



Risk of colorectal cancer associated with the methylenetetrahydrofolate reductase (*MTHFR*) C677T polymorphism in the Kashmiri population

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ABSTRACT. Methylenetetrahydrofolate reductase (*MTHFR*) is a critical enzyme in folate metabolism and is involved in DNA synthesis, DNA repair and DNA methylation. The two common functional polymorphisms of *MTHFR*, 677 C→T and 1298 A→C, have been shown to impact various diseases, including cancer. The 677 C→T polymorphism has been widely investigated in different cancers and has been implicated as a risk factor for the development of various cancers. We investigated *MTHFR* C677T genotype frequency in colorectal cancer cases in the Kashmiri population and correlated this information with the known clinicopathological characters of colorectal cancer, in a case-control study. Eighty-six colorectal cancer cases were studied for *MTHFR* C677T polymorphism, compared to 160 controls taken from the general population, employing the PCR-RFLP technique. We found the frequency of the three different genotypes of *MTHFR* in our ethnic Kashmir population, i.e., CC, CT and TT, to be

68.6, 20.9 and 10.4% among colorectal cancer cases and 75.6, 16.9 and 7.5% among the general control population, respectively. There was a significant association between the *MTHFR* TT genotype and colorectal cancer in the higher age group. We conclude that the *MTHFR* C677T polymorphism slightly increases the risk for colorectal cancer development in our ethnic Kashmir population.

Key words: Colorectal cancer; *MTHFR*; Polymorphism; RFLP; Restriction digestion; Kashmir