KASHI CLINICAL LABORATORIES

MTHFR Genotyping
Healthcare Professional Information

Methylation: Understand your Patient's Risk & Health Implications

Methylation is a ubiquitous process throughout the body. The 5,10-Methylenetetrahydrofolate reductase (MTHFR) enzyme is a central component of DNA methylation, responsible for converting folate into its active form.[1,2] This process occurs constantly as new folate is introduced, and utilized folate is recycled back into the active form. Active folate plays a role in many biological processes and interacts with many other enzymes in the methylation pathway. Inadequate amounts of active folate can lead to insufficient methylation which can contribute to many health disorders involving the cardiovascular, reproductive, and neurological systems as well as others.[2,3] Research has shown that many individuals may be at risk of methylation insufficiency due to reduced MTHFR enzyme function.[5,6]

Associated Disorders

- Hyperhomocysteinemia[3]
- Cardiovascular Disease[3]
- Ischemic Stroke[4]
- Dementia/Alzheimer's Disease[10]
- Neural Tube Defects[3]
- Autism[10]
- Down Syndrome[10]
- Depression[10]
- Bipolar Disorder[10]
- Polycystic Ovary Syndrome[11]
- Fracture Risk[4]

MTHFR Genotyping Can Help Make Informed Treatment Decisions

By identifying a patient's genotype and predisposition to decreased enzyme activity, you may be able to improve your patient's health and mitigate risk to disease. The MTHFR Genotyping test assesses your patient's DNA for the presence of two common single nucleotide polymorphisms (SNPs) that have been shown to impact MTHFR enzyme activity.[5-7] These SNPs differ from what we would expect to find in an individual with optimal MTHFR activity. The degree to which the enzyme function is reduced is determined by how many variant SNPs your patient may carry. The two SNPs screened are MTHFR C677T and A1298C. [5-7] C677T, as compared to A1298C, has been shown to have the greatest influence on many health disorders, but the presence of the A1298C mutation in conjunction with the C677T mutation also exerts a significant effect on the enzyme.

MTHFR Variant Combinations and Approximated Enzyme Activity[6]

Genotype	677CC	677CT Heterozygous	677TT Homozygous
1298AA	**100%** Enzyme activity	**66%** Enzyme activity	**25%** Enzyme activity
1298AC Heterozygous	**83%** Enzyme activity	**48%** Enzyme activity	*Not analyzed*
1298CC Homozygous	**61%** Enzyme activity	*Not analyzed*	*Not analyzed*

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