Variants in DNA double-strand break repair genes and risk of familial breast cancer in a South American population

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The double-strand break (DSB) DNA repair pathway has been implicated in breast cancer (BC). RAD51 and its paralogs XRCC3 and RAD51D play an important role in the repair of DSB through homologous recombination (HR). Some polymorphisms including XRCC3-Thr241Met, RAD51-135G>C, and RAD51D-E233G have been found to confer increased BC susceptibility. In order to detect novel mutations that may contribute to BC susceptibility, 150 patients belonging to 150 Chilean BRCA1/2-negative families were screened for mutations in XRCC3. No mutations were detected in the XRCC3 gene. In addition, using a case-control design we studied the XRCC3-Thr241Met, and RAD51D-E233G polymorphisms in 267 BC cases and 500 controls to evaluate their possible association with BC susceptibility. The XRCC3 Met/Met genotype was associated with an increased BC risk (P = 0.003, OR = 2.44 [95%CI 1.34-4.43]). We did not find an

association between E233G polymorphism and BC risk. We also analyzed the effect of combined genotyp