

GENETIC DISEASES EVALUATION CENTER

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Test Name & Code:

- **Test Name:** MTHFR A1298C Mutation Analysis
- **Test Code:** 3108

Related Diseases & Genes:

- Homocystinuria due to MTHFR deficiency (OMIM #: 236250) || Gene: *MTHFR* (OMIM #: 607093)

Synonyms:

- 5,10-METHYLENETETRAHYDROFOLATE REDUCTASE
- METHYLENETETRAHYDROFOLATE A1298C

Test Details:

- **Methodology:** RT-PCR
- **Performed:** Everyday
- **Reported:** Next day (18:00)

Specimen Details:

- **Specimen Collection:** Peripheral blood
- **Container:** Tube with EDTA
- **Specimen Volume/Amount:** > 3 ml
- **Storage/Transport Condition:** Refrigerated
- **Unacceptable Specimens/Conditions:** Serum; Frozen specimens; Hemolyzed specimens; Icterus; Lipemia

Testing Area(s):

- Predisposition/Risk Factors

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