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Borderline Breast Phyllodes Tumor Coexisting with Intermediate- Grade Ductal Carcinoma In Situ and Focal Invasion: A Case Report and Literature Review

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Case Report

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Abstract

Introduction:

Borderline phyllodes tumor (PT) coexisting with ductal carcinoma in situ (DCIS) and invasive ductal carcinoma (IDC) is an exceptionally rare mixed breast lesion. Due to its low incidence and compositional complexity, it poses substantial diagnostic and management challenges. We present such a case to highlight the pitfalls in preoperative diagnosis and the rationale for a multidisciplinary therapeutic strategy.

Presentation of case:

A 72-year-old Chinese female presented with a 2-week history of a right breast mass. Ultrasound and mammography revealed a large (52.6 mm), well-circumscribed, lobulated solid mass (BI-RADS 4A/4B), suggestive of a phyllodes tumor. Core needle biopsy returned “fibroadenoma with usual ductal hyperplasia.” Subsequent lumpectomy showed borderline PT (80%), intermediate-grade DCIS (20%), and focal Grade II invasive carcinoma of no special type (< 1%, maximal invasive diameter 3 mm). Immunohistochemistry of the invasive component: ER(−), PR(−), HER-2(3+), Ki-67 60%. Given the patient’s preference against breast conservation, a right total mastectomy with sentinel lymph node biopsy was performed (0/1 nodes positive). Final staging: pT1aN0M0, Stage IA, HER-2 overexpression subtype. No adjuvant chemotherapy, targeted therapy, or radiotherapy was given. At 12-month follow-up, no local or distant recurrence was observed.

Conclusion

Definitive diagnosis of PT coexisting with DCIS and IDC relies on adequate excision specimens rather than biopsy alone. Clinical management should integrate PT behavior with standard breast cancer guidelines through multidisciplinary collaboration, balancing complete oncologic control against the risk of overtreatment. Longer follow-up is warranted to confirm durable outcomes.

1. Introduction

Breast phyllodes tumor (PT) is a rare fibroepithelial lesion of the breast, accounting for 0.3%–1% of all primary breast tumors[1]. PT consists of neoplastic stromal cells and non-neoplastic epithelial components, manifesting as a dual proliferation of both. The epithelial component is bilayered, arranged in a slit-like pattern, and surrounded by stromal components to form a lobulated structure, which gives rise to its characteristic "leaf-like" appearance[2]. PT predominantly occurs in women around 40 years of age, typically presenting as a unilateral, painless, well-defined mass, which may rapidly enlarge in a short period. Based on indicators such as stromal cellularity, atypia, mitotic count, stromal overgrowth, and tumor border characteristics, the WHO classifies PT into benign, borderline, and malignant categories[3].

The proportions of the three types exhibit regional variations: benign accounts for 35%–60%, borderline for 15%–25%, and malignant for 10%–15%. All types carry local recurrence as the primary risk, and the degree of malignancy may further increase upon recurrence.

Breast ductal carcinoma in situ (DCIS) originates from the ductal epithelial cells of the terminal ductal lobular unit. It is a non-invasive breast cancer where the lesion is confined within the breast ducts without breaching the basement membrane or invading the surrounding stroma. It accounts for 25% of all diagnosed breast cancers and is associated with a high survival rate[4]. DCIS predominantly occurs in middle-aged and elderly women. Most patients are asymptomatic, but some may present with a breast mass or nipple discharge, particularly bloody discharge. DCIS is considered a heterogeneous disease with diverse biological behaviors: the majority of lesions can remain quiescent in situ for a long time, while others may progress to invasive ductal carcinoma (IDC)[5].

This article reports a 72-year-old female patient diagnosed with a borderline phyllodes tumor (80%) of the right breast coexisting with ductal carcinoma in situ (20%) and focal invasive carcinoma (no special type, histological Grade II, < 1%). The coexistence of these three tumor components is exceedingly rare. By reviewing previous literature, this article discusses the imaging features, pathological characteristics, and diagnostic and treatment processes of this disease, aiming to provide a reference for clinical decision-making regarding such rare mixed lesions.

