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Analysis of MTHFR gene C.677c>t and C.1298a>c polymorphisms in Iranian patients with non-syndromic cleft lip and palate

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Non-syndromic cleft lip with or without cleft palate (nsCL/P) is one of the most common congenital abnormalities of the orofacial region with a multifactorial etiology. The present study aimed to investigate the association of two common polymorphisms of methylenetetrahydrofolate reductase (MTHFR) gene (c.677C>T and c.1298A>C) with the occurrence of nsCL/P in an Iranian population. Forty-five nsCL/P patients, 43 mothers of patients, and 101 unrelated controls participated in the present study. Analysis of c.677C>T and c.1298A>C polymorphisms in MTHFR gene was conducted using polymerase chain reaction and restriction enzyme digestions. There was no statistical difference in genotype and allele frequencies for c.677C>T variants between patients or their mothers and the control group. However, differences in the frequencies of alleles and genotypes of c.1298A>C polymorphism were statistically significant between patients and control group (P=0.01 for alleles and P=0.005 for genotypes). The odds ratios (OR) for the CC versus AA homozygotes were 6.1 (95% CI 1.8-20.5) and 4.2 (95% CI 1.1-15.4), in patients and mothers, respectively. We found no association between genetic polymorphism of MTHFR c.677C>T and the risk of nsCL/P in the population studied. Yet the results suggested that c.1298A>C polymorphism of MTHFR gene may be a risk factor for the occurrence of nsCL/P in the Iranian population.

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