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SUPPLEMENT

The role of MTHFR polymorphism on hyperhomocysteinemia and folic acid and vitamin B metabolism

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Abstract

Non-communicable diseases (NCDs) remain a leading global health concern, with hyperhomocysteinemia recognized as a contributing metabolic disorder linked to cardiovascular disease, neurodegenerative conditions, and developmental anomalies. This presentation explores the pivotal role of methylenetetrahydrofolate reductase (MTHFR) polymorphisms—particularly C677T and A1298C variants—in disrupting homocysteine metabolism through impaired folate and vitamin B pathways. These genetic alterations can reduce enzymatic activity and thermolability of MTHFR, resulting in elevated homocysteine levels and disturbed methylation processes central to the one-carbon cycle. A deficiency in critical B vitamins (B6, B9, and B12), whether dietary or genetically influenced, further exacerbates metabolic imbalance, with implications for DNA synthesis, epigenetic regulation, and oxidative stress. Understanding the biochemical and genetic landscape of MTHFR polymorphisms provides insight into targeted nutritional and clinical strategies for managing NCD risk and improving metabolic health

Keywords: NCD, Hyperhomocysteinemia, MTHFR Variant genotyping, vitamin B6, B9, and B12, Folic Acid Metabolism

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