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# The awareness of the citizens of Swietokrzyskie Voivodeship on the genetic predisposition on cancer

Świadomość mieszkańców województwa świetokrzyskiego na temat genetycznych predyspozycji do zachorowania na nowotwory

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Wstęp: Choroby nowotworowe są istotnym problemem współczesnego świata, stanowią bowiem czwartą co do częstości przyczyne zgonów na świecie a drugą w krajach rozwiniętych. W zapoczątkowaniu rozwoju nowotworu istotną rolę odgrywają czynniki genetyczne. W ciągu ostatnich lat nastąpił ogromny rozkwit genetyki molekularnej i badań laboratoryjnych, umożliwiających wykrycie nowotworu w fazie mutacji.

Cel pracy: Celem pracy jest ocena stanu wiedzy mieszkańców województwa świętokrzyskiego na temat genetycznych predyspozycji do zachorowania na nowotwory.

Materiał i metody: Badaniami objęto 93 osoby zamieszkałe na terenie województwa świętokrzyskiego w wieku 18-75 lat. Metodę badawczą stanowił autorski kwestionariusz zawierający pytania sprawdzające wiedzę na temat genetycznego dziedziczenia nowotworów.

Wyniki: Wśród respondentów najliczniejszą grupę stanowiły kobiety 61%. Średnia wieku mężczyzn wynosiła 48 lat, kobiet 33 lata. Większość ankietowanych (60%) mieszkała w mieście. Poziom wykształcenia badanych był zróżnicowany. Mimo, iż sami badani mają nowotwór (28% kobiet i 53% mężczyzn) i w najbliższej rodzinie ktoś choruje lub chorował na chorobę nowotworową (49% wśród rodziny kobiet biorących udział w badaniu i 64% wśród mężczyzn biorących udział w badaniu) nie mają wiedzy na temat genetycznych predyspozycji do dziedziczenia nowotworów.

Wnioski: Świadomość badanych na temat genetycznych predyspozycji do dziedziczenia nowotworów i możliwości wykorzystania testów genetycznych w kierunku wykrycia choroby nowotworowej jest niewystarczająca. Z uwagi na fakt, że około 30% wszystkich nowotworów ma genetyczne uwarunkowania, dostarczenie kompetentnej i uporządkowanej wiedzy wydaje się istotnym elementem edukacji zdrowotnej bowiem wczesne wykrycie zwiększa szansę wyleczenia.

# Słowa kluczowe: nowotwory, predyspozycje genetyczne, dziedziczenie nowotworów.

Introduction: Cancers are the major concern of the modern world, they are in fact the fourth most common cause of death in the world and second in developed countries. The root cause of tumour development lies in genetics. In the last few years a significant rise in the molecular genetics and laboratory studies, that allow to detect cancer phase mutation.

Aim: The purpose of this dissertation is to evaluate the knowledge of genetic predispositions to cancer among the citizens of Swietokrzyskie Voivodeship.

Material and methodology: The research has been carried out on 93 people living in the Swietokrzyskie Voivodeship aged between 18-75. The chosen measurement method was a survey testing the knowledge of genetic predispositions to cancer.

Results: Among the questioned the majority were women (61%). The average age was 48 among men and 33 among women. The majority of questioned (60%) were living in a city.

The level of education was diverse. Even tough, some of the questioned are or were suffering from cancer (28% of women and 53% of men) or someone in their close relation is or has suffered from cancer (49% in case of women and 64% among men) they do not have the knowledge of the genetic predispositions to cancer.

Conclusions: The awareness of questioned about the genetic predispositions to cancer inheritance or the possibility of using genetic tests to detect the cancer is insufficient.

Regarding to the fact that approximately 30% of cancers have genetic conditions, providing complete and structured knowledge seems to be an important element of health education, because early detection increases the chance of recovering.

### Keywords: cancers, genetic predisposition, hereditary cancer.

Cancer is the second, after the cardiovascular diseases, leading cause of death in Poland and its numbers are still increasing [1]. The World Health Organisation warns against this significant rise in cancer incidence rates [2]. In Poland cancer is the most common cause of premature mortality (before the age of 65) [3]. The National Cancer Registry informs that only in 2001 there have been 144 thousand cases of cancer detected, out of which 92 thousand were fatal and 29,2 thousand concerned people in working age [23].

