Association of CHEK2, PALB2, RAD51C mutations with borderline ovarian tumors in patients from Polish population.

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Borderline ovarian tumors (BOT) are heterogeneous group of noninvasive tumors of uncertain malignant potential and constitute 10-15% of surface epithelial ovarian tumors. BOT are usually diagnosed at an early stage and affect younger women, aged between 20 and 40 years old. In general, prognosis in BOT is favorable, however 11% of tumors may recur and 20-30% of them may undergo malignant transformation. It has been shown that BOT share molecular and genetic alterations with low-grade serous carcinomas.

The aim of the study was to analyze association of RAD51C, PALB2 and CHEK2 mutations with the risk of borderline ovarian tumors among Polish patients.

Molecular analyses included genotyping of recurrent mutations in RAD51C (3), PALB2 (2) and CHEK2 (3) in a group of 340 patients with borderline ovarian tumors and approximately 2000 healthy controls.

We found significant association of CHEK2, but not RAD51C and PALB2 recurrent mutations with the risk of borderline ovarian tumors.

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