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Germ line BRCA1 & BRCA2 mutations in Greek breast/ovarian cancer families: 5382insC is the most frequent mutation observed.

Ladopoulou A, Kroupis C, Konstantopoulou I, Ioannidou-Mouzaka L, Schofield AC, Pantazidis A, Armaou S, Tsiagas I, Lianidou E, Efstathiou E, Tsionou C, Panopoulos C, Mihalatos M, Nasioulas G, Skarlos D, Haites NE, Fountzilas G, Pandis N, Yannoukakos D.

Source

Molecular Diagnostics Laboratory, I/R-RP, National Center for Scientific Research 'Demokritos', Ag. Paraskevi Attikis, 15310, Athens, Greece.

Abstract

BRCA1 and BRCA2 genes were screened for loss-of-function mutations in a series of 85 patients having at least one first- or second-degree relative affected by breast and/or ovarian cancer. All BRCA1 exons and BRCA2 exons 10 and 11 were screened with a combination of methods including SSCP, PTT and direct sequencing. We have found disease-associated mutations in 14 families (16.5%), eleven in BRCA1 and three in BRCA2. The known founder mutation 5382insC of BRCA1 was identified in seven unrelated families. The other mutations identified include the non-sense R1751X, the splice junction variant 5586G>A of BRCA1 and three frameshifts, 2024del5, 3034del4, and 6631del5, of BRCA2. Nine out of these 14 families had a family history of three or more breast/ovarian cancer cases. A large number of polymorphic or unclassified variants is also reported. Combined with our previously published data 5382insC was found in nine out of 20 families (45%), suggesting that this mutation may represent a common founder mutation in the Greek population.