There are many risk factors that may increase the risk of developing cancer. Among them, the most significant are genetic factors [4]. Over the last several years, there has been a significant progress in molecular genetics and laboratory research that allow detecting the disease in its early stages.

# 1. Cancer epidemiology

In Poland the most common types of cancer among men are: lung 20,3%, prostate 14,4%, large intestine 11% [3]. In 2010, lung cancer was the first cancer-related cause of

death among men - 31% of deaths, followed by large intestine cancer - 11%, and prostate cancer -8% [23].

Predominating among women is breast cancer 23%, large intestine cancer 10%, lung cancer 9%, ovarian cancer 5% and cervical cancer 4%. In the case of women, the first cancer-related cause of death was lung cancer - 15% of deaths, followed by breast cancer - 13%, large intestine cancer - 12%, and ovarian cancer - 6%.

In the Swietokrzyskie Voivodeship, the most common types of malignant cancer are lung 21%; large intestine 13%, prostate 11%. The most common cause of death was: lung cancer 33,5%, large intestine cancer 17,2% and prostate cancer 8,6%.

Among women the most common types of cancer were: breast 17,1%, large intestine 10%, lung 6,7%, ovarian 5,1% and the most common cause of death was: breast cancer 14%, large intestine cancer 12,2%, lung cancer 11,9%, ovarian cancer 6% [17].

# 2. Genetics

Cancer initiation is a complex multifactorial and multistage process. Cancer is caused by changes or mutations. The risk of falling ill with hereditary cancer is dependent on the level of gene changes within cells. It is measured by the percentage of illnesses among the carriers of a specific mutation [6].

### 3. Diagnostics

There is no single examination to detect cancer. With some types of cancer e.g. breast, ovarian or large intestine cancer the most significant forms of diagnosis are regular and free of charge screenings. In other cases, it is important to pay attention to some irregularities and symptoms. Furthermore, people with high risk of hereditary cancer and carriers of specific mutations should fall within the scope of a special care programme.

### 3.1. Breast cancer

Women whose relatives were diagnosed with breast or vaginal cancer should start appropriate examination at an early age, according to the recommendations of a gynaecologist. They should be tested as below:

- ✓ manual palpation by a patient, from the age of 18 and then once a month after menstruation
- ✓ manual palpation by a physician from the age of 20 to 30, every six months [7]
- $\checkmark$  mammography, from the age of 30, annually

- ✓ ultrasound examination, from the age of 25, annually (six months after mammography)
- ✓ MR, below 30 years of age
- ✓ primary prevention, breast amputation from the age of 40 to 50, that reduces the risk of breast cancer by 90-95% [8].
- 3.2. Ovarian cancer

Most cases of ovarian cancer are diagnosed at a late stage of development when the patient has low chances of survival. Among the carriers of BRCA1 gene, that is among women with a higher risk factor, control examinations are carried out more frequently. Also in some cases, the primary prevention in the form of bilateral salpingo-oophorectomy is used. This procedure decreases the risk of cancer by 80-95%. However, the procedure must be recommended with a great caution, as many clinical types of research state that if the procedure is made before the menopause it may have a negative impact on the overall health of a patient [11]. The most significant examinations for ovarian cancer are: vaginal ultrasound examination every six months (from the age of 35), abdominal ultrasound examination, gynaecological examination and the detection of the CA 125 antigen. The latter should be done annually, usually six months after ultrasound examination [4,7]. Another type of examination is ROMA test. It consists out of three elements: the assessment of the risk of epithelial ovarian cancer and testing for the HE4 and CA125 markers [12]. The test stratifies cases into two groups: low- or high-risk for ovarian malignancy [13].

### 3.3. Large intestine cancer

Approximately 10-20% of all cases of large intestine cancer is hereditary [5]. In Lynch Syndrome, an inherited disorder that increases the risk of many types of cancer, colonoscopy should be done every two years. For women annual vaginal examination and vaginal ultrasound examinations are recommended, and in some cases, it is also necessary to remove the large intestine.

