Desmoid-type Fibromatosis of the Breast: A Case Report

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Desmoid-type fibromatosis (DTF) is a rare and locally aggressive benign tumor affecting the breasts. It is characterized by infiltrative growth and local recurrence, though it cannot metastasize. We present the case of a woman in her early 20s diagnosed with a palpable mass within her right breast. Imaging evidence was classified as Breast Imaging Reporting and Data System (BI-RADS) category 4 and above. We performed ultrasound-guided core needle biopsy, and pathological examination revealed hyperplasia of ductal epithelium and lobular tissue. Imaging results suggested a greater likelihood of the mass being malignant; however, pathological outcomes indicated that it was benign, resulting in the decision to perform an extended mastectomy. Postoperative pathological results suggested DTF. However, no further treatment was performed after surgery, and the patient's breast color Doppler ultrasound evaluation 12 months after surgery showed no recurrence or metastasis.

Keywords: desmoid-type fibromatosis; breast tumor; diagnosis; treatment

Introduction

Desmoid-type fibromatosis (DTF), also known as aggressive fibromatosis or desmoid tumor, is a clonal proliferation of fibroblastic that occurs in deep soft tissues. It is characterized by infiltrative growth with a propensity of local recurrence, though it lacks metastatic behavior, even when multifocal lesions within the same limb or body part [1]. DTF typically originates from fibroblasts and myofibroblasts and involves activation of the Wingless-type MMTV integration site family (WNT)/β-catenin signaling pathway.

While DTF commonly affects extra-abdominal regions such as the limbs or trunk, it rarely occurs in the breast [2]. When it arises in the breast parenchyma, DTF mimics imaging features of breast cancer, making diagnosis more challenging. Hence, reporting rare cases of DTF in the breast, like the one presented here, is crucial for enhancing awareness and improving the understanding of its clinical and pathological characteristics. This knowledge contributes to enhancing diagnostic accuracy and refining management strategies in clinical practice.

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Case Report

A woman in her early twenties was admitted to the Department of Breast Surgery at the First Hospital of Jilin University on 2 November 2021, two weeks after observing a mass in her right breast. The patient had no family history of breast cancer, no history of breast surgery or trauma, and was unmarried, not pregnant, and childless. She underwent the following assessments:

Clinical Breast Examination

During the physical examination, a tumor about 2.5 × 2.0 cm in size was observed in the outer upper quadrant of the right breast. The mass was hard, with a rough surface, unclear margins, good mobility, and no adhesion to the chest wall or surface derma.

Ultrasound

Imaging indicated a 19.2 × 7.2 mm irregularly shaped hypoechoic mass in the outer upper quadrant of the right breast, with unclear boundaries and visible blood flow (Fig. 1). Additionally, a 23.1 × 13.9 mm irregularly shaped hypoechoic mass with unclear margins and blood flow was also found (Fig. 2).
Furthermore, hyperplastic changes were identified in bilateral breasts, with no substantial abnormalities in the lymph nodes of both armpits. The observations were classified as Breast Imaging Reporting and Data System (BI-RADS) category 4.

