P-44

Genetic polymorphism of methylenetetrahydrofolate reductase as a risk factor for congenital heart defect in Romanian children

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Introduction: Congenital heart defects (CHD) are the most common single group of congenital abnormalities accounting for about 30% of the total abnormalities. Low folate intake as well as alterations in folate metabolism as a result of polymorphisms in the enzyme methylenetetrahydrofolate reductase (MTHFR) have been associated with an increased incidence of neural tube defects, vascular disease, congenital heart defects. To determine whether the C677T and G1793A variants in the MTHFR gene is associated with CHD susceptibility we utilised a case-control study.
Methods: DNA samples were isolated from peripheral blood samples and genotyped using polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP), with the digestion of restriction endonuclease HinfI and BsrBI. The restricted products were analyzed on 2% agarose gel.
Results: Subjects were consisted of 20 patients with CHD and 20 healthy controls. In patients, the genotypes frequencies of the MTHFR G1793A polymorphism were 22% and 88% for the GA and GG genotype respectively, whereas the genotypes frequencies of the MTHFR C677T polymorphism were 47.4%, 47.4% and 5.2% for CC, CT and TT genotypes respectively. The frequency of MTHFR 677CT+TT/1793GA was higher in the patients than controls.
Conclusions: The MTHFR C677T and G1793A polymorphism may influence congenital heart defect, but the MTHFR polymorphisms need to be studied further for confirmation in larger studies.
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