

Methylene tetrahydrofolate reductase

Methylene tetrahydrofolate reductase (MTHFR) is the rate-limiting enzyme in the methyl cycle, and it is encoded by the MTHFR gene.

Methylenetetrahydrofolate reductase catalyzes the conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate, a cosubstrate for homocysteine remethylation to methionine. Natural variation in this gene is common in otherwise healthy people. Although some variants have been reported to influence susceptibility to occlusive vascular disease, neural tube defects, Alzheimer's disease and other forms of dementia, colon cancer, and acute leukemia, findings from small early studies have not been reproduced. Some mutations in this gene are associated with methylenetetrahydrofolate reductase deficiency.

see [MTHFR Polymorphism](#)

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