



TEST: DETECTION OF THE METHYLENE-TETRAHYDROFOLATE REDUCTASE (MTHFR) A1298C GENE POLYMORPHISM BY PCR

PRINCIPLE:

5,10-methylenetetrahydrofolate reductase (MTHFR) is an enzyme that acts as a substrate in the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, a critical step in the homocysteine pathway. Mutations in the *MHTFR* gene cause elevated levels of homocysteine, which are, subsequently, a risk factor for venous and arterial thrombosis. The most common mutation in the *MTHFR* gene is the alanine-to-valine substitution at nucleotide 677 (C677T), which leads to a defective enzyme. The second most common mutation is the glutamate-to-alanine substitution at nucleotide 1298 (A1298C), which also leads to reduced enzymatic activity. MTHRF gene polymorphisms have been associated with vascular diseases, pregnancy complications (such as recurrent spontaneous abortions) and malformations in fetal development. Women who are heterozygous in both *MTHFR* mutations are in increased risk of developing pregnancy complications.

SPECIMEN COLLECTION AND PREPARATION:

10mL whole blood collected in lavender top EDTA tubes (two 5ml tubes). Specimen should be delivered to the laboratory within 72 hours at room temperature. Peripheral blood specimens that are clotted, have not been collected in EDTA, or frozen are not acceptable.

METHOD:

Polymerase chain reaction (PCR) and reverse hybridization.

REFERENCES:

1. Skibola C, Smith M, Kane E et al. *PNAS* 1999;96:12810-12815
2. Friedman G, Goldsmith N et al. *J Nutr* 1999;129:1656-1661

Normal Range: Reported as Normal (A/A), Heterozygous Mutated (A/C) or Homozygous Mutated (C/C)

Turnaround time: 10 business days