodology:**
Amplification of DNA with restriction digest

**References:**
[Fairview University Website](http://fairview.com) (March 2018)

**Updates:**
2/7/2013: CPT update