# 3.4. Prostate cancer

Approximately 60% of all cases of prostate cancer is hereditary [14]. Prostate cancer may show no signs or symptoms in its early stages. The most common diagnostic methods include:

- $\checkmark$  the measurement of the concentration of PSA marker
- $\checkmark$  rectal examination of prostate
- $\checkmark$  transrectal ultrasound examination

# ✓ prostate biopsy

For people with high risk of cancer, periodic health examinations should be made, starting at age 45. Examinations should be done in short intervals [15].

# 4. Purpose

In the last few years, there has been a significant increase in mortality rates caused by cancer. The purpose of this dissertation is to evaluate the knowledge of genetic predispositions to cancer among the citizens of Świętokrzyskie Voivodship.

# 5. Material and methodology:

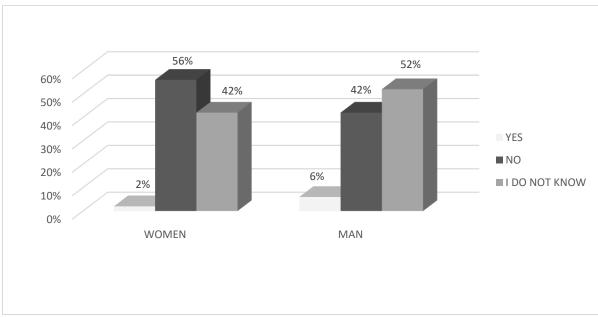
The research has been carried out on 93 people living in the Świętokrzyskie Voivodship aged between 18-75. The chosen measurement method was a self-made survey testing the knowledge of genetic predispositions to cancer.

6. Findings

Among the examined the largest group were women (61%) with the average age of 48. The average age of examined men was 33. The majority of examined are living in the city (60%) and had completed at least secondary education (36%).

At the level of 0,01 of statistical significance, the hypothesis that the variables are individual issues was rejected and it was stated that there is a difference in the knowledge on the researched matter between both sexes. The below diagrams present the results of the survey.

Figure 1. The opinion of the respondents on the claim that every carrier of the defective gene will develop cancer.



#### Source: own study based on research results.

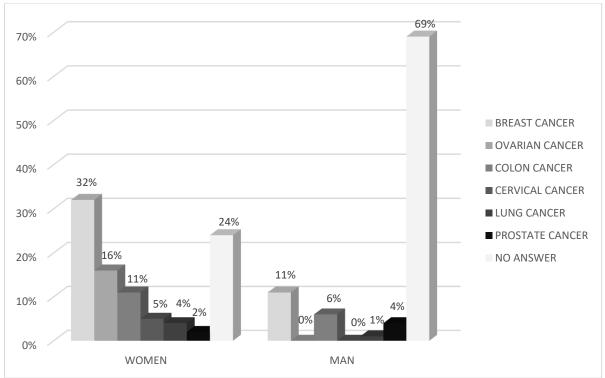
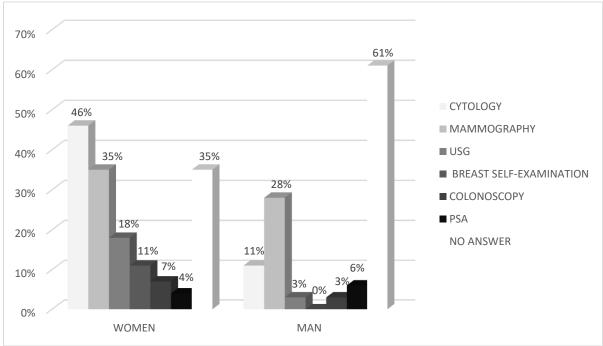


Figure 2. Knowledge of the respondents about hereditary cancer.

Figure 3. The opinion of the respondents on the claim prophylactic surveys that should be done while having predispositions to developing cancer.



Source: own study based on research results.

Source: own study based on research results.

