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Abstract

BRCA1 and BRCA2 Gene Mutations in Breast/Ovarian Cancer Patients from Central and Southern Italy

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Using PTT combined with SSCP, we screened the entire BRCA1 and BRCA2 coding sequences in 136 unrelated patients, recruited from oncology units in Rome and Naples and including 107 females with breast cancer, 10 with ovarian cancer and 19 males affected with breast cancer. Female patients were recruited on the basis of family history, mostly indicative of moderate risk, young age and/or bilateral breast cancer or breast cancer associated with other tumors. Germline BRCA1 and BRCA2 mutations occurred in the same number of female breast cancer patients (~4% of the cases each). BRCA1 mutations were found in 10% of ovarian cancer patients, and BRCA2 mutations in 10% of male breast cancer patients. No BRCA1 mutations were found in male breast cancers and no BRCA2 mutations in ovarian cancers. Relative to BRCA1 mutations, BRCA2 mutations occurred in familial contexts with a more variable disease penetrance. To further evaluate the epidemiological relevance of BRCA 2 mutations, a population-based series of 25 unrelated male breast cancer patients identified in Florence was screened along the entire BRCA2 coding sequence. Germline BRCA2 mutations were identified in three cases (3/25, 12%). One of the three mutations, BRCA2 6696delTC, also occurred in two unrelated female breast cancer patients from the series recruited from Rome and Naples. Combining the two series of patients, BRCA2 mutations were identified in 9 of 161 index patients: interestingly 3 out of 9 (33%) were 6696delTC carriers, which suggests that BRCA2 6696delTC may recur in Italy. The other BRCA1 and BRCA2 mutations were unique and included variants previously reported in high-risk families. (Study supported by an AIRC "CG" grant. CDA is an AIRC fellow.)