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Susceptibility alleles of breast cancer in high, moderate and low penetrance genes in a South American population

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Breast cancer (BC) is the most common cancer among women worldwide. There are few studies of BC susceptibility alleles in South American populations. Here we describe the identification of risk variants in genes BRCA1, BRCA2 (high penetrance), CHEK2, RAD51, XRCC3 (moderate penetrance), rs2981582 and rs1219648 (low penetrance) in Chilean families at high-risk for breast/ovarian cancer (n=326). Germline BRCA1/2 point mutations were found in 7.1% of families. Families with at least three BC and/or OC cancer cases showed the highest frequency of mutation (15.9%). We identified 14 point mutations, of which 3 in BRCA1 and 3 in BRCA2 were recurrent, possibly reflecting region-specific founder effects. In CHEK2, the 1100delC mutation was not detected in the 1320 samples analyzed. The Thr241Met (XRCC3) polymorphism was associated with increased BC risk (OR=2.44, 95%CI=1.34–4.43). The RAD51 135G>C polymorphism increased BC risk among BRCA1/2-negative women with a) a family history of BC and b) age at onset <50 years (OR=2.17, 95%CI=1.11-4.29). The combined Thr/Met–E/G (RAD51D) genotype was associated with increased BC risk among the same group of women (OR=10.5 [95%CI 1.16–94.5]). Our results suggest that variability in XRCC3 and RAD51D plays a role in BC risk via a mutual interaction between the genes. The combined SNPs rs2981582 – rs1219648 (A/A–G/G) of FGFR2 genotypes were associated with increased risk for estrogen-receptor positive BC (OR=2.6, 95%CI=1.2–5.6]). Our results are consistent with a polygenic model for familial BC susceptibility.

Biography

Lilian Jara completed her PhD at 30 at a Chilean University, and postdoctoral studies at Sheffield University (UK). She is a full professor in the Human Genetics Program, University of Chile School of Medicine. She has published over 60 papers in reputed journals and serves as a WJMG editorial board member. She conducts longitudinal studies of women from Chilean families with genetic mutations related to breast cancer. She is also interested in gene-gene interactions underlining hereditary breast cancer. She collaborates with research groups studying the genetics of breast cancer from Argentina, Colombia, Spain, Canada, and the USA.