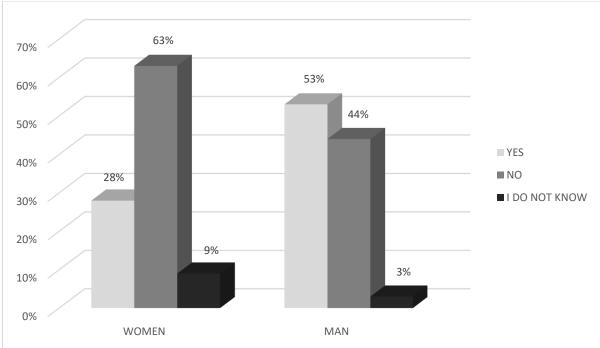
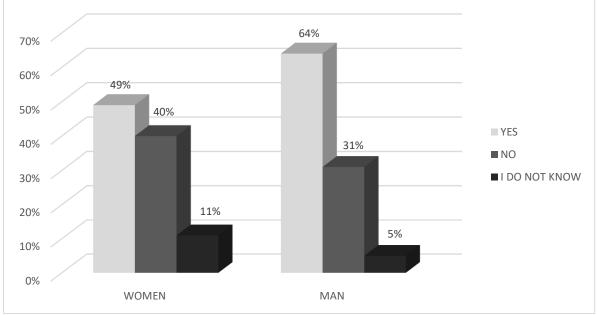


Figure 4. Number of the respondents having cancer.

Figure 5. Number of the respondents that have in the closest family someone that has or had cancer.



Source: own study based on research results.

Source: own study based on research results.

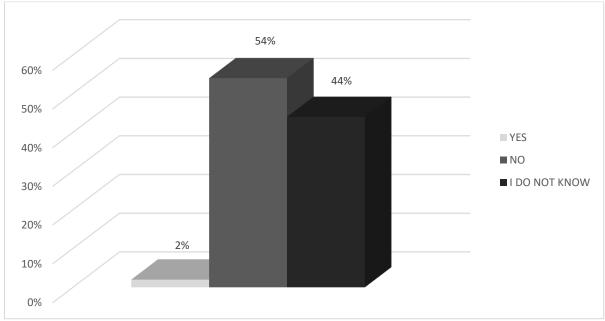
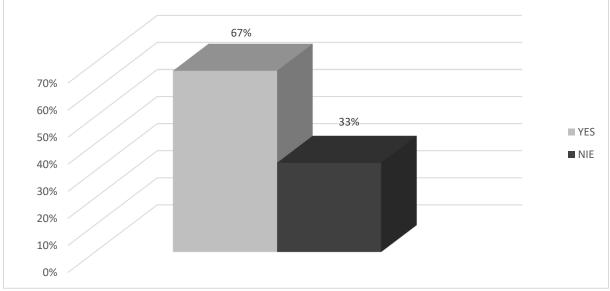


Figure 6. The opinion of the respondents on the claim that finding of a mutation means the occurrence of cancer.

Figure 7. Knowledge of the respondents about cases of breast amputations that were made because there was a possibility of inheritance risk of developing breast cancer.



Source: own study based on research results.

Source: own study based on research results.

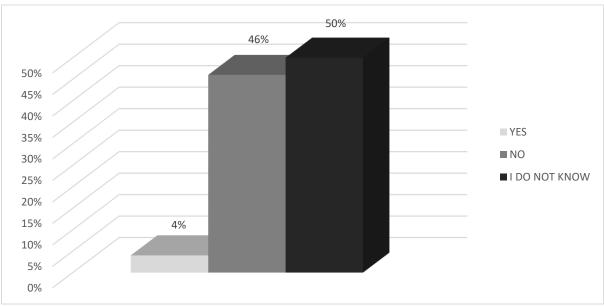
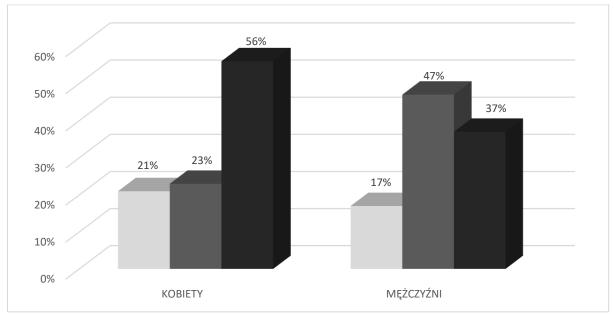


Figure 8. The opinion of the respondents on the claim that one genetic test could diagnose predisposition to all types of cancer.

Source: own study based on research results.

Figure 9. The opinion of the respondents on the claim of the number of genetic tests that should be made in direction of inheritance of one type of cancer, for example the colon cancer in a lifetime.



Source: own study based on research results.

