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Geographical and ethnic distribution of MTHFR gene polymorphisms and their associations with diseases among Chinese population.

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Abstract

Numerous studies have investigated the distribution of methylenetetrahydrofolate reductase (MTHFR) C677T and A1298C polymorphisms and their associations with diseases in China. In this study we conducted a systematic review and meta-analysis of these studies (715 eligible studies in total).Results revealed that the frequencies of the MTHFR C677T and A1298C polymorphisms varied markedly in different areas and ethnicities, and even showed geographical gradients. The MTHFR C677T polymorphism was significantly associated with 42 clinical disorders ($p < 0.05$), mostly relating to the diseases of circulatory system, birth defects and cancers. The association of the A1298C polymorphism with three diseases (coronary heart disease, breast cancer and neural tube defects fathers) was statistically significant ($p < 0.05$). However, according to the Venice criteria, only the associations of the C677T polymorphism with breast and ovarian cancers were assessed as having strong epidemiological credibility. This is the first study to provide a comprehensive assessment of the current status and gaps in genetic epidemiological study of the two polymorphisms in China, and its findings may be useful for medical and public health practices. Future studies are warranted to focus on the interactions of MTHFR genes with environmental exposure and with other genes, and to improve their methodological quality and reporting of findings.

KEYWORDS: A1298C; C677T; Chinese; methylenetetrahydrofolate reductase; polymorphism; systematic review

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