2. Case Description

A 72-year-old Chinese female patient was admitted due to "discovery of a right breast mass for half a month." Half a month prior, the patient incidentally noticed a walnut-sized mass on the lateral side of her right breast. There was no pain, no chills, no fever, and no nipple discharge. Her spirit, diet, urination, and defecation were normal, with no significant weight change. She was previously healthy with no history of infectious diseases, no surgical history, and no family history of breast tumors.

Physical Examination: Both breasts were symmetrical and of normal size. The skin of both breasts showed no redness, swelling, or ulceration, and no "dimpling sign" or "peau d'orange" appearance. Both nipples were symmetrical, with no fixation, retraction, or elevation, and no nipple discharge or erosion. A mass measuring approximately 5 cm × 3 cm could be palpated at the 9 o'clock position of the right breast, about 3 cm from the nipple. It was hard in texture, with an irregular surface, blurred borders, and poor mobility. No obvious mass was palpated in the left breast. No enlarged lymph nodes were palpated in the bilateral axillary and supraclavicular regions.

After admission, color Doppler ultrasound of bilateral breasts and axillary lymph nodes was performed: At the 9 o'clock position of the right breast, 28 mm from the nipple, a solitary nodule was observed, measuring approximately 52.6 mm × 22.5 mm. It was oriented parallel to the skin, oval in shape, with large lobulations and smooth margins. The internal echo was hypoechoic, with no obvious calcifications. Color Doppler flow imaging (CDFI) showed blood flow signals within the lesion, and an arterial blood flow spectrum was detected. Imaging conclusions: 1. Solid nodule in the right breast, BI-RADS category 4A,

consider phyllodes tumor? 2. No obvious space-occupying lesion in the left breast, BI-RADS category 1; 3. No obviously enlarged lymph nodes in bilateral axillae. Bilateral mammography suggested: A slightly high-density mass (CC: 18/40, MLO: 25/49) was seen in the middle and posterior 1/3 of the outer quadrant of the right breast, measuring approximately 50.4 mm × 38.7 mm, with relatively clear borders and smooth margins, showing obvious large lobulations (Fig. 2A). Additionally, a round-like high-density nodule (MLO: 8/49) was seen in the middle 1/3 of the central area of the right breast, measuring approximately 14.2 mm × 14 mm, with clear borders and smooth margins, appearing to have ring-like calcifications. Imaging conclusions: 1. Right breast mass, BI-RADS category 4B, consider phyllodes tumor? 2. Right breast central area nodule, BI-RADS category 3; 3. Left breast calcifications (BI-RADS category 2).(Fig. 1) Other laboratory test results showed no obvious abnormalities.

Due to the large volume of the right breast mass and its high BI-RADS category, combined with factors such as the patient's age, there was a high clinical suspicion of malignancy. After informing the patient of the condition, a core needle biopsy of the right breast mass was performed. Postoperative pathology indicated: breast fibroadenoma with partial usual ductal epithelial hyperplasia. Given the inconsistency between imaging findings and biopsy pathology results, and that the tumor had surgical indications, a right breast lumpectomy was performed under local infiltration anesthesia. Postoperative pathology indicated: borderline phyllodes tumor (80%), intermediate-grade DCIS (20%), focal invasive carcinoma of no special type Grade II (< 1%), with a maximum invasive diameter of approximately 3 mm. Immunohistochemistry (invasive carcinoma component): GATA-3 (+), P120 (+), E-Cad (membrane +), P63 (-), CK5/6 (partial +), Calponin (-), ER (-), PR (-), HER-2 (3+), Ki-67 (60%).(Fig. 2)

The patient was diagnosed with breast cancer coexisting with a phyllodes tumor, which carried indications for further radical resection. A right radical mastectomy or breast-conserving surgery could be chosen, and intraoperative sentinel lymph node frozen section could determine whether axillary lymph node dissection was necessary. The patient had no desire for breast conservation or reconstruction. After completing systemic auxiliary examinations and excluding surgical contraindications, a right radical mastectomy combined with right axillary sentinel lymph node biopsy was performed. One right sentinel lymph node was detected intraoperatively, with no cancer metastasis (0/1). Postoperative pathology of the right radical mastectomy specimen indicated: Microscopically, a small amount of residual intraductal carcinoma was seen, surrounded by inflammatory cell infiltration and granulation tissue proliferation. No vascular tumor thrombi or nerve bundle invasion was observed. No Paget's disease was found in the nipple and breast skin. The breast skin, base, superior, inferior, medial, and lateral margins showed no cancer involvement. Postoperative diagnosis: 1. Malignant tumor in the lateral-central quadrant of the right breast (invasive carcinoma: pT1aN0M0, Stage IA, HER-2 overexpression subtype); 2. Borderline breast phyllodes tumor.

