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MTHFR Polymorphisms and Disease

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Methylenetetrahydrofolate reductase (MTHFR) is an enzyme critical in both folate and homocysteine metabolism. It is a FAD-dependent enzyme that catalyzes the irreversible conversion of 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate which in turn serves as a methyl donor in the remethylation of homocysteine (Hcy) to methionine. The enzyme therefore resides at an important metabolic branch point directing the folate pool towards Hcy remethylation and DNA methylation at the expense of DNA and RNA biosynthesis.

A missense mutation at bp677 predisposes to mild or moderate hyperhomocysteinemia. In addition, cardiovascular disease, neural tube defects, pregnancy complications, neuropsychiatric disease, renal failure and colorectal neoplasias have been reported to be associated with the 677C→T mutation.

Investigations of MTHFR deficiencies recapitulate the transition in medical genetics from isolation of genes involved in single gene disorders to identification of variants involved in complex multifactorial diseases.

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