Cancer Genet. 2015 Apr;208(4):129-34. doi: 10.1016/j.cancergen.2015.02.006. Epub 2015 Feb 20.

CHEK2 c.1100delC allele is rarely identified in Greek breast cancer cases.

Apostolou P[1], Fostira F[2], Papamentzelopoulou M[2], Michelli M[2], Panopoulos C[3], Fountzilas G[4], Konstantopoulou I[2], Voutsinas GE[5], Yannoukakos D[6].

Abstract

The CHEK2 gene encodes a protein kinase that plays a crucial role in maintenance of genomic integrity and the DNA repair mechanism. CHEK2 germline mutations are associated with increased risk of breast cancer and other malignancies. From a clinical perspective, the most significant mutation identified is the c.1100delC mutation, which is associated with an approximately 25% lifetime breast cancer risk. The distribution of this mutation shows wide geographical variation; it is more prevalent in the Northern European countries and less common, or even absent, in Southern Europe. In order to estimate the frequency of the CHEK2 c.1100delC mutation in Greek breast cancer patients, we genotyped 2,449 patients (2,408 females and 41 males), which was the largest series ever tested for c.1100delC. The mean age of female and male breast cancer diagnosis was 49 and 59 years, respectively. All patients had previously tested negative for the Greek BRCA1 founder and recurrent mutations. The CHEK2 c.1100delC mutation was detected in 0.16% (4 of 2,408) of females, all of whom were diagnosed with breast cancer before the age of 50 years. Only one c.1100delC carrier was reported with breast cancer family history. The present study indicates that the CHEK2 c.1100delC mutation does not contribute substantially to hereditary breast cancer in patients of Greek descent.