P8

The genetic factor in the peculiarities of clinical development of endometrial cancer in patients with aggravated oncopathology of clinico-genealogical anamnesis

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Endometrial cancer refers to the multifactor pathologies which is much due to the role of external and hereditary factors. The problems of the peculiarities of clinical manifestation of endometrial cancer depending on aggravation of hereditary oncopathology are still unknown. The objective of the present research is the comparative study of clinical development and biological peculiarities of tumors in patients with aggravating and non-aggravating hereditary factors in respect of oncopathology. We have analyzed the reproductive and menstrual functions, biological peculiarities of tumor and the data of clinical and genealogical anamnesis of 482 patients with endometrial cancer. As a result of this research two groups of patients have been singled out. The first group of patients totals 261 patients (54.14%) with the absence of oncopathology in hereditary factors. The second group included 221 patients (45.86%) with endometrial cancer, whose relatives were subjected to have malignant tumors in the families. The data analysis has shown that endometrial cancer patients with the presence of oncopathology in heredity have differences in the clinical development of the oncotic process. These patients are characterized by earlier or later beginning of menstruations, early or late menopause; moderate or low level of differentiation of cancer, deeper invasion into miometry is observed more often. It is reflected in the decreasing values of five-year survival. All this proves that the peculiarities of heredity and aggravation in oncopathology in particular, modify the clinical peculiarities of the neoplastic process.

P9

Proband-mediated information dissemination does not meet the expressed wishes in families with a BRCA1/2 gene mutation

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Background: Genetic counseling for hereditary breast and/or ovarian cancer (HBOC) is usually based on a protocol of non-directive counseling from the international guidelines used for the Huntington families. When a BRCA1/2 gene mutation is found in a family, the possibility of predictive counseling and testing is also offered to the other family members, but only through informing the proband. We have examined the efficiency of information transfer from the proband to the other relatives, and we compared the level of transferred information to the needs in these families.

Methods: Fourteen families (with a BRCA1/2 mutation) with 107 subjects participated in the study. Subjects were eligible for participating if they were first-degree relatives of an affected person with breast cancer, ovarian cancer, or another primary cancer, or if they were first-degree relatives of a known or probable mutation carrier. Data were collected with semi-structured interviews.

Results: This study clearly reveals that the transfer of information from probands to their relatives is highly defective. In contrast and surprisingly, almost all participating relatives (97,8%) wanted to be informed about the various aspects concerning HBOC, and even wanted to have a predictive genetic test (96,6%).

Conclusion: The results of this study lead to the conclusion that the current practice of proband-mediated information dissemination is inefficient and does not meet the needs expressed by members of high risk cancer families for whom preventive measures (such as clinical surveillance, prophylactic oophorectomy and prophylactic mastectomy) are available. We therefore propose to inform relevant relatives with an informative letter, without revealing personal genetic test results.

P10

Genome instability of endometrial and colorectal cancer patients

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Aim: To investigate the changes of chromosomal instability in patients with endometrial (EC) and colorectal cancer (CC).

Materials and methods: It was performed the cytogenetic analysis of peripheral blood lymphocytes (PBL) from patients with EC and CC and healthy womens: the characteristics of chromosomal aberrations and fragile sites, heterochromatic regions of chromosome 1,9 and 16, nucleolus organizer regions (NOR's) activity and level of acrocentric chromosome association.

Results: It was shown the reliable increased frequency of PBL chromosome aberrations (per 100 cells) of EC patients (4,8+0,8) and CC patients (5,2+0.5) compared with healthy donors (0.7+0.2) and increased quantity of common and rare fragile sites. C-band variability (including extremal variants and polymorphism of homologues) in EC patients was differed from that in control. The number of active nucleolus organizer regions and the frequency of 13 and 21 acrocentric chromosome association was higher in patients with EC compared with that in control. Cytogenetic markers in PBL are correlated with histopathology grade and were more expressed in patients from families with oncopathology aggregation.

Conclusion: Cytogenetic research revealed the increasing chromosomal aberrations and fragile sites, variable changes of C-heterochromatin and increasing of NOR's activity that reflect of genome instability in EC and CC patients and may be of addition criterion of early diagnostic of CC and EC.