

Association of *IL4* (rs79071878) and *TYMS* (rs51264360) Polymorphisms with Colorectal Cancer (CRC) Risk in Brazilian Populations

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INTRODUCTION: Colorectal cancer (CRC) is the third most common cancer in men and the second in women, worldwide. CRC is associated with various risk factors, but the genetic factors are a major component of CRC predisposition. Among several mutations that may occur in the human DNA, the influence of Insertion-Deletions (INDEL) on cancer risk has barely been explored in an admixed population, especially in Brazil. Moreover, to date, no study evaluated the possible effect of INDEL polymorphism on CRC risk in an admixed population. **OBJECTIVES:** Thus, in this study, we investigated the possible effect of 16 INDEL (rs4646994, rs34667759, rs3834129, rs56228771, rs28892005, rs371194629, rs3783553, rs3730485, rs28362491, rs11267092, rs17878362, rs17880560, rs151264360, rs3213239, *CYP2E1* [96 bp], *UCP2* [45 bp]), and 2 Variable Number Tandem Repeat (VNTR)

(rs79071878 and rs8175347) polymorphisms in 140 patients with CRC and 140 healthy individuals from Rio Grande do Norte state, Brazil. We also evaluated the autosomal ancestry distribution between cases and controls, considering that the Brazilian population is genetically influenced by many ethnic groups. **MATERIAL AND METHODS:** The polymorphisms and ancestry distribution were typed by Multiplex polymerase chain reaction (PCR) using the ABI PRISM 3130 and analyzed with GeneMapper ID v3.2. **RESULTS AND DISCUSSION:** The groups studied were uniform in term of ancestry distribution ($p > 0.05$). After multivariate logistic regression, the polymorphism variations in *IL4* (rs79071878) and *TYMS* (rs151264360) genes were associated with increased ($p = 0.003$) and decreased ($p = 0.023$) CRC risk, respectively. **CONCLUSION:** In summary, the association of *IL4* and *TYMS* polymorphisms with CRC risk suggests that these polymorphisms could be useful to predict disease outcome at diagnosis and establish future therapeutic targets. Furthermore, this is the first study showing the association of this *IL4* polymorphism with the risk of CRC development.

Key words: Admixed Population, Biomarker, Risk Association

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