

Optimization of the genetic diagnostics algorithm in patients with ovarian cancer from south-eastern Poland

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Abstract

Background: Mutations in *BRCA1* and *BRCA2* genes are well-established risk factors of breast and ovarian cancer. In our former study, we observed that approximately 6% of unselected ovarian cancer patients in the region of Podkarpacie (South-East Poland) carry *BRCA1* causative founder variants, which is significantly lower than in other regions of Poland. Therefore, it is deeply justified to do research based on the sequencing of whole *BRCA1* and *BRCA2* genes.

Methods: We examined 158 consecutive unselected cases of ovarian cancer patients from the region of Podkarpacie. We performed *BRCA1* and *BRCA2* genes Next-Generation Sequencing study in all cases.

Results: Altogether, in 18 of 158 (11.4%) ovarian cancer patients with *BRCA1* or *BRCA2* pathogenic mutations were found. *BRCA1* pathogenic variants were detected in 11 of the 158 (7.0 %) ovarian cancer cases. 10 of 11 (91%) detected *BRCA1* mutations were founder mutations, detectable with the standard test used in Poland. *BRCA2* pathogenic variants were found in 7 of the 158 (4.4 %) cases. No *BRCA2* pathogenic variants were founder mutations. The median age of patients at the diagnosis of the 18 hereditary ovarian cancers was 57.5 years.

Conclusions: The frequency of *BRCA1* or *BRCA2* gene mutation carriers among patients with ovarian cancer from the Podkarpacie region is comparable to other regions of Poland. However, a significantly higher percentage of *BRCA2* gene mutations was observed, that were not detectable with a standard test for detection of founder mutations. Diagnostics based only on testing the *BRCA1/2* Polish founder mutations is characterized by relatively low sensitivity in the case of ovarian cancer patients from South-East Poland and should be supplemented by NGS study, in particular of the *BRCA2* gene.