

Psychosocial context of the decision-making process and the consequences of preventive procedures in a patient with BRCA1 genetic mutation

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Summary

Background: Patients with a genetic mutation that increases the risk of cancer are often offered preventive surgery – mastectomy and adnexectomy. The objective of this case report is to present the psychosocial context of the decision to perform prophylactic breast and ovarian removal surgery and the consequences of the indicated procedures on the example of a patient with the BRCA1 genetic mutation, to adapt educational activities and support for patients.

Case presentation: Patient with BRCA1 genetic mutation, without cancer diagnosis – now and in the past. Cancers occurred in the family. Due to high cancer risk, in 2018 performed two preventive procedures – mastectomy and adnexectomy. The decision to carry out preventive surgical procedures made two years after receiving a recommendation from a geneticist, which was due to the family situation and the patient's responsibilities. The decision was associated with a special mental state which means that the risk of cancer may contribute to the patient's feeling of fear or recurring concerns about their health and requires meeting specific support needs.

Conclusions: Patient needs covered educational or emotional and social aspects, but also support provided by qualified specialists and medical staff. The patient's experience described indicates that specialist psychological help is needed not only at the stage preceding the decision to carry out preventive surgical procedures, but also after their execution, because it is associated with changes that affect the patient's daily life and, consequently, her psychophysical condition.

BRCA1, prophylactic mastectomy, prophylactic adnexectomy

INTRODUCTION

The development of cancer diagnostics as well as genetic counseling and public awareness and the popularization of knowledge about prevention increase the number of women who are choos-

ing to perform a genetic test for the presence of BRCA1 (breast cancer1) and BRCA2 (breast cancer 2) gene mutations. These genes increase the risk of developing breast or ovarian cancer. According to data obtained at the International Hereditary Cancer Center in Szczecin for the Polish population, the risk of developing cancer is about 66% for breast cancer and 44% for ovarian cancer, respectively [1]. Current observations indicate that cancer risk depends on the type of mutation and its location in the gene [2]. Pro-

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phylactic removal of the breast and reproductive organs is associated with a reduction in the risk of cancer, but not all patients decide to perform these procedures [3]

The decision to carry out preventive treatment by carriers of the BRCA1 / 2 mutation can be conditioned by family circumstances, such as having children or planning offspring, and is also related to the age of women. Long-term satisfaction assessment of patients who underwent preventive procedures indicates positive results in terms of reducing emotional concerns related to the development of breast cancer and overall favorable results in the areas of psychological and social functioning [4]. The results obtained so far indicate satisfaction of patients who underwent prophylactic mastectomy in a time perspective of 6 to 30 months after the surgery [5, 6]. On the other hand, literature covering psychosocial issues in the decision-making process and psychosocial consequences experienced by the patients after performing preventive procedures is significantly limited.

In the Polish literature, according to the author's knowledge, the psychosocial context of the decision-making process and the emotional consequences of preventive procedures performed by patients with the BRCA1 and / or BRCA2 genetic mutation have not yet been described. For this reason, it is worth presenting the case of a patient with a genetic mutation who underwent preventive procedures, taking into account the motivation regarding the moment when she decided to perform the surgery and her individual perspective regarding the changes that were associated with preventive mastectomy and adnexectomy.

CASE DESCRIPTION

A 42-year-old patient, married for 16 years, having two daughters – aged 12 and 16, living in a rural area, with higher education and professionally active, was diagnosed in 2015 as a carrier of the BRCA1 genetic mutation. At the time of diagnosis, the patient did not have a personal cancer history, she was healthy. In the patient's family, the sister, aunt and cousin received a diagnosis of breast cancer. In 2016, the doctor offered the patient to undergo surgery to reduce

the risk of cancer. In 2018, she underwent preventive mastectomy with reconstruction and preventive adnexectomy. The decision to perform the procedures was preceded by a 2-year postponement period, conditioned by the family situation – mainly caring for a sick father, who required disposition and physical fitness.

The patient's account shows that the decision to perform preventive procedures was primarily determined by the parenting perspective. Due to the role of the mother in relation to two adolescent daughters, who, considering the family history of cancer, may also be carriers of a genetic mutation, the patient decided to take action to serve as an example worth following in the future. She also wanted to support her daughters more effectively and have practical knowledge regarding the course of surgery and convalescence, as well as the consequences of their performance.

The leading fears arising during the decision-making process, which the patient indicated, were connected with the fact that the operation took place in a private clinic, so in the event of complications there was no possibility of quick and direct transfer to a specialized department, e.g. an emergency department. The patient also pointed to her apprehension associated with the psychophysical condition of the doctor who on the day of the surgery worked in the operating theater from 8.00 am to 2.00 pm, "then consulted 5 patients and stood at the table again", proceeding to the surgery with the participation of the patient. In the area covering the consequences of the procedures, the patient indicated greater fears associated with performing preventive mastectomy than before adnexectomy. At the time, fear of "greater surgical intervention and longer surgery time" dominated. When deciding on the prophylactic removal of ovaries and appendages, the patient was primarily afraid of the effects of menopause, possible changes in appearance, i.e. weight gain and external signs of aging – graying hair, skin deterioration. In the patient's perspective, fear of experiences similar to those of her loved ones, above all her sister, who at the age of 51 performed an adnexectomy (after cancer diagnosis) and as a result "she gained over 15 kilos, turned gray, has depression" were seen.

The patient relied heavily on her husband's opinion when deciding to perform the preven-

tive procedures. The most difficult of the topics in the talks at that time were the long-term consequences of the procedures performed, which were considered by the patient – including primarily depression and loss of physical attractiveness. The patient declares that if she had not received her husband's approval then she would probably not have decided to undergo preventive surgery.