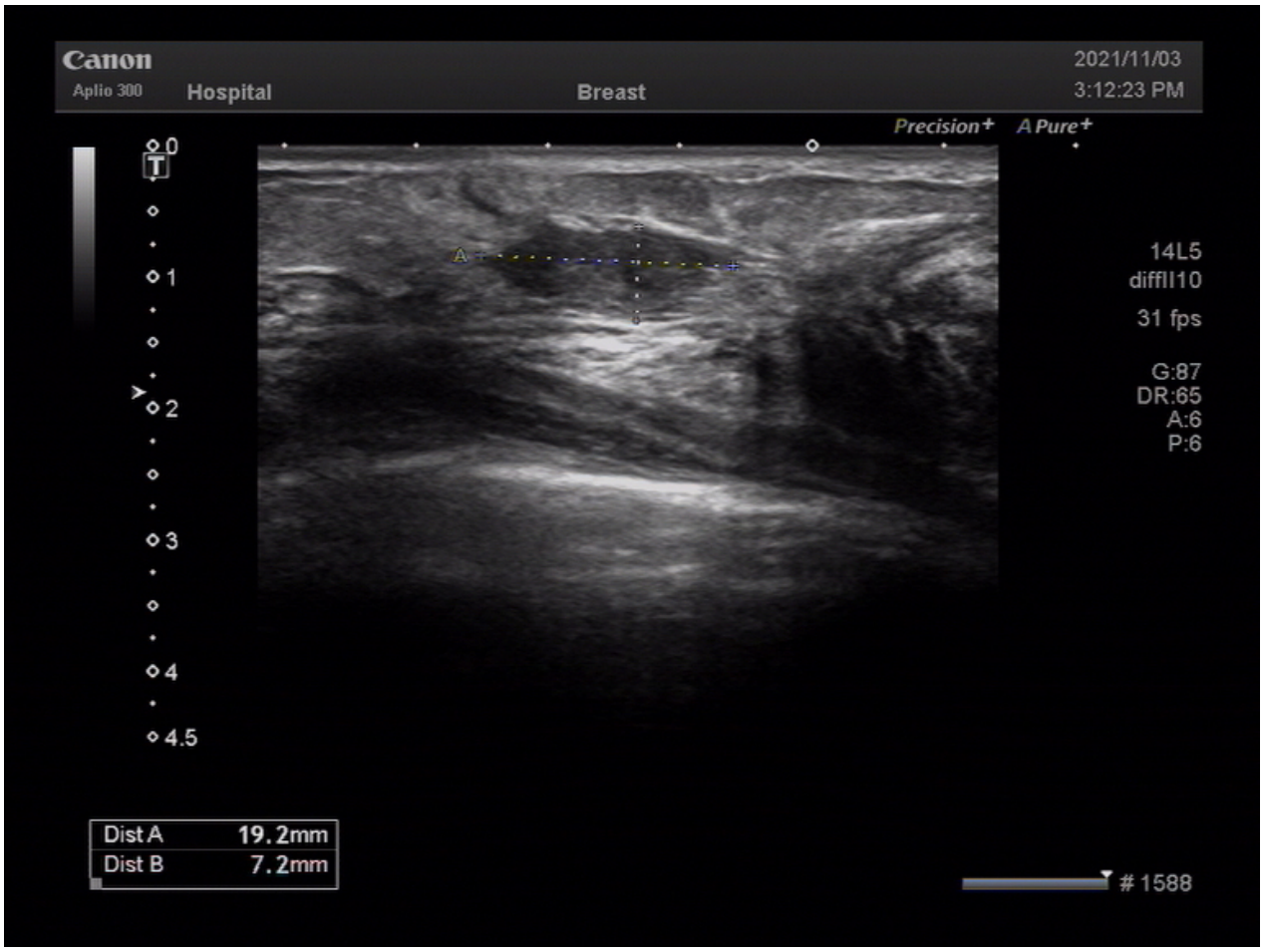

Fig. 1. Ultrasound indicated a 19.2 × 7.2 mm irregularly shaped hypoechoic mass in the outer upper quadrant of the right breast with unclear boundaries and visible blood flow.

Mammography

The bilateral mammary glands showed a polyglandular pattern, with structure distortions, radial changes, and skin thickening in the upper outer quadrant of the right breast (Fig. 3). However, no masses or abnormal calcifications were observed in the left breast. Furthermore, no enlarged axillary lymph nodes were detected on either side, and the skin and nipples were found normal. The findings were classified as BI-RADS category 4B.

Magnetic Resonance Imaging (MRI)

MRI indicated abnormal glandular shape in the upper and lower outer quadrants of the right breast, and a nodule measuring about 1.2 × 0.9 cm was observed in the upper outer quadrant. The nodule had rough edges and uneven enhancement. It showed isointensity on T1-weighted images, hyperintensity on T2-weighted images, and isointensity on diffusion-weighted imaging. The time-signal curve revealed a rapid-rising plateau pattern (Fig. 4). No clear enlarged lymph nodes were identified in the bilateral axilla. In conclusion, the nodule in the outer upper quadrant of the right breast was categorized as BI-RADS category 4.

An ultrasound-guided core needle biopsy was conducted on the center of the mass presented in Fig. 2 of the right breast. Pathological examination indicated ductal epithelium and lobular tissue hyperplasia, along with stromal cell and fibrous tissue hyperplasia in certain areas, some of which showed hyaline degeneration. Furthermore, active cell proliferation was observed in some regions.

