Letter to the Editor

MTHFR gene polymorphisms analyzed in population from Kolkata, West Bengal

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Sir,

Importance of hyperhomocysteinemia in association with several pathological conditions as well as ischemic stroke^[1-3] is a well-discussed subject. Methylenetetrahydrofolate reductase (*MTHFR*) enzyme converts 5,10-methylenetetrahydrofolate to 5-methyltetrahydrofolate (the predominant folate in circulation), which acts as a methyl donor for remethylation of homocysteine (Hcy) to generate methionine. Optimum activity of *MTHFR* is, therefore, essential for both DNA synthesis/methylation and in maintaining Hcy level. Genetic polymorphisms in *MTHFR*, leading to disruption in enzymatic activity, may perturb this phenomenon and eventually may cause an increase in Hcy.

Ethnicity of population is one of the most important aspects for conducting genetic studies. We have investigated two polymorphisms in *MTHFR* in cardiovascular disease (CVD) patients (N=30) and control individuals (N=30), both groups being native inhabitants of Kolkata, West Bengal. Allelic frequencies for the T allele of C677T and for the C allele of A1298 C were 0.13 and 0.38, respectively. Allelic frequencies for the T and C alleles of the two polymorphisms in control individuals were 0.17 and 0.45, respectively. No significant difference between the two groups was observed for the polymorphisms studied.

Earlier investigators have failed to detect the T allele of C677T in subjects from West Bengal (N=36); however, values obtained for the A1298C in their study were comparable to that observed in the present investigation. Significant correlation between the presence of hyperhomocysteinemia and the presence of 1298 C allele was reported in CVD patients, while no significant association was reported with the C677T.^[2] The C677T polymorphism was studied in the North Indian population (N=32); the "T" allele in homozygous condition was found to be associated with hyperhomocysteinemia in three out of four stroke patients and folate depletion was observed in two out of three patients.^[3] Significant association with the C677T polymorphism with CVD and hyperhomocysteinemia was also documented earlier from other parts of India.^[4] Further new born screening program was suggested to "reduce mortality and morbidity rate associated with hyperhomocysteinemia".^[1]

Data obtained in the present investigation for the C677T polymorphism in population from Kolkata does not match with that reported previously in population from West Bengal.^[2] It could be inferred that it is necessary to explore homocysteine level as well as genetic polymorphisms in the *MTHFR* in large number of individuals from different locations of the country to correlate the contributions made by genetic polymorphisms in the etiology of CVD.

References

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