
Lab Dept: **Anatomic Pathology**

Test Name: **MTHFR GENOTYPE**

General Information

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

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

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| 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 |

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: | N/A |
| Methodology: | Amplification of DNA with restriction digest |
| References: | Fairview University Website (March 2018) |
| Updates: | 2/7/2013: CPT update |