Contribution of BRCA1 germ-line mutations to breast cancer in Greece: a hospital-based study of 987 unselected breast cancer cases.

Armaou S, Pertesi M, Fostira F, Thodi G, Athanasopoulos PS, Kamakari S, Athanasiou A, Gogas H, Yannoukakos D, Fountzilas G, Konstantopoulou I.

Source

Molecular Diagnostics Laboratory, I/R-RP, National Center for Scientific Research Demokritos, Aghia Paraskevi, Athens, Greece.

Abstract

BACKGROUND:

In most Western populations, 5-10% of all breast cancer cases can be attributed to major genetic factors such as predisposing mutations in BRCA1 and BRCA2, with early-onset cases generally considered as an indicator of genetic susceptibility. Specific BRCA1 and BRCA2 mutations or different mutation frequencies have been identified in specific populations and ethnic groups. Previous studies in Greek breast and/or ovarian cancer patients with family history have shown that four specific BRCA1 mutations, c.5266dupC, G1738R, and two large genomic rearrangements involving deletions of exons 20 and 24, have a prominent function in the population's BRCA1 and BRCA2 mutation spectrum.

METHODS:

To estimate the frequency of the above mutations in unselected Greek breast cancer women, we screened 987 unselected cases independently of their family history, collected from major Greek hospitals.

RESULTS:

Of the 987 patients, 26 (2.6%) were found to carry one of the above mutations in the BRCA1 gene: 13 carried the c.5266dupC mutation (1.3%), 6 carried the exon 24 deletion (0.6%), 3 carried the exon 20 deletion (0.3%), and 4 carried the G1738R mutation (0.4%). Among 140 patients with early-onset breast cancer (<40 years), 14 carried one of the four mutations (10.0%).

CONCLUSION:

These results suggest that a low-cost genetic screening for only the four prominent BRCA1 mutations may be advisable to all early-onset breast cancer patients of Greek origin.