The patient's general condition was good postoperatively. The drainage tube was removed on the 14th postoperative day, and she was discharged smoothly. No chemotherapy, targeted therapy, or radiotherapy was administered postoperatively. Follow-up at 3, 6, 9, and 12 months postoperatively

revealed no local or distant recurrence. The diagnosis and treatment process of the patient is shown in Fig. 3.

3. Discussion

PT was first described by the German pathologist Johannes Müller in 1838. It was named "cystosarcoma phyllodes" due to its cystic, fleshy, and leaf-like appearance on the cut surface, and he emphasized its benign nature[6]. After Lee reported the first case of metastasis in 1931, its potential malignancy was recognized[7]. Because the term "sarcoma" is easily misunderstood as highly malignant, the WHO adopted the neutral term "phyllodes tumor" in its 1982 classification, and in 2003 further classified it into benign, borderline, and malignant categories based on stromal cellularity, atypia, mitotic figures, stromal overgrowth, and border characteristics[8, 9].

PT is a fibroepithelial tumor of the breast, originating from the intralobular or periductal stroma of the terminal ductal lobular unit. It consists of benign epithelium and proliferating stroma that together form the "leaf-like" structure. Its growth driving force primarily originates from the monoclonal proliferation of the stroma. Recent studies indicate that as MED12 mutations, TERT promoter alterations, and chromosomal imbalances such as 1q gain and 13q loss accumulate, the stroma gradually escapes epithelial regulation and enters an autonomous growth state[10, 11]. Upon progression to malignancy, stromal sarcomatous changes, heterologous components, and hematogenous metastasis (primarily to the lungs and bones) may occur.

The diagnosis of PT requires a synthesis of clinical presentation, imaging, and pathological results. Clinically, it often presents as a painless mass within a unilateral breast that slowly enlarges over a long period; a minority of patients may present with rapid enlargement in a short time, even occupying the entire breast. Due to the expansive growth of PT, the tumor continuously pushes the superficial soft tissues, which may cause skin color changes, but it almost never invades the skin, nor does it present with peau d'orange. On ultrasound, it typically appears as a well-defined, lobulated solid mass with heterogeneous internal echoes. Sometimes, scattered slit-like structures can be seen internally, which is one of the characteristics of PT. This is caused by the abnormally abundant stroma within the tumor continuously compressing the glandular epithelium, trapping it within tiny slit-like regions, which appears as scattered, slit-like small anechoic areas under ultrasound[12]. Mammography lacks specificity, mostly presenting as a well-circumscribed round, oval, or lobulated mass, similar to a fibroadenoma. On MRI, it often presents as a high signal on T2-weighted imaging with marked enhancement post-contrast. The dynamic contrast-enhanced curve usually shows a rapid wash-in/wash-out or persistent rising pattern, which helps in evaluating the actual tumor size and its relationship with surrounding tissues. Histopathological examination is the gold standard for diagnosing and grading PT. Core needle biopsy can be used for preliminary diagnosis, but due to the heterogeneity of PT, the histological result is often described as a "fibroepithelial lesion," making it difficult to clearly distinguish PT from fibroadenoma; ultimately, diagnosis relies on a completely excised specimen.

