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POLYMORPHIC VARIATION OF MONONUCLEOTIDE MICROSATELLITES AND ITS IMPLICATION FOR MICROSATELLITE INSTABILITY SCREENING

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Background - Colorectal cancer (CRC) is the sixth most common tumor and the fifth in mortality in Brazil. Molecular markers have been associated with disease prognosis, especially in relation to therapeutic response and overall survival rates. Among these, microsatellite instability (MSI) has been extensively studied. Microsatellite stability status is usually determined by comparison of normal and tumoral tissues from the same patient and instability is characterized by the difference in the PCR-amplification profile of these tissues at a given locus. Usually, a panel of five markers is used for this purpose. Two of them (BAT-25 and BAT-26) are considered monomorphic in populations of European origin. Aim - The aim of this study was to analyze the frequency of constitutive polimorphic variaion at BAT-25 and BAT-26 loci in a sample of individuals from Southern Brazil. Patients/Methods – Two-hundred and sixteen healthy and unrelated individuals were analised to assess the frequency of allelic variation at the BAT-25 and BAT-26 loci in DNA extracted from peripheral blood. Analysis was done by PCR-SSCP. Results – From the sample of patients studied, 7% and 6% of the patients had possible constitutive alelic variation at the BAT-25 and BAT-26 loci, respectively. Conclusions - These results indicate that significant constitutive allelic variation of these loci does occur in heterogeneous populations such as ours, and reinforce the importance of comparative studies between tumoral and corresponding normal tissue to determine microsatellite stability status and correctly identify MSI in selected populations.