

RAD51 135G>C polymorphism and risk of familial breast cancer in a South American population

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Several studies have reported that mutations in genes involved in maintenance of genome integrity may be responsible for increased cancer risk. Human RAD51, known to function in DNA repair, interacts with a number of proteins implicated in breast cancer (BC), including BRCA1 and BRCA2. Few studies have investigated the role of RAD51 gene variations in familial BC. To detect potential novel gene defects that may contribute to hereditary BC susceptibility, 143 patients belonging to 143 Chilean families tested for BRCA1 and BRCA2 mutations were screened for mutations in RAD51, using conformational sensitive gel electrophoresis (CSGE) and DNA sequencing. No mutations were detected in the exon or splice-boundary regions of the RAD51 gene in these families. The RAD51 135G>C polymorphism (c.-98G>C, rs1801320) was studied in a case-control design, to evaluate its possible association with BC susceptibility. The frequency of the RAD51 135C allele was established in 143 cases and 247 controls, u