Online journal ISSN - 1676-5680
GMR Genetics and Molecular Research
EVOLUTION AND TECHNOLOGY

C677T polymorphism of the methylenetetrahydrofolate reductase gene does not affect folic acid, vitamin B_{12}, and homocysteine serum levels in Turkish children with neural tube defects

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Genet. Mol. Res. 9 (2): 1197-1203 (2010)
Received February 25, 2010
Accepted March 29, 2010
Published June 22, 2010
DOI 10.4238/vol9-2gmr816

ABSTRACT. Association between neural tube defects (NTDs) and C677T polymorphism of the methylenetetrahydrofolate reductase (MTHFR) gene was suspected, because the MTHFR gene codes for a key enzyme in folate metabolism. Its deficiency usually leads to significant reductions in plasma concentrations of folate, vitamin B_{12} and methionine, whereas homocysteine levels are

increased. We examined folate, vitamin B_{12} and homocysteine serum concentrations and polymorphism of the C677T MTHFR gene in Turkish children with neural tube defects. Thirty-three children with NTDs, 26 mothers and 48 healthy individuals were studied. C677T MTHFR polymorphism was determined by melting curve analyses (LightCycler®). The levels of folate, vitamin B_{12} and homocysteine serum concentrations in NTDs were evaluated and compared, along with information concerning alleles of the MTHFR gene. C677T allele frequencies in NTD children and their mothers were similar to those found in controls. Serum folate and vitamin B_{12} concentrations were significantly higher in NTD children than that of controls. Serum homocysteine concentrations were not significantly higher in NTD children and mothers. We concluded that C677T MTHFR gene polymorphism does not affect folic acid, vitamin B_{12} and homocysteine metabolism in Turkish children with NTDs. C677T polymorphism of the MTHFR gene cannot be regarded as a major risk factor for NTDs in Turkish children.

Key words: C677T polymorphism; Folate; Homocysteine; MTHFR; Vitamin B_{12}

