

FREQUENCY OF BRCA1 AND BRCA2 FOUNDER MUTATIONS AMONG OVARIAN CANCER PATIENTS FROM PODKARPACKIE VOIVODESHIP.

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It was shown that about 12-13% of ovarian cancers in Polish population are caused by founder mutations in BRCA1 gene. Some regional differences were reported. In our study we evaluated the frequency of BRCA1 and BRCA2 founder mutations among ovarian cancer patients from Podkarpackie Voivodeship. DNA testing for 8 BRCA1 and 5 BRCA2 founder mutations was performed in 160 consecutive patients diagnosed with ovarian cancer in Rzeszów Regional Hospital in 2013-2016. BRCA1 mutation was diagnosed in 10 (6,25%) patients. BRCA1 mutations - 5382 ins C was observed in 6 patients; ex.5 300 T G in 3 patients; and 794delT I 1 patient. No BRCA2 mutation was diagnosed. The frequency as well as mutation spectrum in some extent are different from general population. BRCA1/2 NGS should be performed in larger number of patients.