EJMA

DOI:10.14744/ejma.268077
EJMA 2026;(2):77–80

Research Article

Investigation of the Clinical and Demographic Characteristics of Our Patients With Breast Cancer Diagnosed Before the Age of 40

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Abstract

Objectives: Breast cancer diagnosed before the age of 40 accounts for 7% of all cases and is associated with aggressive disease. This study aimed to investigate the clinical, demographic, and pathological characteristics of young patients with breast cancer in Türkiye.

Methods: A retrospective analysis was conducted on 73 patients diagnosed with breast cancer before the age of 40 at Necmettin Erbakan University between 2011 and 2020. Demographic data, clinical presentation, pathology, treatment, and recurrence/metastasis were analyzed.

Results: All patients were female, with a mean age of 30.9±4.1 years. The most common symptom was a palpable breast mass (61.6%). Invasive ductal carcinoma was predominant (87.7%). The molecular subtypes were as follows: luminal, 65.8%; HER2-positive, 20.5%; and triple-negative, 13.7%. Metastasis at diagnosis occurred in 21.9% of patients, and recurrence during follow-up occurred in 23.3%. In the luminal subgroup, recurrence was more common than metastasis (82.4% vs. 43.8%, $p<0.05$). In the HER2-positive subgroup, metastasis was more common than recurrence (56.3% vs. 5.9%, $p<0.05$). Among the 13 patients tested for BRCA mutations, 3 (23%) had pathogenic variants.

Conclusion: Young Turkish patients with breast cancer showed high rates of aggressive molecular subtypes. The different recurrence and metastasis patterns between the luminal and HER2-positive subtypes suggest the need for subtype-specific follow-up strategies.

Keywords: Breast cancer, clinical characteristics, Türkiye, under 40, young

Cite This Article: *Bülbül B, Karaağaç M, Artaç M. Investigation of the Clinical and Demographic Characteristics of Our Patients With Breast Cancer Diagnosed Before the Age of 40. EJMA 2026(2): 77-80.*

Breast cancer is the most frequently diagnosed cancer among women worldwide, with an estimated 2.3 million new cases in 2020.[1] Although breast cancer predominantly affects older women, approximately 7% of newly diagnosed cases occur in women under 40 years of age.[2] In Türkiye, breast cancer accounts for 23.9% of all female cancers.[3]

Young-onset breast cancer presents unique clinical challenges. These patients are not included in routine screening programs, leading to delayed diagnosis and a more advanced stage at presentation.[4] A young age at diagnosis is associated with more aggressive tumor biology, a higher grade, greater lymph node involvement, and a poorer prognosis.[5,6]

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Submitted Date: April 16, 2026 **Accepted Date:** August 5, 2026

Table 1. Demographic and clinical characteristics (N=73)

Characteristic	n (%)
Mean age (years, ±SD)	30.9±4.1
Married	66 (90.4)
Oral contraceptive use	10 (13.7)
IUD use	10 (13.7)
Nulliparous	11 (15.1)
Presenting symptom: breast mass	45 (61.6)
Tumor location: upper outer quadrant	42 (57.5)
Clinical T4	16 (21.9)
Axillary lymph node positive	43 (58.9)

SD: Standard deviation; IUD: Intrauterine device.

Table 2. Pathological and molecular characteristics

Characteristic	n (%)
Invasive ductal carcinoma	64 (87.7)
Grade 3	15 (20.5)
ER positive	46 (63.0)
PR positive	40 (54.8)
HER2 3+	26 (35.6)
Luminal	48 (65.8)
HER2-positive	15 (20.5)
Triple-negative	10 (13.7)

ER positive: Estrogen receptor-positive breast cancer; PR positive; Progesterone receptors-positive breast cancer; HER2: Human epidermal growth factor receptor 2.

Table 3. Metastasis and recurrence by molecular subtype

Subtype	Metastasis (n)	Recurrence (n)
Luminal	7 (43.8)	14 (82.4)
HER2-positive	9 (56.3)	1 (5.9)
Triple-negative	0 (0)	2 (11.8)
Total	**16 (100)**	**17 (100)**

HER2: Human epidermal growth factor receptor 2.

The molecular landscape of breast cancer in young women differs from that in older patients, with higher proportions of triple-negative and HER2-positive subtypes.[7,8] Genetic predisposition also plays a prominent role, with BRCA mutations accounting for a substantial proportion of early-onset cases.[9]

Data from Turkish populations remain limited. This study aimed to investigate the clinical, demographic, and pathological characteristics of patients with breast cancer diagnosed before the age of 40 at a tertiary care center in Türkiye.

Methods

Study design and population

This retrospective, single-center study was conducted at Necmettin Erbakan University Meram Faculty of Medicine, Department of Medical Oncology. The study protocol was approved by the Institutional Ethics Committee (approval date: 2022; approval no.: 2022/3665) and was conducted in accordance with the Declaration of Helsinki.

Patients were included if they: 1) were diagnosed with breast cancer between January 2011 and December 2020; 2) were under 40 years of age at diagnosis; 3) received at least part of their treatment at our institution; and 4) had complete medical records available.

