The BARD1 Cys557Ser variant and risk of familial breast cancer in a South-American population

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Since the discovery of the BRCA1 and BRCA2 genes, much work has been carried out to identify further breast cancer (BC) susceptibility genes. BARD1 (BRCA1-associated ring domain) was originally identified as a BRCA1-interacting protein but has also been described in tumor-suppressive functions independent of BRCA1. Some association studies have suggested that the BARD1 Cys557Ser variant might be associated with increased risk of BC, but others have failed to confirm this finding. To date, this variant has not been analyzed in Spanish or South-American populations. In this study, using a case-control design, we analyzed the C-terminal Cys557Ser change in 322 Chilean BC cases with no mutations in BRCA1 or BRCA2 and in 570 controls in order to evaluate its possible association with BC susceptibility. BARD1 Cys557Ser was associated with an increased BC risk (P = 0.04, OR = 3.4 [95 % CI 1.2-10.2]) among cases belonging to families with a strong family history of BC. No difference between si