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Meeting abstract

Molecular characterization of BRCA1 and BRCA2 genes in breast cancer patients under 40 years-old or with familiar cancer history

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Background

Breast and ovarian cancer are the most frequent causes of death in women, generating an important problem. A small proportion of these tumors results from alterations in cancer susceptibility genes. Two of these genes are BRCA1 and BRCA2, which are described as hereditary breast and ovarian cancer genes. BRCA1 mutations have been identified in 15 to 20% of women with familiar breast cancer, and in 60 to 80% of women with familiar breast and ovarian cancer; to date over 500 sequence variants have been reported for this gene. It is estimated that mutations in BRCA2 cause approximately 35% of hereditary breast cancer cases, besides, is associated with breast cancer in men, ovarian, pancreas and prostate cancer. Specific mutations to particular ethnic groups have been described. There are no known mutations for the Mexican population. The purposes of this work were to determine the mutation frequency of BRCA1 and BRCA2 genes in breast cancer patients under 40 years old or patients with familiar breast and/or ovarian cancer history, through DHPLC analysis and, to establish genotype-phenotype correlations. Those families with mutations will be followed-up for an early detection, and will receive genetic counseling.

Materials and methods

Forty breast and/or ovarian cancer patients were included. DNA was obtained from peripheral leukocytes, and was amplified for the 24 exons of BRCA1 using 31 pairs of oligonucleotides, and for the 26 exons of BRCA2 with 39 pairs of primers. The primers were designed to include each exon flanked by a small portion of the corresponding introns. The amplifications were analyzed in a DHPLC (Transgenomics).

Results

We have analyzed the entire BRCA2 gene for the 40 patients, and found 1 polymorphism at exon 4 in 4 patients and 4 different polymorphisms at exon 11 in 24 patients. We identify a patient with a mutation in exon 11 already reported at Breast Cancer International Core (BIC), which was also present in her father, for this reason the rest of the family will be analyzed and receive the appropriate genetic counseling. Regarding BRCA1, we have completed the amplifications, and we are now performing the DHPLC analysis.

Conclusion

We have found an elevated percentage of patients with polymorphisms (up to 30%). Until now we have found only one mutation, which has been reported only three times in the BIC. It seems that mutations in BRCA2 are not frequent in our hereditary breast cancer patients.