In this case, preoperative ultrasound and mammography suggested a well-circumscribed, large lobulated solid mass, consistent with PT; the calcifications in the medial right breast also raised suspicion of coexisting DCIS. However, the core needle biopsy result was only "fibroadenoma with usual ductal hyperplasia," which is a typical underestimation due to the intra-tumoral heterogeneity of PT. The 2025 UK Association of Breast Surgery (ABS) guidelines note that core needle biopsy often only samples the stromal component, carrying a high risk of missing coexisting malignant epithelial components in borderline or malignant PT[13].Therefore, final grading and epithelial component assessment must rely on an adequate excision specimen. The coexistence of carcinoma in PT is not merely a coincidental occurrence. Although the epithelial component of PT is usually non-neoplastic, it may undergo DCIS due to paracrine stimulation by stroma-derived factors and further progress to invasive carcinoma in some cases. To our knowledge, previous reports in the literature regarding PT coexisting with DCIS or invasive carcinoma are already scarce, and cases with all three components coexisting are even rarer. This specific biological composition also explains why core needle biopsy easily misses the malignant epithelial component: the cancer foci are scattered and focally distributed within the stroma, making them difficult to capture with limited biopsy sampling. This further highlights the importance of an adequate excision specimen for accurate grading and component assessment.

We searched PubMed for all accessible full-text cases of PT, DCIS, and IDC coexisting, including this case, totaling 9 cases: 3 borderline PTs, 4 benign PTs, and 2 malignant PTs (Table 1).

Table 1
Cases of coexisting PT, DCIS and IDC components.

Refer	Age	Gender	Chief complaint	Compositional profile	Size(mm)	Surgical approach
Bozkurt et.[14]	42	female	A progressively enlarging mass in the right breast for 3 months.	Borderline PT+ DCIS + IDC	150	Total mastectomy +ALND
Trabelsi et.[15]	52	female	Pain and a mass in the right breast for 5 months.	Borderline PT+ DCIS + IDC	150	Total mastectomy + ALND
Panko et.[16]	70	female	A mass in the right breast for 9 years.	Benign PT+ DCIS + IDC	23	Total mastectomy +SLNB
Parfitt et.[17]	26	female	A mass in the right breast for 1 month.	Benign PT+ DCIS + IDC	33	Excision of breast mass + ALND
Salem et.[18]	40	female	A mass in the right breast for 1 year.	Benign PT+ DCIS + IDC	80	Excision of breast mass + SLNB
Ahmad et.[19]	33	female	Pain in the right breast and yellow nipple discharge for 4 days.	Benign PT+ DCIS + IDC	66.3	Excision of breast mass + SLNB
Korula et.[20]	51	female	A mass in the left breast for 9 months.	Malignant PT+DCIS + IDC	210	Total mastectomy + ALND
Erdogan et.[21]	22	female	A mass in the right breast was found	Malignant PT+DCIS + IDC	90	Total mastectomy + ALND
Our	72	female	A mass in the right breast for half a month	Borderline PT+ DCIS + IDC	52.6	Total mastectomy +SLNB

SLNB: Sentinel Lymph Node Biopsy; ALND: Axillary Lymph Node Dissection

Surgical resection is the preferred treatment for PT, aiming to completely excise the tumor and ensure adequate margins. The surgical approach depends on tumor size, histological grade, and the patient's specific condition. For benign PT, local excision, including ultrasound-guided vacuum-assisted excision, can be performed, with the goal of complete tumor removal without emphasizing margin width[22]. Borderline and malignant PT require wider excision (≥ 1 cm negative margin) to reduce the risk of recurrence[23]. For large tumors, unfavorably located tumors, or those where satisfactory margins cannot be achieved, a mastectomy may be necessary. However, due to the limitations of preoperative core needle biopsy, when postoperative histopathological examination reveals borderline or even

malignant PT with insufficient or positive margins, a secondary wider excision should be considered to reduce the risk of local recurrence, or postoperative adjuvant radiotherapy can be considered. It should be noted that there is currently no evidence that postoperative adjuvant radiotherapy improves overall survival in patients[24, 25].

Regarding the management of axillary lymph nodes, routine axillary lymph node surgery is generally not recommended because PT primarily metastasizes hematogenously, and lymph node metastasis is rare[26]. However, for patients with clear histopathological evidence of lymph node involvement, axillary lymph node dissection should still be performed. In this case, because DCIS with focal invasive carcinoma components was present, the surgical plan was formulated according to the surgical principles for breast cancer.

