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High prevalence of BRCA1 founder mutations in Greek breast/ovarian families.

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Abstract

We have screened 473 breast/ovarian cancer patients with family history, aiming to define the prevalence and enrich the spectrum of BRCA1/2 pathogenic mutations occurring in the Greek population. An overall mutation prevalence of 32% was observed. Six BRCA1 recurrent/founder mutations dominate the observed spectrum (58.5% of all mutations found). These include three mutations in exon 20 and three large genomic deletions. Of the 44 different deleterious mutations found in both genes, 16 are novel and reported here for the first time. Correlation with available histopathology data showed that 80% of BRCA1 carriers presented a triple-negative breast cancer phenotype while 82% of BRCA2 carriers had oestrogen receptor positive tumours. This study provides a comprehensive view of the frequency, type and distribution of BRCA1/2 mutations in the Greek population as well as an insight of the screening strategy of choice for patients of Greek origin. We conclude that the Greek population has a diverse mutation spectrum influenced by strong founder effects.