Additionally, due to inconsistency between imaging results and biopsy findings, a local extended resection of the lesion in the right breast was performed under general anesthesia on 5 November 2021. Rapid frozen pathology revealed adenosis of the breast, along with focal irregular fibrous tissue and smooth muscle tissue hyperplasia.

Paraffin Pathology Findings

The diagnosis was DTF with breast adenopathy. The tumor had no capsule, displacing surrounding breast tissue, and grew around some glandular ducts. The tumor cells exhibited mild abnormality with localized collagenization and partial infiltration of surrounding adipose tissue (Fig. 5). No clear tumor cells were found at the upper, lower, inner, outer, or bottom margins marked during surgery.

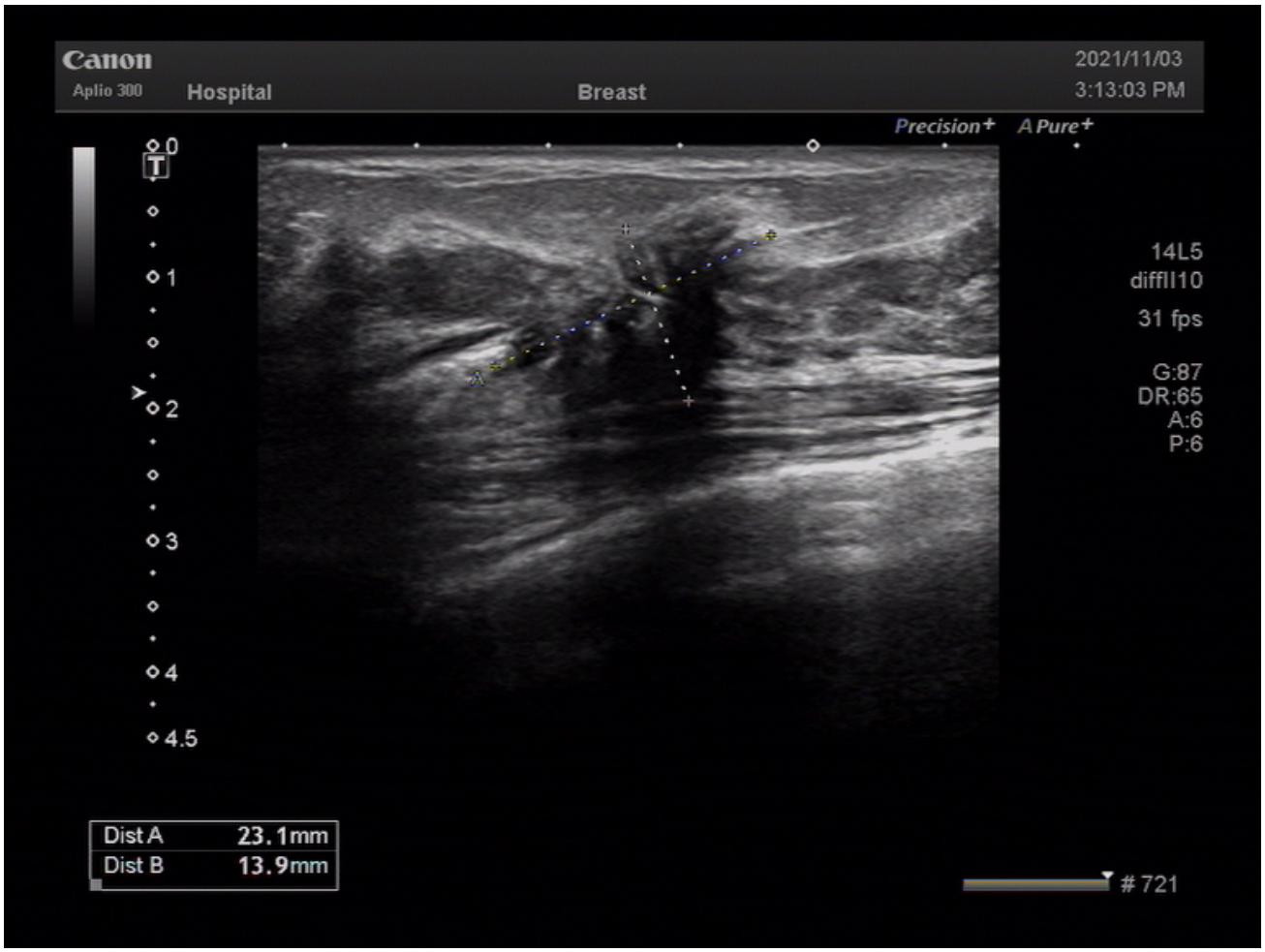

Fig. 2. Ultrasound indicated a 23.1 × 13.9 mm irregularly shaped hypoechoic mass with unclear margins and viable blood flow adjacent to it.