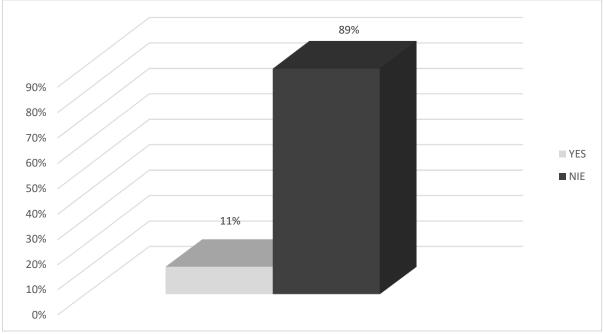
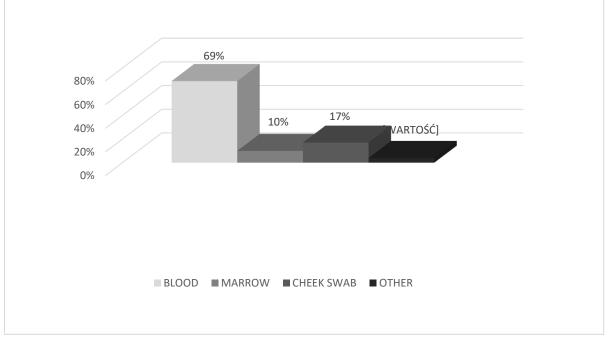


Figure 10. Knowledge of the respondents about look of genetic test in direction of, for example breast cancer.

Figure 11. The opinion of the respondents on the claim of the body fluids that have to be collected to genetic test in direction of cancer disease.



Source: own study based on research results.

Source: own study based on research results.

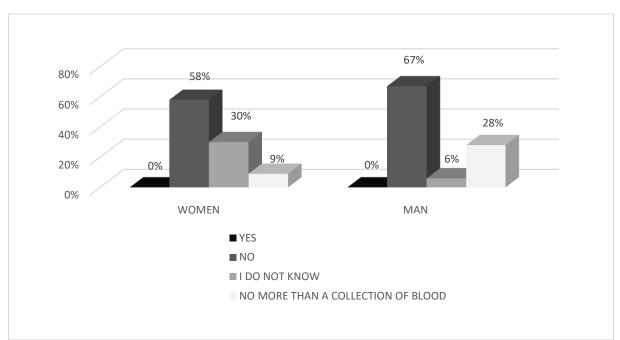


Figure 12. The opinion of the respondents on the claim that the genetic test is dangerous for health.

Source: own study based on research results.

### 7. Discussion

Cancer is one of the most common diseases and it proved to be a serious health, social and economic problem. In the female population (45-64 years of age) the percentage of mortality due to cancer rapidly increases. In the male population, malignant cancer remains the reason of death in 29% cases [16]. In the European Union in 2015, there have been 1 250 000 deaths caused by cancer, an increase of 11% in comparison to 2000 [18].

Approximately 30% cases of cancer are a result of the high, genetic predisposition [24]. Epidemiological studies show that there is a certain population of people whose characteristic feature is the increased incidence of cancer, and these patients usually get sick at a younger age. It is important that being the carrier of a specific gene mutation means that there is a higher possibility of developing a particular type of cancer. What is inherited is only the predisposition to the disease, not the disease itself. [6] Genetic research has led to findings that some people from birth are carriers of the defective gene. A classic example is the gene BRCA1, which damage is associated with an increased risk of incidence of breast cancer. The predisposition to cancer is autosomal dominant [24].

The studies show that the awareness of the examined population in the field of genetic inheritance of cancer is low. It is widely known that genetic test is performed once in a

lifetime, and its result does not change. Currently, there is no way to perform a single genetic test to assess the risk for all types of cancer. The studies also show that half of the respondents does not know that one genetic test cannot diagnose predisposition to all types of cancer (shown in Figure 8). 56% of women and 37% men (shown in Figure 9) said that the genetic test is performed only once in a lifetime. The vast majority (89%) does not know how a genetic test for the breast cancer looks like (shown in Figure 10). 69% of respondents mentioned that a genetic test involves collecting blood samples (presented in Figure 11). 58% of women and 67% of men claims that genetic tests are not dangerous for health (presented in Figure 12). The positive result of the test does not mean that a person will fall ill with the disease. It only means that the person is more susceptible to it. 54% of respondents indicated that the mutation is not equivalent to having cancer, of cancer, the rest of the respondents did not know the answer to this question (shown in Figure 6). It is stated that there are 100 thousand women with a suspected BRCA1 gene mutation living in Poland [19], out of which only 7-8 thousand cases are diagnosed.

