Eur J Med Genet. 2015 Jan;58(1):1-10. doi: 10.1016/j.ejmg.2014.10.004. Epub 2014 Nov 4.

Methylenetetrahydrofolate reductase (MTHFR) C677T polymorphism: epidemiology, metabolism and the associated diseases.

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Abstract

The Methylenetetrahydrofolate reductase (MTHFR) C677T polymorphism is associated with various diseases (vascular, cancers, neurology, diabetes, psoriasis, etc) with the epidemiology of the polymorphism of the C677T that varies dependent on the geography and ethnicity. The 5,10-Methylenetetrahydrofolate reductase (MTHFR) locus is mapped on chromosome 1 at the end of the short arm (1p36.6). This enzyme is important for the folate metabolism which is an integral process for cell metabolism in the DNA, RNA and protein methylation. The mutation of the MTHFR gene which causes the C677T polymorphism is located at exon 4 which results in the conversion of valine to alanine at codon 222, a common polymorphism that reduces the activity of this enzyme. The homozygous mutated subjects have higher homocysteine levels while the heterozygous mutated subjects have mildly raised homocysteine levels compared with the normal, non-mutated controls. Hyperhomocysteinemia is an emerging risk factor for various cardiovascular diseases and with the increasing significance of this polymorphism in view of the morbidity and mortality impact on the patients, further prevention strategies and nutritional recommendations with the supplementation of vitamin B12 and folic acid which reduces plasma homocysteine level would be necessary as part of future health education. This literature review therefore focuses on the recent evidence-based reports on the associations of the MTHFR C677T polymorphism and the various diseases globally.

KEYWORDS: C677T polymorphism; Diseases; Folic acid; Methylenetetrahydrofolate reductase (MTHFR) gene; Single nucleotide polymorphism; Vitamin B(12)

PMID: 25449138 DOI: 10.1016/j.ejmg.2014.10.004