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EXPERIENCE IN INTRODUCING MEDICAL-GENETIC COUNSELING INTO EARLY DIAGNOSIS PROGRAMS FOR PRE-CANCER AND CANCER OF THE FEMALE REPRODUCTIVE SYSTEM

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SUMMARY
Hereditary ovarian cancer is one of the most common genetic pathologies. Medical genetic counseling for patients with hereditary forms of cancer and their family members is an integral part of providing oncological care, as it allows for the development of a set of diagnostic, preventive and therapeutic measures aimed at monitoring healthy individuals and creating personalized approaches to treating patients.
KEY WORDS: *familial forms of cancer, hereditary breast cancer and ovarian cancer, BRCA 1/2 genes, medical genetic counseling, prevention*

RELEVANCE

Ovarian cancer ranks third in the structure of cancer incidence of the reproductive system in women. According to the International Agency for Research on Cancer (IARC), more than 165 thousand new cases of ovarian cancer and more than 101,000 deaths from the progression of this pathology are registered annually in the world[1,5]. The individual risk of ovarian cancer is approximately 1.6%. The peak incidence is observed between 60-70 years and is approximately 65 cases per 100,000 female population per year, while before 40 years of age this figure is almost 5 times lower (Ozols RF et al ., 2005). Average incidence rates, taking into account women of all ages, slightly exceed 10 cases of ovarian cancer per 100,000 women per year (in Northern Europe - 12.7; in Western Europe - 11.6; in North America - 11.2; in Eastern Europe - 10.0). The lowest incidence rates of ovarian cancer are observed in the countries of Asia and North Africa (Daly M.V., 2003).

In most industrialized countries of the world, ovarian cancer has the highest mortality rates among tumors of the female reproductive system, which is associated with late diagnosis of the disease. The mortality rate of patients with ovarian cancer within a year after diagnosis is about 35%. According to aggregate data from population-based cancer registries in European countries, the three-year survival rate of patients with ovarian cancer is only slightly higher than 40%. Over the past decade, 5-year survival in Europe has increased by 3% (from 32 to 35%), and in the United States by 4% (from 36 to 40%). This positive trend is explained not only by improved diagnosis, but also by the use of effective chemotherapy in the treatment of disseminated forms of ovarian cancer[6]. The age of patients is not only a statistical, but also a prognostic parameter: the 5-year survival rate of patients who fell ill at the age of 15-29 years is 77.8%, 30-39 years - 71.1%, 40-49 years -57.4 %, 50-59 years old - 47.5%, 60-69 years old - 41.5%; at 70-79 years old - 34.1% [Aksel E.M., Kozachenko V.P., 2001; Black RJ et al ., 1997; Heintz APM et ai ., 2000].

Among all malignant neoplasms in Russian women, ovarian cancer ranks seventh in frequency and accounts for 4-6% of all cancer incidence. Every year there are 11,000 new cases of ovarian cancer in the country, and the number of patients under observation is approaching 80,000[7]. The average age of those affected is 59 years, the average age of those who died from ovarian cancer is 64 years. The peak incidence occurs at the age of 65-69 years, that is, a predominantly unorganized, socially unprotected part of the population that needs the implementation of affordable but highly effective programs for the early detection and treatment of this disease. There is an annual increase in the incidence of ovarian cancer, reaching 4% per year. In the structure of mortality from oncogynecological diseases, ovarian cancer accounts for 49-56% (Davydov M.I., Aksel E.M., 2006).

Prevention and early diagnosis of malignant neoplasms of the female reproductive system is the most important socio-biological problem of modern oncology. The role of hereditary factors in the tendency to develop cancer in a number of localizations has been proven. Medical and genetic counseling was carried out on healthy women, including families with familial cancer syndrome, for early detection of cancer pathology. A clinical and genealogical questionnaire has been developed for self-completion by women who contacted a gynecologist for a medical examination during 2007-2010. Medical and genetic counseling was carried out on 663 patients [9,10] .

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MATERIALS AND METHODS OF RESEARCH

According to the developed questionnaires, out of 663 women, 63 (9.95%) were found to have pedigrees that, according to the results of medical genetic counseling, correspond to familial cancer syndrome: type I syndrome - in 38 women (16-68 years old), type II syndrome - in 25 women (23-66 years old). Among them, women of reproductive age (up to 40 years) predominated, the number of which did not depend on the type of familial cancer syndrome and amounted to 20 (52.6%) and 14 (56%), types I and II, respectively.

RESULTS

According to the questionnaires, the relatives of these 63 women had cancer in the past: parents (mother and/or father) - 30, brothers and sisters - 6, grandparents (maternal or paternal) - 55, aunts and uncles - 23. Relatives had malignant neoplasms of various localizations: cancer of the rectum and colon (16), stomach (18), pancreas (3), uterine body (10), ovary (8), breast (21), lungs (12), prostate (7), kidney (3), other localizations (13), lymphoproliferative diseases (6) After in-depth clinical, instrumental and laboratory examinations, 17 (26.98%) of 63 women were diagnosed with oncological diseases at the initial stages of the process (body cancer uterus – 2, cervical cancer – 3, breast cancer – 6, rectal cancer – 2, thyroid cancer – 2, lymphogranulomatosis – 2), and 7 patients (11.1 %) were diagnosed with precancerous gynecological pathology (CIN III , atypical endometrial hyperplasia, adenomyosis of the uterine body, ovarian cysts). All patients underwent special and surgical treatment.

CONCLUSIONS

The introduction of medical and genetic counseling into gynecological practice is one of the effective measures aimed at early diagnosis of precancer and cancer of the female reproductive system. As part of medical genetic counseling, people of reproductive age should be informed about the possibility of genetic testing for the presence of mutations in the BRCA1, BRCA2, etc. genes. The best tactic today is a combination of transvaginal ultrasound examination and determination of the level of tumor-associated antigen CA-125 with periodicity carriers of mutations in the BRCA1 and BRCA21 genes once every 6 months from the age of 25 [26].

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