After performing the procedures, the patient does not feel any changes in the direct perception of her femininity. She indicates weight gain as the most severe of the negative effects of the treatments. Since the surgery, the patient has not done any sports that in the past helped her maintain a satisfactory figure. The decision to stop activity, such as running, was the result of a surgeon's suggestion that "shocks are very bad for the appearance of the breasts." In the patient's perspective, "an additional 8 kilograms more affect the sense of femininity than the mere fact of having the breast operated."

After performing the preventive procedures, the sexual relations of the patient with her husband "weakened". The leading reason, however, was not the mere removal of the breast, but above all a long recovery after surgery, pain, a decrease in the sense of femininity resulting from weight gain. Also, in the long run "a decrease in libido, noticed a year after the surgery". The patient's husband declares that he has not noticed any changes in the sexual relationship with his wife.

RESULTS

The decision-making process regarding the implementation of preventive procedures, the course of surgery and convalescence were associated with a specific psychological condition and the needs for support indicated by the patient. The accounts show that she primarily felt the lack of "system support", understood as social education and support in "lifting the burden of consciousness" after receiving a positive result of the genetic test. In the patient's perspective, the genetic counseling center that she used aroused her fear of a potential disease and was trying to convince her that the treatment was not reimbursed by the National Health Fund.

The patient claims that the support system either does not exist at all or is not comprehensive – one doctor focused only on the need for mastectomy, the other only on the need for adnexectomy. The patient used psychological consultation only once and it was a consultation necessary to receive a certificate of mental readiness to perform the procedure that the surgeon required before performing mastectomy. This meeting was not satisfactory for the patient because, according to the report, it did not provide her with support. Other forms of psychological assistance were not offered to the patient, although she now sees that it would be justified to use such help. The patient's experience shows that forms of psychological help should be offered to patients also, and maybe even above all, after performing the procedures, because they are associated with significant changes affecting everyday life and, as a consequence, also the psychophysical condition.

Experiences resulting from undergoing preventive procedures have changed the patient's lifestyle. She began to eat healthier meals and approach them with greater attention, avoiding alcohol, sun and stress. The patient is a member of a discussion group on a social networking site that brings together women with the BRCA1 and BRCA2 genetic mutation, where she is involved in supporting people who are considering preventive surgeries.

DISCUSSION

Taking into account the conclusions contained in the presented description, it seems reasonable to introduce a more extensive analysis of the needs that can be externalized to patients deciding to perform preventive mastectomy and adnexectomy.

The results of research conducted around the world show that improving the support system provided to patients can lead to an increase in their commitment, a greater sense of understanding and also help in making decisions regarding the selection of preventive actions. Farrelly and colleagues [7] described the process of implementing a program based on mutual assistance. The goal was to reduce the discomfort felt by carriers of the BRCA mutation through reg-

ular 4-month contact with a trained peer, also a carrier of the mutation who was a volunteer. Each of the parties, both supporting and supported, expressed high satisfaction from participating in the program. This shows the importance of both social and emotional support. This is particularly valuable when non-invasive risk reduction methods, such as lifestyle changes, offered to patients are insufficient [8]. At the same time, it is worth showing the positive aspects of preventive surgery in educational activities. It has been proven that these operations not only reduce cancer risk, but also reduce fear of cancer [9].

The decision to perform preventive surgery seems to be a complex process. This was shown, among others in research by Underhill and Crotsler [10], that the emotional and physical consequences of the decision (actual as well as potential, implied) are important for the decision-making process and that they require not one-time, but permanent and long-term support, also by the medical staff and healthcare professionals. This was also confirmed by the analysis of the case described in this article. In turn, studies by Puski and colleagues [11] showed that patients with the BRCA mutation, under 40 years old, and people without children showed difficulties in making decisions regarding cancer risk management. The patient described in this work also clearly emphasized that being a mother was important for making a decision about the operations. Sharlene Hesse-Biber and An Chen studies [12] have shown that women who felt guilty and afraid of being able to pass the BRCA gene on to their children were more likely to opt for risk-reducing surgery. Moreover, the sex of the child also mattered. Patients who had at least one daughter were more likely to perform surgery than women who had only sons.

The case described shows that genetic counseling is one of the key elements in the decision-making process regarding preventive measures. It is also often the only form of support that patients benefit from. This is confirmed by modern research report, showing that doctors are an integral part of the decision-making process. They are a source of support in risk management. Family members, other close relatives and other carriers also support patients, but their role is different from the roles of doctors – it is

more about emotional support [11]. However, it is worth remembering that, just like the described patient, also other carriers of BRCA mutations, when receiving the result, often are under the influence of strong emotions, which may not fully understand the information provided to them, especially if they contain too complicated medical terms. Lack of proper communication with the patient may contribute to difficulties in understanding complicated concepts [13].

McQuirter and colleagues [14] have shown that women who decide to perform preventive surgery experience the emotional key moment that makes the decision final. This means that the described decision-making process is not so much acting in accordance with specific procedures, but rather an intuitive and emotional process.

However, it's worth remembering that the decision to perform preventive surgery may be conditioned not only by the patient's internal emotional state, but also be dependent on other factors such as the economic situation [15], family history of cancer [16] or ethnicity [17].

Understanding what makes women decide to perform cancer risk reduction procedures, despite the lack of illness, seems to be the basis for implementing the most effective educational and preventive measures. Bearing in mind the complex needs of patients (technical, emotional, social, psychological support) in the future it would be worth considering the possibility of organizing a complex support system in Polish conditions that would meet the needs of patients and could at the same time facilitate the decision-making process regarding preventive mastectomy and adnexectomy.

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