BRCA1/2, CHEK2, PALB2 and *RAD51C* mutations in ovarian cancer patients from Polish population

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In Poland ovarian cancer represents the second cause of cancer among gynecological malignancies and the fourth cause of cancer deaths among women. Literature data shows that more than one-fifth of ovarian cancer cases have been related to hereditary factors. It has been recognized that the most frequently germline mutation in hereditary ovarian cancer are *BRCA1/2* mutations. Nevertheless, several other genes, as *RAD51C*, have been suggested to be associated with hereditary ovarian cancer. Mutations in other genes which are known to be associated with high breast cancer risk in Polish population, as *PALB2* and *CHEK2*, have not been tested in ovarian cancer patients up to now.

The aim of the study is to estimate the frequency of recurrent Polish germline mutations in *BRCA1/2, RAD51C, PALB2* and *CHEK2* genes among unselected and familial ovarian cancer patients. Additionally, an association of *RAD51C, PALB2* and *CHEK2* mutations with ovarian cancer risk was assessed.

Molecular analyses included genotyping of recurrent mutations in *BRCA1/2* (13), *RAD51C* (3), *PALB2* (2) and *CHEK2* (3) in a group of ~2000 unselected OC, ~250 HOC and 2000 healthy controls .

The frequency of *BRCA1/2* was 11.02%. We found significant association of *RAD51C* and *PALB2* mutations, but not *CHEK*2 mutations, with ovarian cancer risk.

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