Immunohistochemistry Findings

Ki 67 (+2%), Calponin (+), Desmin (focal+), B-catenin (membrane+), Estrogen Receptor (ER) (scattered+), α-Smooth Muscle Actin (SMA) (section+), CD34 (–), CKpan (–), S-100 (–), Signal Transducer and Activator of Transcription 6 (STAT6) (–) (Fig. 6).

Furthermore, the patient did not receive postoperative radiotherapy or chemotherapy and her breast color ultrasound Doppler was reviewed annually. Notably, no recurrence or metastasis was observed during the 12-month follow-up period after surgery.

Discussion

The pathogenesis of DTF remains unclear. Some cases have been linked to previous trauma or breast surgery [1,2]. Recent case reports have shown the occurrence of DTF after breast prosthesis reconstruction, but it has not been confirmed whether breast implants are associated with the development of DTF [2]. In this case, the patient had no previous history of breast trauma and surgery, and the etiology remains unknown.

DTF primarily occurs in women, though it can occasionally occur in men [1,3–5]. The median age of onset is 46.6 years, ranging form15.5 to 92.2 years [5]. The incidence is roughly similar between the left and right breasts, with few cases involving both breasts at the same time [5,6]. The initial symptom of DTF is a palpable breast mass, occasionally accompanied by nipple discharge, though some asymptomatic patients are observed during routine physical examination [1]. The mass generally presents as hard, with unclear boundaries and irregular shape, sometimes causing a dimple sign on the surface of the skin [7,8]. Nipple retraction is commonly seen in tumors located close to the nipple. If the tumor invades the chest wall, the mass may become fixed and immobile. The imaging features of DTF are similar to those of breast cancer, often resulting in a BI-RADS classification of 4 or 5, which frequently leads to misdiagnosis of breast cancer.

On ultrasound images, most of them showed hypoechoic with irregular shape and unclear boundary, and some cases showed attenuation of acoustic shadow behind the lesions [7]. DTF is generally categorized as BI-RADS category 4 or 5. On mammography, DTF often manifests as a

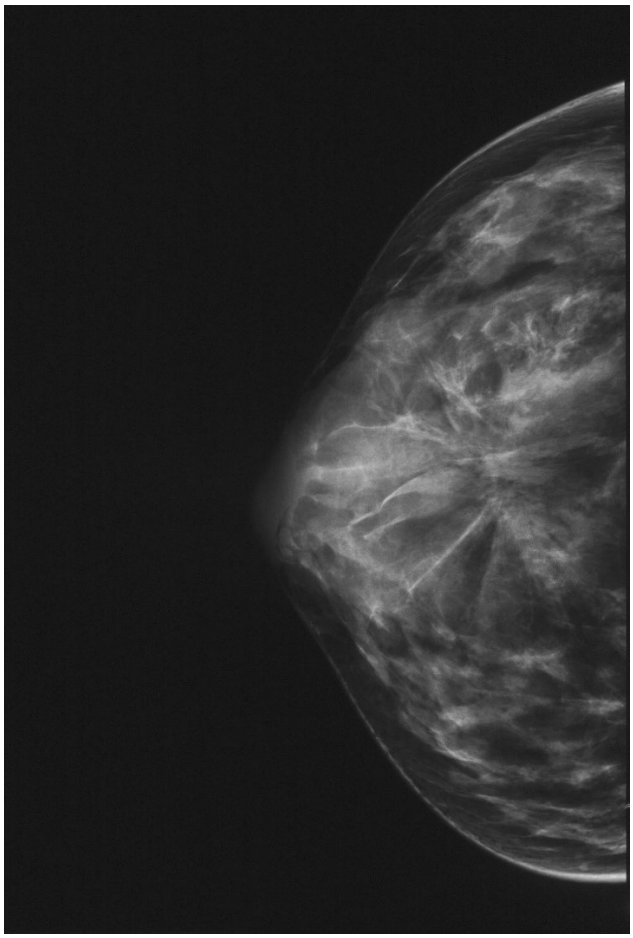

Fig. 3. Mammography showed a structural distortion, radial changes, and skin thickening in the upper outer quadrant of the right breast.

