Summary

Despite the growing knowledge about ovarian cancer, it has not yet been possible to develop an effective screening test for this cancer. Therefore, it seems necessary to identify new risk factors, such as genetic polymorphisms. Ovarian cancer is a hormone-dependent cancer and its steroid hormones are estrogens. Estrogens affect cells through the estrogen receptors ERα and ERβ. An imbalance between ERα and ERβ receptor expression may therefore be a key critical step in estrogen-dependent carcinogenesis. In 60% of cancer cases, significantly elevated levels of ERα receptors are detected. The ERα receptor is encoded by the *ESR1* gene. The *ESR1* gene is a polymorphic gene.

In this study, the aim was to demonstrate whether polymorphisms of the *ESR1* gene rs2234693 and rs9340799 may be involved in the development of ovarian cancer.

The material for the study consisted of 100 paraffin blocks containing specimens from ovarian cancerous tumors and 100 paraffin blocks containing specimens from benign ovarian lesions, collected in the archives of the Department of Clinical Pathomorphology of the Polish Mothers Memorial Hospital Research Institute in Lodz. DNA was isolated from the above preparations. The polymorphisms were determined by the PCR-RFLP technique.

Statistical analysis of the distribution of genotypes and alleles in the study and control groups was carried out after it was previously confirmed that the obtained systems remain in equilibrium according to the Hardy and Weinberg rule. The result was considered statistically significant with a significance level p less than 0.05. The assessment of genotypes and alleles in terms of their relationship to a given trait was carried out by using the analysis of the odds ratio (OR) and the 95% confidence interval, which were calculated according to the logistic regression model. The wild-type genotype and allele was the reference group.

It has been shown that the presence of the CC genotype (rs2234693) more than doubles the risk of ovarian cancer. The presence of the TT genotype (rs2234693) significantly reduces the risk of developing this type of cancer. In the case of the second of the studied polymorphisms - rs9340799, carrier of the GG genotype more than doubles the risk of ovarian cancer. Analysis of the *ESR1* gene haplotypes in relation to the rs2234693 and rs9340799 polymorphisms showed that the occurrence of the TCAG and CCGG systems may be associated with a significant increase in the risk of ovarian cancer.

In the group of patients with ovarian cancer, a correlation was found between rs2234693 and rs9340799 polymorphisms in the tissues of the lowest stage ovarian cancers compared to more advanced ovarian cancers, which may indicate a relationship between these factors and

the stage of the cancer. The age of the women had no effect on the prevalence of individual genotypes, nor on the associated risk of disease.

The study shows that the polymorphisms rs2234693 and rs9340799 of the *ESR1* gene are characterized by a statistically significant relationship with the occurrence of ovarian cancer.

The results indicate that in women with ovarian cancer, *ESR1* rs2234693 and rs9340799 polymorphisms may be associated with the occurrence of this disease. The presented work indicates that on the basis of genetic studies of variants of these polymorphisms, patients can be classified into the group of increased risk of cancer development. However, further work is needed on much larger groups of subjects.