Data Collection

Data were extracted from electronic medical records. The collected variables included demographic data, clinical presentation, imaging findings, pathological features (ER, PR, HER2, Ki-67, and grade), treatment modalities, and clinical outcomes (metastasis at diagnosis and recurrence during follow-up).

ER and PR positivity was defined as ≥1% nuclear staining. HER2 positivity was defined as 3+ staining by immunohistochemistry or 2+ staining with FISH amplification. Molecular subtypes were classified as luminal (ER/PR-positive and HER2-negative), HER2-positive, and triple-negative.

Statistical Analysis

Statistical analyses were performed using SPSS version 22.0. Categorical variables were compared using the chi-square test. Continuous variables were expressed as the mean±standard deviation. A p-value of <0.05 was considered statistically significant.

Results

Demographic characteristics

A total of 73 patients were included. All patients were female, with a mean age of 30.9±4.1 years (range: 20–39 years). At diagnosis, 90.4% (n=66) were married. Oral contraceptive use was reported in 13.7% (n=10), and intrauterine device (IUD) use was reported in 13.7% (n=10). The nulliparity rate was 15.1% (n=11) (Table 1).

Clinical presentation

The most common presenting symptom was a palpable breast mass (61.6%; n=45). Tumors were most frequently localized in the left breast (56.1%; n=41) and the upper outer quadrant (57.5%; n=42). The clinical T-stage distribution

was as follows: T1, 38.4% (n=28); T2, 38.4% (n=28); and T4, 21.9% (n=16). Axillary lymph node positivity was present in 58.9% (n=43) (Table 1).

Pathological and molecular characteristics

Invasive ductal carcinoma was the most common histological type (87.7%; n=64). The tumor grade was grade 2 in 31.5% (n=23) and grade 3 in 20.5% (n=15). ER positivity was observed in 63.0% (n=46), and PR positivity was observed in 54.8% (n=40). HER2 3+ expression was observed in 35.6% (n=26). The molecular subtype distribution was as follows: luminal, 65.8% (n=48); HER2-positive, 20.5% (n=15); and triple-negative, 13.7% (n=10) (Table 2).

Metastasis, recurrence, and molecular subtypes

Metastatic disease at diagnosis was present in 21.9% (n=16). Recurrence during follow-up developed in 23.3% (n=17). In the luminal subgroup, recurrence was significantly more frequent than metastasis (82.4% vs. 43.8%, $p<0.05$). In the HER2-positive subgroup, metastasis was significantly more frequent than recurrence (56.3% vs. 5.9%, $p<0.05$) (Table 3).

BRCA mutations

BRCA1/2 mutation analysis was performed in 13 patients (17.8%). Pathogenic variants were detected in 3 patients (23%).

Discussion

In this study, we described the clinical, demographic, and pathological characteristics of 73 patients diagnosed with breast cancer before the age of 40 in Türkiye. The mean age of 30.9 years is consistent with the literature.[2,8]

The finding that a palpable breast mass was the most common presenting symptom (61.6%) reflects the absence of routine screening in this age group.[4,10] The 87.7% rate of invasive ductal carcinoma is consistent with previous reports.[11,12]

Our triple-negative rate (13.7%) was lower than those reported in Western series (20%–30%).[7,8], which may be due to the sample size, genetic differences, or environmental factors. HER2 positivity (20.5%) was consistent with the literature.[7]

A striking finding was the difference in recurrence and metastasis patterns between the luminal and HER2-positive subgroups. The higher recurrence rate in luminal patients suggests the need for closer long-term follow-up. The higher rate of metastasis at diagnosis in HER2-positive patients indicates a more aggressive initial course, although current anti-HER2 therapies appear to be effective.[13,14]

The prevalence of BRCA mutations (23% among those tested) emphasizes the importance of genetic counseling in young patients.[9,15]

Study limitations

The limitations of this study include its single-center, retrospective design, small sample size, and high rates of missing data for tumor grade (45.2%) and Ki-67 (61.6%). BRCA testing was performed in a limited number of patients.

Conclusion

Young Turkish patients with breast cancer showed high rates of aggressive molecular subtypes. The different recurrence and metastasis patterns between the luminal and HER2-positive subgroups suggest the need for subtype-specific surveillance strategies. Larger multicenter studies are needed.

Disclosure

Ethical Committee Approval: This retrospective, single-center study was conducted at Necmettin Erbakan University Meram Faculty of Medicine, Department of Medical Oncology. The study protocol was approved by the Institutional Ethics Committee (approval date: 2022; approval no: 2022/3665) and conducted in accordance with the Declaration of Helsinki.

Informed Consent: Written informed consent was obtained from all study participants before enrollment.

Conflict of Interest: The authors declare no conflict of interest.

Funding: No funding was received for this study.

Use of AI for Writing Assistance: No artificial intelligence (AI)-assisted technologies, including Large Language Models (LLMs), chatbots, or image creators, were used in the production of this submitted work.

Author Contributions: Concept: BB, MA, MK; Design: BB, MA, MK; Supervision: MA; Data Collection and/or Processing: BB, MK; Analysis and/or Interpretation: BB, MA; Literature Review: BB; Writing: BB; Critical Review: MA, MK.

Peer-review: Externally peer-reviewed.

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