Greek BRCA1 and BRCA2 mutation spectrum: two BRCA1 mutations account for half the carriers found among high-risk breast/ovarian cancer patients.

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Source

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Abstract

127 Greek breast/ovarian cancer families were screened for germline BRCA1/2 mutations by dHPLC followed by direct sequencing. Our results indicated 16 and 5 breast/ovarian cancer families bearing deleterious mutations in the BRCA1 and BRCA2 genes, respectively. Two novel BRCA2 germline mutations (G4X and 3783del10) are reported here for the first time. Subsequent compilation of our present findings with previously reported mutation data reveals that in a total of 287 Greek breast/ovarian cancer families, 46 and 13 carry a deleterious mutation in BRCA1 and BRCA2, respectively. It should be noted that two BRCA1 mutations, 5382insC and G1738R, both located in exon 20, account for 46% of the families found to carry a mutation. Based on our mutation analysis results, we propose here a hierarchical, cost-effective BRCA1/2 mutation screening protocol for individuals of Greek ethnic origin. The suggested protocol can impact on the clinical management of breast-ovarian cancer families on a national healthcare system level.