Identification and phylogenetic characterization of Methylene tetrahydrofolate reductase (MTHFR) gene varients among various genera

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ABSTRACT

Methylene tetrahydrofolate reductase (MTHFR) is a key regulatory enzyme involving in folate and homocysteine metabolism. MTHFR gene is located on chromosomes 1p36.22 and plays a vital role in chemical reactions and involving in vitamin-B9 metabolic pathways. Specifically, these enzymes convert 5, 10-methylene tetrahydrofolate to 5-methyl tetrahydrofolate. This product is used to convert homocysteine to methionine by the enzyme methionine synthase. MTHFR deficiency causes homocystinuria, thrombophilia and metabolic disorders. An MTHFR gene mutation has the ability to alter the metabolic process thereby converting important nutrients leading to changes in hormonal levels. The genetic variations in Homo sapiens do not stop at the boundaries of genus/species level and comparison of these variants may throw a light on better understanding in major diseases such as cancer, cardiac ailments and metabolic disorders. In this current study, an extensive literature search on MTHFR gene variants was performed on multiple databases, public repositories and bioinformatic analysis was executed to compare the variants across different genera (Homo sapiens, Chimpanzee, Rhesus monkey and Orangutan). The identification of these genetic variations provides us evidence on complex genetic diversity of human evolution.

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