LOKMAN HEKIM HEALTH SCIENCES

DOI: 10.14744/lhhs.2021.80011
Lokman Hekim Health Sci 2021;1(3):81–84

lokmanhekimhs.com

ORIGINAL ARTICLE

Evaluation of Family History in Terms of Breast Cancer in Cases Applying for Breast Ultrasonography

Meme Ultrasonografisi Amacıyla Başvuran Olgularda Meme Kanseri Açısından Aile Öyküsünün Değerlendirilmesi

Deniz Esin Tekcan Şanlı[1,2]

[1]Department of Medical Imaging Techniques, Vocational School of Health Services, İstanbul Rumely University, İstanbul, Turkey
[2]Department of Radiology, Acıbadem Kozyatağı Hospital, İstanbul, Turkey

Abstract

Introduction: The present study aims to investigate the family history of breast cancer in patients who underwent breast ultrasonography for any reason.

Methods: Patients at any age who underwent breast ultrasonography examination in our clinic due to family history, complaints of a palpable mass in the breast–breast pain, and cancer screening over the age of 40 years were included in the study between 2019 and 2021. Regardless of whether there is an individual diagnosed with breast cancer in each case's family, the degree of proximity was asked. Additionally, the age of the cases and whether they gave birth or not were questioned and recorded.

Results: Of the 3783 breast ultrasonography patients, 349 had a family history of breast cancer. Breast cancer was diagnosed in one relative in 79% (n=276), in two relatives in 15.5% (n=54), in three relatives in 5% (n=17), and in four relatives in 0.5% (n=2) with breast cancer in 3% (n=10). Of the cases, 181 had breast cancer in first-degree relatives; 169, in second-degree relatives; and 48, in third-degree relatives.

Discussion and Conclusion: Although the genetic transmission of breast cancer, which is the most common cancer in women, is high, it is possible to achieve a significant decrease in mortality and morbidity rates with early diagnosis, thanks to routine breast examination and imaging by all women with or without a family history of breast cancer.

Keywords: BRCA 1; BRCA 2; Breast ultrasonography; Hereditary breast cancer

Breast cancer is the most common cancer in women and is an important cause of mortality and morbidity in the female population worldwide.[1,2] Nowadays, one out of every 9–12 women is diagnosed with breast cancer.[3] These rates are higher in large and industrialized cities. With the widespread use of screening programs, it has become possible to diagnose at an early stage. The most important risk factor for breast cancer is the female

Cite this article as: *Tekcan Şanlı DE. Evaluation of Family History in Terms of Breast Cancer in Cases Applying for Breast Ultrasonography. Lokman Hekim Health Sci 2021;1(3):81–84.*

Correspondence: Deniz Esin Tekcan Şanlı, M.D. İstanbul Rumeli Üniversitesi Sağlık Hizmetleri Meslek Yüksekokulu, Tıbbi Görüntüleme Teknikleri Anabilim Dalı, İstanbul, Turkey
E-mail: tekcandenizesin@gmail.com **Submitted:** 26.08.2021 **Accepted:** 28.10.2021

gender, and the most important underlying cause is hormonal factors.[1,2] Among the main hormonal risk factors, it is possible to list the causes such as early menarche, late menopause, use of exogenous hormonal drugs, not giving birth, advanced maternal age, and obesity. Breast cancer is one of the cancers that has the highest genetic transmission rate among all cancers.[1,4] Since breast cancers show a dominant inheritance feature, especially, cases diagnosed with breast cancer in their first-degree relatives should participate in screening programs at an earlier age and they should take care by having their radiological examinations such as breast ultrasonography, mammography, and breast magnetic resonance imaging performed regularly at specified intervals. Although the risk of breast cancer cannot be reduced, it is possible to reduce morbidity and mortality rates, improve prognosis, and increase survival with early diagnosis. This study was aimed to reveal the demographic characteristics of patients who applied to our clinic for breast ultrasonography examination by questioning their family history in terms of breast cancer.

Materials and Methods

Patients who underwent breast ultrasonography examination in our clinic for any reason (cancer screening over 40 years of age, routine breast examination due to family history, and palpable mass in the breast–breast pain) between March 2019 and April 2021 were included in the study. Regardless of whether there is an individual diagnosed with breast cancer in each case's family, the degree of proximity, if any, was asked. Additionally, the age of the cases and whether they gave birth or not were questioned and recorded. Family history was questioned only in terms of breast cancer, and cases with a history of endometrium/ovarian/colon cancer or other cancer were excluded from the study. IRB was obtained from the ethics committee of our hospital (date: 09.06.2021 number: 2021-10/01). Since we used anonymous clinical data and the individual could not be identified on the basis of the available data, patients were not required to give informed consent for inclusion in this retrospective study.

Statistical Analysis

SPSS 21 (SPSS Inc., IBM Company, Chicago, USA) program was used for statistical analysis. Descriptive data were presented as mean, standard deviation, minimum and maximum values, frequency, and ratio. Since the total number of cases was relatively small, no inferential statistical analysis was performed.

