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Commentary

Significance of BRCA genetic testing for preoperative breast cancer patients

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Abstract

Examining BRCA mutations in preoperative breast cancer patients is very important when selecting a surgical procedure. Although there are advantages and disadvantages associated with knowing about the presence of genetic mutations, including for the patient's family, there are many benefits for the patient. BRCA genetic testing should be recommended for patients who are strongly suspected of being positive for a BRCA mutation. However, it is up to the patient to decide whether or not to undergo a genetic test. Therefore, medical staff should provide accurate information and interact with the patient, but the patient should not be pressured to undergo testing.

Keywords: Breast cancer, BRCA mutation

Approximately 5%-10% of cases of breast cancer are hereditary and thought to carry germline mutations in genes involved in the development of breast cancer. This is the so-called hereditary breast cancer and ovarian cancer syndrome. The BRCA1 and BRCA2 genes are the most frequent causative genes of breast cancer [1,2]. Patients with pathological mutations in these genes have an increased risk of developing breast cancer, ovarian cancer, prostate cancer, and pancreatic cancer compared with the general population. By the age of 80 years, the prevalence of breast cancer is 72% for a BRCA1 mutation and 69% for a BRCA2 mutation, and the prevalence of ovarian cancer is 44% for a BRCA1 mutation and 17% for a BRCA2 mutation [3].

In the past, genetic testing and counseling for cancer was not covered by public medical insurance in Japan, and medical personnel were not fully aware of hereditary breast cancer and ovarian cancer syndrome. In addition, risk-reducing surgery was not commonly performed. Therefore, the current situation is that few medical institutions and specialists carry out testing for cancer-associated genes. However, it has become clear that many Japanese patients have hereditary breast cancer with mutations in BRCA1 or BRCA2. Given this situation, it has been difficult to measure the prevalence of mutations in BRCA1 and BRCA2 in Japan.

In recent years, the number of BRCA genetic tests being performed in Japan has increased dramatically due to increased awareness and availability of BRCA genetic tests as part of the treatment for recurrent breast cancer. These genetic tests were intended only for patients with recurrent breast cancer and could not be performed for preoperative patients. However, in Japan, BRCA genetic testing finally started to be covered by public medical insurance in April 2020. For those who have already developed breast cancer, genetic testing is indicated when there are two or more primary breast cancers or when there is a third-degree relative who has breast or ovarian cancer.

For BRCA mutation-positive patients, preoperative mutation-positive findings are critical for the choice of treatment and procedure. The approach for BRCA mutation-positive breast cancer patients is very different from that for general breast cancer patients who do not have a mutation. If a mutation is present, mastectomy is recommended for radical surgery, even if the breast can be preserved, and it has a great influence on the decision for the surgical procedure [4,5]. In addition, prophylactic resection of the contralateral breast is recommended for patients with breast cancer who have not yet developed contralateral breast cancer. This is also covered by public medical insurance in Japan. Thus, it must be understood that the treatment approach for BRCA mutation-positive breast cancer patients is very different from that of other patients.

We encountered a case of simultaneous bilateral breast cancer that was found to be BRCA mutation-positive preoperatively and treated by bilateral total subcutaneous mastectomy and breast reconstruction [6]. The surgical procedure was also changed significantly in our case.

The tumors on both sides were significantly reduced by preoperative chemotherapy, and breast-conserving surgery was possible. There was no lymph node metastasis, and we would usually perform bilateral partial breast resection and radiation therapy to the remaining breast. The patient had a notable family history and underwent a BRCA genetic test. As a result, she was diagnosed as BRCA1 mutation-positive and we changed her procedure to bilateral total subcutaneous mastectomy and breast reconstruction.

It is very important to check for mutations in BRCA genes before treatment, especially before surgery. A mastectomy on the affected side is recommended, and simultaneous reconstruction is an option. Furthermore, it is possible to perform preventive resection on the contralateral side at the same time. If breast-conserving surgery is performed, there may be local recurrence in the preserved breast. If the patient then undergoes genetic testing and is found to be positive for a BRCA mutation, the patient's feelings when it later turns out that a mastectomy was desirable are beyond speculation.

However, genetic testing cannot be enforced in all cases and it is not always necessary. It should be offered first to eligible patients who fulfill the criteria for genetic testing. The frequency of BRCA mutations is by no means high, but accurate information should be provided to patients who are to decide whether or not to undergo genetic testing.

At that time, care must be taken not to impose a genetic test on the patient for the convenience of the medical staff. Patients should not be forced or pressured to undergo testing. Our patient had undergone preoperative chemotherapy, so we had plenty of time to explain the test to her and to devise appropriate therapeutic strategies. The patient chose to undergo the test, and based on the results, she was able to assess what kind of surgery was best for her. Our patient was a lucky case. However, there is not much time to spare for patients who have surgery as initial treatment.

It is a heavy burden for patients to decide treatment and surgical options in a limited time. When conducting an examination, it is necessary to give a sufficient explanation, including the significance of the examination. However, from the perspective of a breast surgeon, we should be careful not to burden the patient unnecessarily with regard to genetic tests and their results. Depending on the degree of the patient's understanding, they may be ambivalent, and so a genetic test may be performed after surgery. Such a situation may be unavoidable, but there is tremendous merit in performing genetic testing before surgery.

At present, recognition and interest in BRCA genes is increasing in Japan. However, the reality is that BRCA genetic testing is still rare, including when cancer recurs. In addition, preoperative examinations are rarely performed. I believe that medical professionals must reform their own practice and actively perform genetic testing for the benefit of their patients.

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