



# MTHFR DNA Test

Hyperhomocysteinemia is a widely recognized risk factor for coronary artery disease, venous thrombosis, and stroke. It is also involved in the pathogenesis of neural tube defects, stillbirths, and recurrent pregnancy loss. The leading cause of hyperhomocysteinemia is folate deficiency. Other determinants include insufficient B12 intake, impaired renal function, and genetic variations including those in the MTHFR gene. Folate supplementation can correct for most causes of hyperhomocysteinemia.

Methylenetetrahydrofolate reductase (MTHFR) is a key enzyme in the metabolism of homocysteine. Mutations in the MTHFR gene have been reported as causes of hyperhomocysteinemia. The most common MTHFR mutation, C677T, is present in the homozygous state in 5-10% of the general Caucasian population. In homozygous individuals, this results in a thermolabile variant of the enzyme with decreased activity. Individuals homozygous for the C677T mutation are predisposed to developing hyperhomocysteinemia, particularly when deficient in folate. The frequency of C677T homozygosity is increased in individuals with coronary artery disease (to 17%), arterial disease (to 19%), and venous thromboembolism (to 11%).

The presence of a second mutation in the MTHFR gene, A1298C, in conjunction with C677T, has been associated with decreased MTHFR activity and hyperhomocysteinemia. The frequency of the A1298C mutation is reported to be as high as 30% in the general Caucasian population. Heterozygosity or homozygosity for A1298C alone does not result in hyperhomocysteinemia. MTHFR mutations, when present with other genetic thrombophilic factors (e.g., factor V Leiden), dramatically increase risk for venous thrombosis.

Testing for mutations in MTHFR is useful in identifying a genetic etiology for persistent hyperhomocysteinemia. Also, in individuals known to have other genetic thrombophilic factors (e.g., factor V Leiden), detection of MTHFR mutations signifies a dramatically increased risk for venous thrombosis.

## Indications for MTHFR DNA Testing:

- Hyperhomocysteinemia
- History of venous thromboembolism, coronary artery disease, and/or stroke
- History of pregnancy complications including neural tube defects, stillbirths, and/or recurrent pregnancy loss
- Individuals with other genetic hypercoagulabilities (e.g., factor V Leiden)
- Relatives of individuals with hyperhomocysteinemia and MTHFR gene mutations

## Our MTHFR DNA Test Service Provides:

- PCR analysis for the C677T and the A1298C mutations in the MTHFR gene
- Detailed reports with genetic interpretation, recommendations, and education
- Free genetic counseling for physicians, patients, and families
- Free shipping via FedEx

## Specimen Requirements:

- 5 ml blood in an EDTA (lavender top) tube, room temperature

## Turnaround Time:

- 1 business day

Please call Kimball Genetics for more information.

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