The value of postoperative adjuvant therapy for PT remains controversial: benign PT does not require radiotherapy; the role of radiotherapy in borderline and malignant PT remains to be clarified, and whether adjuvant chemotherapy is needed is also unclear. The European Society for Medical Oncology (ESMO) considers that the main indications for radiotherapy are high-grade, deep-seated tumors larger than 5 cm; for superficial lesions, even if larger than 5 cm, wide excision is usually the first choice, and whether to administer adjuvant radiotherapy requires comprehensive evaluation during multidisciplinary discussion based on tumor grade, margin status, location, and recurrence risk[27]. PT has limited sensitivity to conventional chemotherapy, and routine use is generally not recommended; however, for unresectable malignant PT or metastatic cases, chemotherapy regimens based on anthracyclines or ifosfamide can be considered[28]. Endocrine therapy, targeted therapy, and immunotherapy currently lack sufficient evidence to support their use.

Most importantly, the mixed presence of PT and DCIS/IDC fundamentally altered the clinical decision-making logic in this case. On one hand, the TNM staging of the mixed lesion should be determined based on the invasive carcinoma component—in this case, the final pathological stage was T1aN0M0 (Stage IA, HER-2 overexpression subtype), rather than being determined by the PT itself. On the other hand, the presence of DCIS/IDC components made the surgical margin requirements more stringent: a purely borderline PT requires a ≥ 1 cm negative margin, whereas when coexisting with DCIS, the margin criteria for breast cancer must also be met. In this case, because the patient had no desire for breast conservation, a total mastectomy was performed, thereby satisfying both margin requirements simultaneously; if breast-conserving surgery had been chosen, this contradiction would have been more prominent. The indication for sentinel lymph node biopsy also derived entirely from the invasive carcinoma component, not from the PT itself. Regarding postoperative adjuvant therapy, although this case was of the HER-2 (3+) overexpression subtype, the invasive carcinoma burden was extremely low (< 1%) and lymph nodes were negative. Combined with the limited sensitivity of PT to conventional chemotherapy, no chemotherapy, targeted therapy, or radiotherapy was administered following multidisciplinary discussion, and no recurrence was observed during one year of follow-up. For rare mixed lesions such as PT coexisting with carcinoma, adjuvant therapy should comprehensively evaluate tumor burden, molecular subtyping, and margin status to avoid overtreatment.

4. Conclusion

PT coexisting with DCIS and invasive carcinoma is extremely rare and is prone to underestimation by core needle biopsy. Definitive diagnosis relies on adequate excision specimens, and clinical management should follow a multidisciplinary collaborative model that integrates the behavioral characteristics of PT with breast cancer diagnosis and treatment guidelines, so as to avoid both under-treatment and over-treatment. Longer follow-up is still needed to confirm durable clinical oncological outcomes.

Abbreviations

ABS
Association of Breast Surgery
ALND
Axillary Lymph Node Dissection
BI-RADS
Breast Imaging Reporting And Data System
CC
Craniocaudal
CDFI
Color Doppler Flow Imaging
DCIS
Ductal Carcinoma In Situ
ER
Estrogen Receptor
ESMO
European Society for Medical Oncology
HER-2
Human Epidermal Growth Factor Receptor 2
IDC
Invasive Ductal Carcinoma
MLO
Mediolateral Oblique
PR
Progesterone Receptor
PT
Phyllodes Tumor
SLNB
Sentinel Lymph Node Biopsy;
WHO
World Health Organization

Declarations

Ethics approval and consent to participate

This case report was conducted in accordance with the principles of the Declaration of Helsinki. This study was approved by the Ethics Committee of the First Affiliated Hospital of Gannan Medical University, and written informed consent was obtained from the patient.

Consent for publication

The authors affirm that human research participants provided informed consent for publication.

Competing interests

The authors declare that they have no competing interest.

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Author Contribution

D.Y.: Original draft writing Data collection and processing, K. H.: Original draft writing, prepared figures 1-3 and table 1,H. G.: Contribution to content validation, H.T.: data analysis, S. H. and K. X.: Writing – Review & Editing, Y.L.: Data collection and processing, L. L.: Contribution to content validation, X. X.: Data collection and processing Writing – Review & Editing.

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Data Availability

The data without confidential proprietary information are available from the corresponding author on reasonable request.