Results

A total of 3783 breast ultrasonography examinations were performed within the specified date range. Of these cases, 349 had a family history of breast cancer. These 349 cases with breast cancer in at least one family member were included in the study. The mean age of the cases was 43.7 (21–75) years; 13.5% of the cases (n=47) did not give birth, and 86.5% (n=302) gave birth.

Breast cancer in one relative in 79% (n=276), in two relatives in 15.5% (n=54), in three relatives in 5% (n=17), and in four relatives in 0.5% (n=2) was diagnosed; 3% (n=10) of the cases were also diagnosed with breast cancer themselves. Of the cases, 181 had first-degree relatives with breast cancer; 169 had second-degree relatives; and 48 had third-degree relatives. Mothers in 148 cases, aunts in 90 cases, grandmothers in 41 cases, cousins (aunts' daughters or aunts' sons) in 49 cases were diagnosed with breast cancer. Aunts in 44 cases, grandmothers in 13 cases, grandfathers in two cases, and fathers in two cases were diagnosed with breast cancer. Daughters or siblings of 24 patients were diagnosed with breast cancer. When their daughters and siblings were excluded, there was maternal side in 79% (256/325), paternal side in 15% (48/325), and a relative diagnosed with breast cancer on both maternal and paternal sides in 6% (21/325) of cases.

Discussion

Family history is a well-defined risk factor for breast cancer.[1,2] Although a normal woman's lifetime risk of breast cancer is 10%–12%, this rate is even higher in cases with a family history of breast cancer.[1] The genetic similarity rate with first-degree relatives (mothers, fathers, siblings, and children) is 50%. Those with second-degree relatives (aunts, grandmothers, grandfathers, uncles, and nieces) account for 25%, and those with third-degree relatives (cousins), 12.5%. Considering this degree of genetic similarity, the risk of breast cancer genetically increases as the degree of consanguinity decreases. Colditz et al.[5] reported that 4.6% of women had mothers, 1.5% had at least one sister, and 0.1% had both mother and sister. In our study, breast cancer was diagnosed in first- and second-degree relatives in most of the cases. The incidence of breast cancer on the maternal side was significantly higher. In most of our case series, mothers were diagnosed with breast cancer; 3% of our patients who had breast cancer in at least one family member also had their own diagnosis of breast cancer.

Although hereditary and familial breast cancers are used with the same name, they are different in definition. Hered-

itary breast cancer is defined in breast/ovarian cancer syndromes, in at least three first-degree relatives with a history of breast–ovarian–endometrium–colon cancer or in at least one person under 50 years of age with a history of cancer. Familial cancer is defined in patients with at least two first- or second-degree relatives with a history of breast/ovarian cancer at an early age.[3,6] Although males with breast cancer are very rare and constitute only 1% of all breast cancer cases, the probability of genetically transmitted breast cancers significantly increases when a male family member has breast cancer.[7] In our series, the male relatives (1.2%) of four patients (two fathers and two grandfathers) had breast cancer.

Having a family history of breast cancer has also an effect on the stage and grade of the tumor. However, women who had a family history of breast cancer have a significantly higher probability of axillary lymph node metastasis and cancer stages at the time of diagnosis. The majority of women with no family history of breast cancer have a low histological grade (grade 1). In other words, tumors seen in women without a family history of breast cancer are less aggressive but are diagnosed late.[8,9] Again, in a study conducted on young women with breast cancer, those with a family history of cancer had smaller tumor sizes and lower axillary lymph node metastasis rates at the time of diagnosis than women with no family history.[10] It can be suggested that the reason for this situation is that women with a family history of breast cancer are more conscious of cancer and are diagnosed earlier because they do not disrupt their routine breast controls.

Different studies have shown that the cancer subtype and aggressiveness observed in the family, the degree of consanguinity of the case with cancer, and the number of relatives with cancer may also affect the stage of cancer. Several studies on this subject have shown that patients with a family history of breast cancer have a higher TNM stage, higher grade, and more triple-negative breast cancer.[8] Although the probability of triple-negative cancer is higher in those with a family history of triple-negative cancer or breast/ovarian cancer syndrome, such a relationship was not found in other histopathological subtypes and the Ki-67 proliferation rate.[8] The conclusion to be drawn from this is that the genetic transmission of breast cancer such as the triple-negative one with an aggressive course and poor prognosis is higher than other subtypes.

Most of all breast cancers occur spontaneously, with no family history. Familial or hereditary breast cancers constitute 10%–15% of all breast cancer cases.[3] Of the familial breast cancers, 30%–50% are associated with BRCA 1 and BRCA 2 mutations.[11,12] Although the lifetime risk of breast cancer in a normal woman is 10%–12%, this risk increases to 40%–85% in those with a BRCA mutation.[3] Moreover, more aggressive tumors develop in breast cancer cases with BRCA mutations. It has been shown that more than half of the tumors associated with the BRCA 1 mutation are in the triple-negative and approximately 40% of the tumors associated with the BRCA 2 mutation are in the Luminal B histopathological subgroup.[12–14] Thus, risk-reducing methods such as prophylactic mastectomy, prophylactic oophorectomy, and chemoprevention (anti-estrogenic hormone therapy) may be available in cases with BRCA 1/2 mutation.[3]

Study Limitations

Although our case series is quite high when compared with several clinics, our single-center study has some limitations. Other factors such as mammographic breast density, increased body mass index, hormonal status, which are considered risk factors for breast cancer, were not evaluated in our study. In relatives diagnosed with breast cancer, the histopathological subtype of the tumor and the stage and extent of the disease at the time of diagnosis were not known or remembered by most cases, so they were ignored in the study. Additionally, since the patients included in the study did not have long-term follow-ups, whether most of these patients developed breast cancer is not known.