Not so long ago, a Hollywood star Angelina Jolie has sparked a discussion about cancer prophylaxis as she revealed her double mastectomy. The news reverberated in global media, Poland included. The majority of respondents (67%) indicated that they know cases of women, who because of the possibility of hereditary risk of breast cancer, have decided to take the same precautions (shown in Figure 7). Some of them have written Angelina Jolie's name as an example. 56% of women and 42% of men believe that not every carrier of the defective gene will develop cancer (shown in Figure 1). And they are right in their assumptions because there are genes defects that are associated with a very high increase in the risk and genes with medium or even low-risk growth. An important issue is not only genetics but also the environment in which we live.

If a person with a defective gene, for example, MSH2 which predisposes to colon cancer, does not care for his or hers health through the use of proper diet, it increases the risk of falling ill. However, if the same person applies to basic medical principles, there is a chance that he or she will never have cancer. Recent studies show that self-knowledge of hereditary cancer is not high. Breast cancer was the most common response and has been selected by <sup>1</sup>/<sub>3</sub> of the women. 16% of respondents indicated ovarian cancer, 11% of colon carcinoma and 5%, 4% and 2% of cervical cancer, lung and prostate tumours. 24% of women do not know any hereditary cancer. Lorenc, Pop and Boychuk stated in their studies that 78% of women said that breast cancer can be a genetic disease [20]. Among men, this knowledge is sparse, the

most common answer was breast cancer (11%). 69% of respondents did not know the answer to this question (shown in Figure 2). Self-carried research indicates that women respondents know such prophylactic examinations as: cytology (46%), mammography (35%), ultrasound (18%), breast self-examination (11%) and colonoscopy (7%) (shown in Figure 3).

Cichońska and co. state that <sup>1</sup>/<sub>4</sub> of female respondents had heard about the prevention program for early detection of cervical cancer and 20% of breast cancers. It is worth mentioning that none of the respondents in our study indicated magnetic resonance imaging. Lubiński pointed out that breast cancer in women with defective BRCA1 gene can be detected by using magnetic resonance, and the probability of detecting cancer without metastases in armpits reaches 77% in case of resonance and only 20% while using mammography [22].

### 8. Results

Even tough, the people taking part in the survey have cancer (28% of women and 53% of men) or someone in their close relation has or had cancer (49% among the women and 64% among men (illustrated on the graph 4 and 5) they have small knowledge on the genetic predispositions to cancer.

The awareness of respondents about the genetic predisposition to cancer and the possibility of using genetic testing to detect the cancer is insufficient. Due to the fact that about 30% of all cancer is conditioned genetically, a competent and organised knowledge seems to be an essential part of health education, because early detection increases the chances of a successful cure.

### 9. Bibliography

Didkowska J. Epidemiologia nowotworów złośliwych w Polsce. W: Meder J. (red.).
Podstawy onkologii klinicznej. Wydawnictwo Centrum Medyczne Kształcenia
Podyplomowego w Warszawie, Warszawa 2011; 5-16.

 Didkowska J. Prognozy rozwoju chorób nowotworowych w Polsce. W: Kwiatkowska L. (red.). Zachorowalność i umieralność na nowotwory a sytuacja demograficzna Polski. Rządowa Rada Ludnościowa, Warszawa 2014; 147-163,

3. Zatoński W, Sulkowska U, Przewoźniak K, Zatoński M. Epidemiologia nowotworów złośliwych w Polsce. W: Kwiatkowska L. (red.). Zachorowalność i umieralność na

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nowotwory a sytuacja demograficzna Polski. Rządowa Rada Ludnościowa, Warszawa 2014; 30-49.

4. Kulig A, Smolarz B. Badanie genetyczne apoptozy i mutacji genu BRCA1 u kobiet obciążonych dziedzicznie rakiem piersi. Przegląd Menopauzalny 2004; 5: 19-23.