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Figures

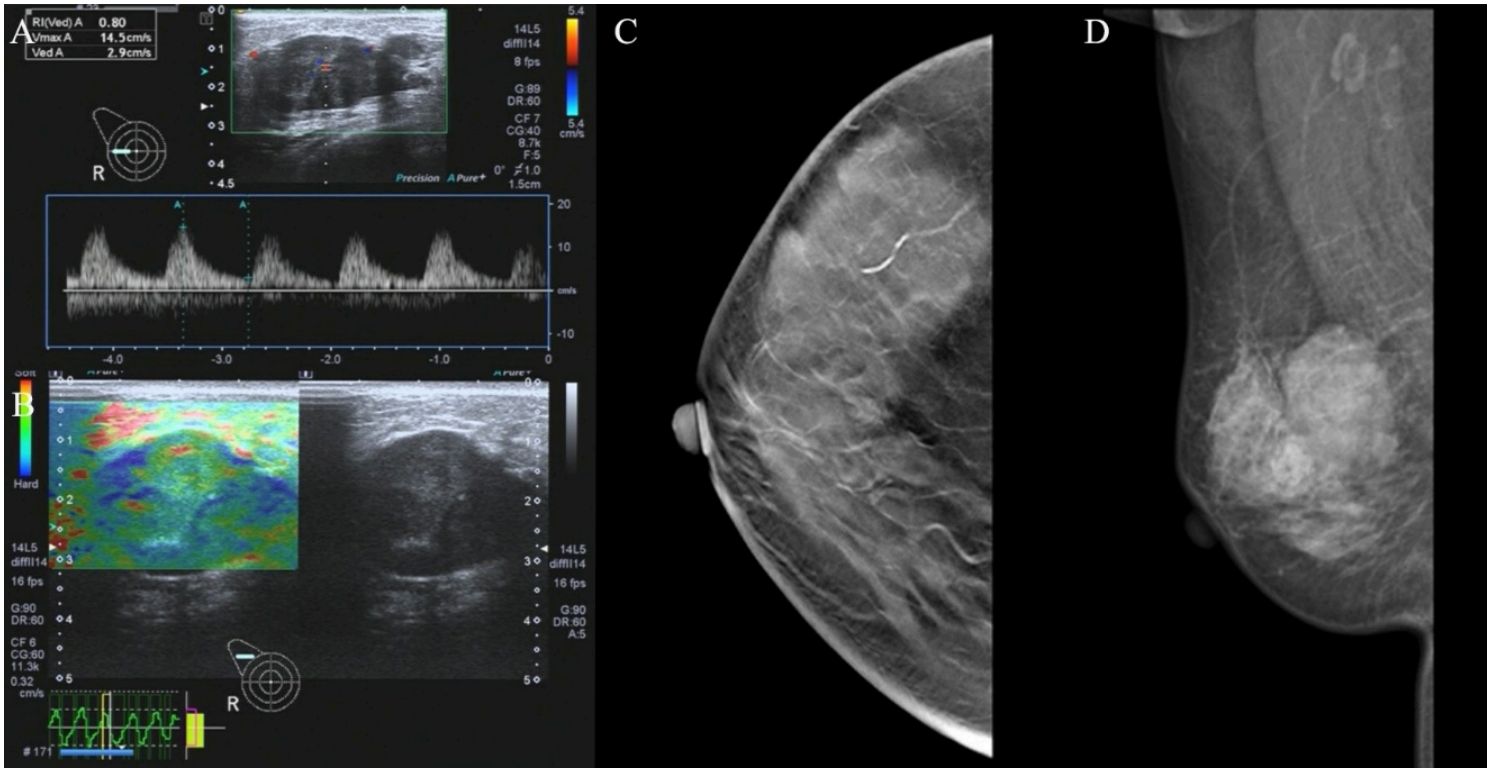


Figure 1

Multimodal imaging findings of the right breast lesion. (A) B-mode ultrasound with Color Doppler and pulsed-wave Doppler spectral imaging show an oval, hypoechoic, well-circumscribed, lobulated nodule with internal blood flow and a high resistive index (RI = 0.80, Vmax = 14.5 cm/s). (B) Ultrasound elastography demonstrating heterogeneous stiffness (mixed soft and hard areas) corresponding to the lobulated mass. (C, D) Craniocaudal (CC) and mediolateral oblique (MLO) mammography views demonstrate a large, slightly high-density mass with well-defined margins in the right breast, containing central ring-like calcifications.

