Lab Dept: Anatomic Pathology

Test Name: MTHFR GENOTYPE

General Information

Lab Order Codes:	MTHFR
Synonyms:	Hyperhomocysteinemia; Methylenetetrahyfrofolate 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 he 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 (March 2018)
Updates:	2/7/2013: CPT update