5. Lovett E. Family studies in cancer of the colon and rectum. British Journal of Surgery 1976; 63 (1) : 13-18.

6. <u>http://onkologia.org.pl/nowotwory-genetyczne</u> (dostęp: 2016.05.03).

7. Lubiński J, Górski B, Kurzawski G, Jakubowska A, Dębniak T, Cybulski C. "i wsp." Genetyka we wczesnej diagnostyce nowotworów. Współczesna Onkologia 2000; 4; 5: 186-189.

8. Badora A, Kaleta B. Mnogie nowotwory pierwotne u kobiet nosicielek mutacji genu BRCA1 – dwa przypadki kliniczne. Ginekologia Polska 2013; 84(10):892-896.

9. Markowska J, Mądry R. Stan obecny w leczeniu nowotworów złośliwych narządów rodnych u kobiet. W: Kwiatkowska L. (red.). Zachorowalność i umieralność na nowotwory a sytuacja demograficzna Polski. Rządowa Rada Ludnościowa, Warszawa 2014; 103-116.

10. Moszyński R, Szpurek D, Pawlak M, Englert-Golon M, Sajdak S. Wpływ typu histopatologicznego guza jajnika na wartości prognostyczne ultrasonograficznego indeksu morfologicznego. Ginekologia Polska 2006; 77: 516-522.

11. Berek JS, Chalas E, Edelson M, Moore D, Burke W, Cliby W. "i wsp." Prophylactic and Risk-Reducing Bilateral Salpingo-oophorectomy: Recommendations Based on Risk of Ovarian Cancer. Obstetrics & Gynecology 2010; 116(3):733-743.

12. Gumółka I. Przydatność oznaczeń surowiczego stężenia CA 125 i HE4 w diagnostyce raka jajnika. Annales Academiae Medicae Silesiensis 2015; 69: 138–149.

13. Li A. Nowe markery biologiczne w raku jajnika. Przydatność OVA1 i ROMA w ustalaniu rozpoznania. Ginekologia po dyplomie 2012; 14-19.

14. Hjelmborg JB, Scheike T, Holst K. Skytthe A, Penney K, Graff R. "i wsp.". The heritability of prostate cancer in the Nordic Twin Study of Cancer. Cancer Epidemiol Biomarkers Prev. 2014; 23(11):2303-10.

15. Bratt O. Hereditary prostate cancer: clinical aspects. The Journal of Urology 2002; 168(3):906-13.

16.http://www.rynekzdrowia.pl/Uslugi-medyczne/Nowotwory-zlosliwe-w-Polsceznamynajnowsze-dane-epidemiologiczne (dostęp: 2016.05.03).

17. Góźdź S, Karpacz T, Stępień D. Nowotwory złośliwe w województwie świętokrzyskim w 2010 roku. Ministerstwo Zdrowia, Kielce 2012.

18. Quinn M.J, d'Onofrio A, Moller B, Black R, Martinez-Garcia C, Moller H. "i wsp.". Cancer mortality trends in the EU and acceding countries up to 2015. Annals of Oncology 2003; 14: 1148–1152.

19. Lubiński J. Przyszłość onkologii należy do genetyki. Przegląd Urologiczny 2008; 3 (49):22.

20. Lorenc A, Pop T, Boychuk T. Wiedza kobiet po 40 roku życia o czynnikach ryzyka i profilaktyce raka piersi. YOUNG SPORT SCIENCE OF UKRAINE 2012; 4 : 59-65

21. Cichońska M, Borek M, Krawczyk W, Maciąg D. Wiedza kobiet w zakresie zapobiegania nowotworom piersi i raka szyjki macicy.

22. Gronwald J, Byrski T, Huzarski T, Oszurek O, Menkiszak J, Rzepka-Górska I, Lubiński J. Genetyka kliniczna raka piersi i jajnika. Postępy Nauk Medycznych 2008; 7 : 446-455.

23. Didkowska J, Wojciechowska U, Zatoński W. Krajowy Rejestr Nowotworów. Nowotwory złośliwe w Polsce w 2011 roku. Warszawa 2013; 14-15.

24. Dębniak T, Lubiński J. Zasady dziedziczenia predyspozycji do nowotworów. Postępy Nauk Medycznych 2008; 7: 427-430.