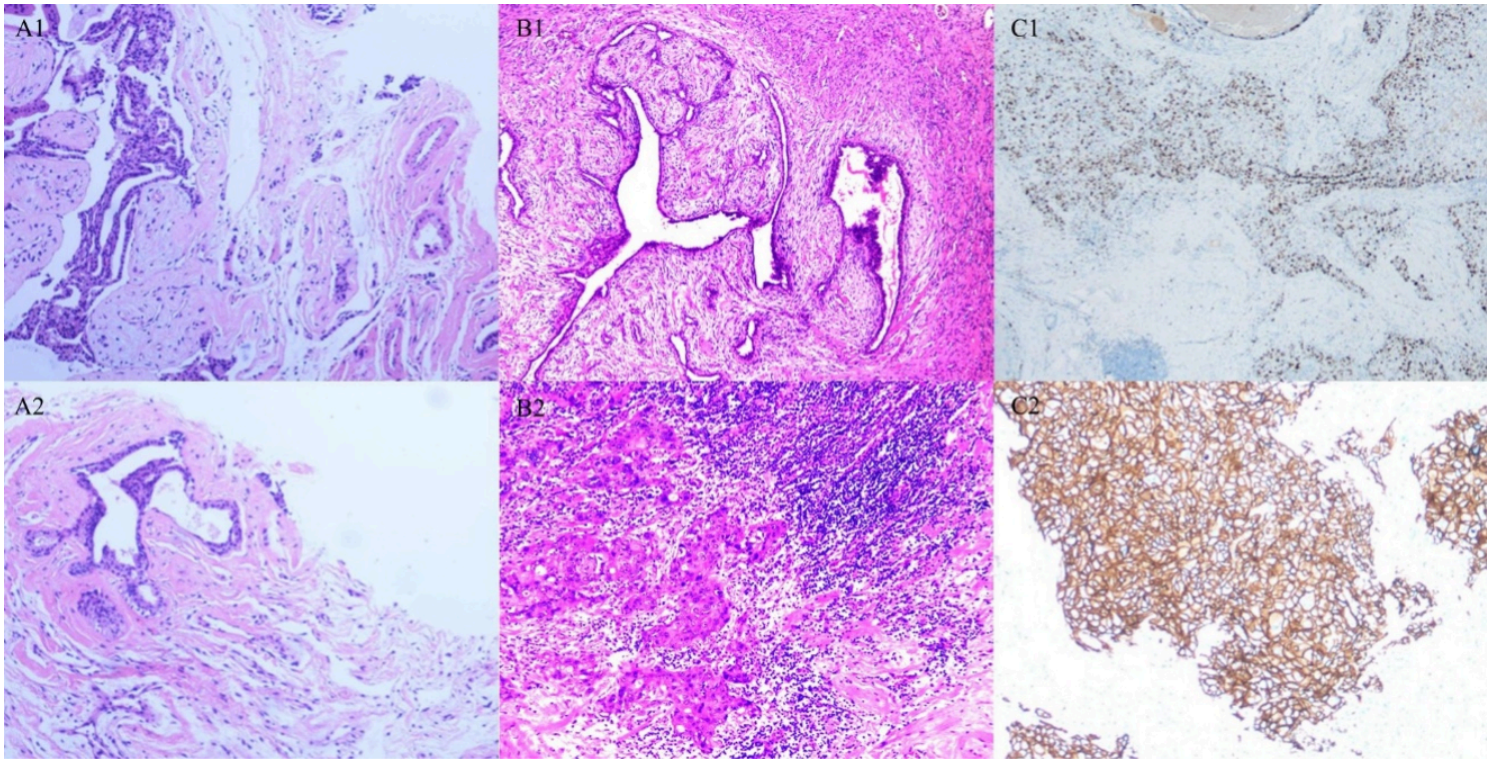


Figure 2

A1-A2: Core needle biopsy showing fibroadenoma with usual ductal hyperplasia(H&E). B1-B2: Postoperative specimen showing borderline PT+DCIS+IDC(H&E). C1-C2: Immunohistochemistry further confirmed the pathological diagnosis.