Conclusion

Our aim in this study was not to evaluate breast cancer, the role of ultrasonography in the diagnosis of breast cancer, and the relationship between breast cancer and family history. Our aim in this study was to question the family history of breast cancer in patients who came for routine breast ultrasonography examination for any reason (cyst, fibroadenoma, family history, and screening program over 40 years of age). Hence, this study was aimed to cross-sectionally reveal the family history of breast cancer in cases who underwent breast ultrasonography in our center. It was evaluated multidimensionally in terms of the number of people with breast cancer, degree of relationship, and parents' side. On the basis of the result of the study, we aimed to raise awareness in terms of public health and general cancer screening programs by showing that the patient population applying to our clinic for ultrasonography has a high family history and cancer burden in terms of breast cancer. Also, we wanted to emphasize again that screening programs should be started earlier than normal in cases with a family history of breast cancer.

In summary, having a family history of breast cancer is one of the most important reasons that increase a woman's risk of breast cancer. Patients with a breast cancer diagnosis in their family, especially in their first-degree relatives, are diagnosed at an early stage because they are more conscious and sensitive in terms of breast cancer screening and they are meticulous in their follow-up. All women, with or without a family history of cancer, should be sensitive to breast cancer screening. In this way, early diagnosis may enhance prognosis and survival, even with aggressive tumoral histology.

Peer-review: Externally peer-reviewed.

Conflict of Interest: None declared.

Financial Disclosure: The author declared that this study received no financial support.

References

1. Rojas K, Stuckey A. Breast cancer epidemiology and risk factors. Clin Obstet Gynecol 2016;59(4):651–72. [CrossRef]
2. Sun YS, Zhao Z, Yang ZN, Xu F, Lu HJ, Zhu ZY, et al. Risk factors and preventions of breast cancer. Int J Biol Sci 2017;13(11):1387–97. [CrossRef]
3. Lalloo F, Evans DG. Familial breast cancer. Clin Genet 2012;82(2):105–14. [CrossRef]
4. Valencia OM, Samuel SE, Viscusi RK, Riall TS, Neumayer LA, Aziz H. The role of genetic testing in patients with breast cancer: a review. JAMA Surg 2017;152(6):589–94. [CrossRef]
5. Colditz GA, Rosner BA, Speizer FE. Risk factors for breast cancer according to family history of breast cancer. For the Nurses' Health Study Research Group. J Natl Cancer Inst 1996;88(6):365–71. [CrossRef]
6. Shiovitz S, Korde LA. Genetics of breast cancer: a topic in evolution. Ann Oncol 2015;26(7):1291–9. [CrossRef]
7. Lax SF. Hereditary breast and ovarian cancer. [Article in German]. Pathologe 2017;38(3):149–55. [CrossRef]
8. Liu L, Hao X, Song Z, Zhi X, Zhang S, Zhang J. Correlation between family history and characteristics of breast cancer. Sci Rep 2021;11(1):6360. [CrossRef]
9. Jannot AS, Usel M, Bouchardy C, Schubert H, Rapiti E. Breast cancer family history leads to early breast cancer detection and optimal management. Cancer Causes Control 2017;28(9):921–8. [CrossRef]
10. Malone KE, Daling JR, Doody DR, O'Brien C, Resler A, Ostrander EA, et al. Family history of breast cancer in relation to tumor characteristics and mortality in a population-based study of young women with invasive breast cancer. Cancer Epidemiol Biomarkers Prev 2011;20(12):2560–71. [CrossRef]
11. Antoniou A, Pharoah PD, Narod S, Risch HA, Eyfjord JE, Hopper JL, et al. Average risks of breast and ovarian cancer associated with BRCA1 or BRCA2 mutations detected in case Series unselected for family history: a combined analysis of 22 studies. Am J Hum Genet 2003;72(5):1117–30. [CrossRef]
12. Neuhausen S, Gilewski T, Norton L, Tran T, McGuire P, Swensen J, et al. Recurrent BRCA2 6174delT mutations in Ashkenazi Jewish women affected by breast cancer. Nat Genet 1996;13(1):126–8. [CrossRef]
13. Sønderstrup IMH, Jensen MR, Ejlertsen B, Eriksen JO, Gerdes AM, Kruse TA, et al. Subtypes in BRCA-mutated breast cancer. Hum Pathol 2019;84:192–201. [CrossRef]
14. Ha SM, Chae EY, Cha JH, Kim HH, Shin HJ, Choi WJ. Association of BRCA mutation types, imaging features, and pathologic findings in patients with breast cancer With BRCA1 and BRCA2 mutations. AJR Am J Roentgenol 2017;209(4):920–8.