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Frequency of rearrangements versus small indels mutations in BRCA1 and BRCA2 genes in Turkish space patients with high risk breast and ovarian cancer

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Statement of the Problem: The current rearrangement ratio of BRCA1 and BRCA2 genes is not known in the Turkish population. Rearrangements are not routinely investigated in many Turkish laboratories. This creates problems and contradictions between clinics. Therefore, the aim of this study was to evaluate the distribution and frequency of rearrangements in BRCA1 and BRCA2 genes in high-risk families and to clarify the limits of BRCA1 and BRCA2 testing in Turkey.

Methodology: The study included 1809 patients at high risk of breast cancer or ovarian cancer. All patients were investigated for both small indels and rearrangements of BRCA genes using DNA sequencing and multiplex ligation-dependent probe amplification (MLPA) analysis.

Findings: The overall frequency of rearrangements was 2% (25/1262). The frequency of rearrangements was 1.7% (18/1086) and 4% (9/206) in patients with breast cancer and ovarian cancer, respectively. The frequency of rearrangements was 3.7% (8/215) in patients with triple-negative breast cancer. The rearrangement rate was 7.7% (2/26) in patients with both breast and ovarian cancer.

Conclusion & Significance: Rearrangements were found with high rates and were strongly associated with bilateral and triple-negative status of patients with breast cancer, which are signs of high risk for breast and ovarian cancer. Analysis of rearrangements should definitely be included in routine clinical practice in Turkey for high-risk families and also for improved cancer risk prediction for families.

Biography

I love this work because it keeps me motivated intrinsically which is the utmost necessity for success. Research is an organized method that keeps all scientist dissatisfied with what you have discovered and this dissatisfaction is what drives more and more discovery. For so many years the question: 'How to cure cancer?', which has gone unanswered drives me so ambitious in order to find the answer. I believe that the work conducted our lab may help cancer patients always improve my motivation.

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