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Contribution of BRCA1 5382insC mutation to triplene-gative and luminal types of breast cancer in Ukraine

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Abstract

Purpose: The gene BRCA1 plays a key role in DNA repair in breast and ovarian cell lines and this is considered one of target tumor suppressor genes in same line of cancers. The 5382insC mutation is among the most frequently detected in patients (Eastern Europe) with triple-negative breast cancer (TNBC). In Ukraine, there is not enough awareness of necessity to test patients with TNBC for BRCA1 mutations. That is why this group of patients is not well-studied, even through is known the mutation may affect the course of disease.

Methods: The biological samples of 408 female patients were analyzed of the 5382insC mutation in BRCA1. We compared the frequency of the 5382insC mutation in BRCA1 gene observed in Ukraine with known frequencies in other countries.

Results: For patients with TNBC, BRCA1 mutations frequency was 11.3%, while in patients with luminal types of breast cancers, the frequency was 2.8%. Prevalence of 5382insC among TNBC patients reported in this study was not different from those in Tunisia, Poland, Russia, and Bulgaria, but was higher than in Australia and Germany.

Conclusion: The BRCA1 c.5382 mutation rate was recorded for the first time for TNBC patients in a Ukrainian population. The results presented in this study underscore the importance of this genetic testing of mutations in patients with TNBC. Our study supports BRCA1/2 genetic testing for all women diagnosed with TNBC, regardless of the age of onset or family history of cancer and not only for women diagnosed with TNBC at <60y.o., as guidelines recommend.

Keywords: 5382insC mutation of BRCA1; Luminal breast cancer; Triple-negative breast cancer; Ukraine.

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