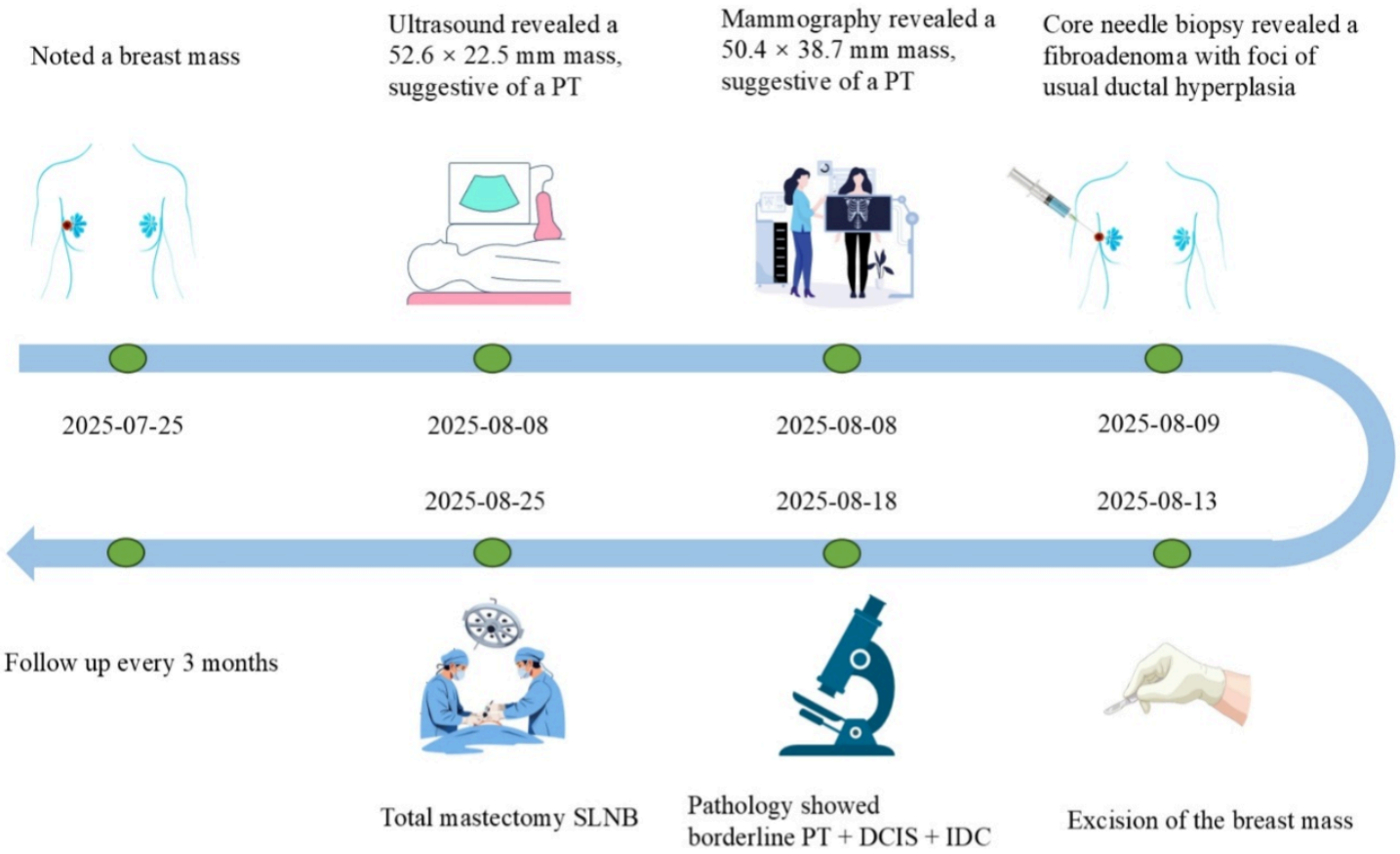


Figure 3

Flowchart of the patient's clinical course from initial symptom onset to hospital presentation.