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BRCA1 mutation analysis in breast/ovarian cancer families from Greece.

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Source

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Abstract

Germline mutations in BRCA1 gene account for varying proportions of breast/ovarian cancer families, and demonstrate considerable variation in mutational spectra coincident with ethnic and geographical diversity. We have screened for mutations the entire coding sequence of BRCA1 in 30 breast/ovarian cancer women with family history of two or more cases of breast cancer under age 50 and/or ovarian cancer at any age. Genomic DNA from patient was initially analyzed for truncating mutations in exon 11 with PTT followed by DNA sequencing. In the cases where no frameshift mutation was observed in exon 11, all other exons were screened with direct sequencing. Two novel (3099delT, 3277insG) and three already described (3741insA, 1623del5-TTAAA, 5382insC-twice) truncating mutations were identified. In addition, 6 point mutations (L771L, P871L, E1038G, K1183R, S1436S, S1613G) which are already classified as polymorphisms were identified. Three unclassified intronic variants (IVS16-68 G>A, IVS16-92 G>A, IVS18+65G>A) were also detected. These results show that BRCA1 deleterious mutations are present in a fraction (20%) of Greek breast/ovarian cancer families similar to other European countries. Mutations were detected in high- (>/=3 members) as well as in moderate-risk (2 members) families. This is the first report of BRCA1 mutation analysis in Greece.

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