high-density mass shadow with irregular shape and unclear boundary, which is easy to be confused with breast cancer, and sometimes also manifests as local structural distortion, invading the muscle tissue of the chest wall [9]. In the present case, mammography revealed localized glandular structural disorder with radial changes, which differs from the imaging features reported in the previous studies. In some cases, DTF arises from the underlying chest wall rather than the breast parenchyma, and MRI can help discern whether the tumor is within the breast tissue or arises from the pectoralis muscle, with the breast displaced anteriorly [10]. MRI can show mass shadows or structural disorders. The lesions are hypointense or isointense on T1-weighted images (WI), hyperintense on T2WI, and the time signal curve is mostly a rapid-rising plateau type [1,11].

Ultrasound-guided fine needle aspiration cytology is usually insufficient for diagnosing DTF [12,13]. Ultrasound-guided core needle biopsy (CNB) can sometimes diagnose DTF through histomorphological characteristics and immunohistochemical staining. Lorenzen J *et al*. [7] reported that 86% of 15 CNB cases suggested DTF, though many cases could only distinguish between benign and malignant lesions, while others were misdiagnosed as phyllodes tumors [12]. Therefore, surgical excision biopsy is often required to obtain sufficient tissue for accurate histopathological assessment. Histopathologically, DTF is manifested as uniform, elongated, spindle-shaped cells forming interwoven, arcuate bundles, with infiltration into adjacent tissues, such as mammary ducts/lobules, adipose tissue, and skeletal muscle [13]. The nuclei are small, lightly stained, and may contain a few small nucleoli, with mitoses usually absent or rare, and the cytoplasm is eosinophilic. The interstitial collagen is abundant and varies morphologically, sometimes resembling keloid-like scars. Although the stroma of early lesions may appear myxoid, lesions of longer duration may have calcifications or chondro-osseous metaplasia. Lymphocytes often cluster at the periphery of the lesion. Nuclear positivity for βcatenin is seen in 76%–80% of cases [14]. Most tumors express smooth muscle markers like α-SMA and calponin, while Desmin is occasionally expressed in a few tumor cells.

The differential diagnosis of DTF includes nodular fasciitis, neurofibroma, fibroadenoma, phyllodes tumor, lipomatous myofibroblastoma, and low-grade fibromatosis-like spindle cell metaplastic carcinoma of the breast. In nodular fasciitis, fibroblasts are usually plumper, mitotic figures are abundant and inflammatory cells are diffusely dispersed at the periphery without conspicuous lymphoid infiltrates [15]. Neurofibromas are characterized by neoplastic cells with tapered, buckled nuclei, and the stromal collagen often shows a “shredded carrot” appearance [16]. Fibroadenomas and phyllodes tumors are usually distinguishable by the identification of an epithelial component [17]. Low-grade fibromatosis-like spindle cell metaplastic carcinoma can be microscopically indistinguishable from DTF but shows at least focal cytokeratin expression [17].

In contrast, DTF typically expresses α-SMA and calponin without cytokeratin, CD34 or S-100 expression. Lipomatous myofibroblastoma mimics DTF due to its pseudo infiltrative fibromatosis-like pattern caused by the admixture of the spindle cell with the lipomatous [18,19]. Unlike DTF, lipomatous myofibroblastoma has pushing borders, and the fatty component is not the result of the entrapment of surrounding adipose tissue [18,19]. Although both lipomatous myofibroblastoma and DTF share expression of α-SMA, the former is also positive for desmin, CD34, and estrogen/progesterone receptors, which are negative in the latter [18,19]. Notably, nuclear positivity for β-catenin could not specifically differentiate DTF from metaplastic carcinoma and phyllodes tumors.

Although European Society for Medical Oncology (ESMO) proposed in a consensus on the diagnosis and treatment of sporadic DTF, the first-line treatment for DTF after diagnosis by CNB is watch & wait, after disease progression, different treatments are selected according to the location of the lesion [20]. However, it does not specify treatment

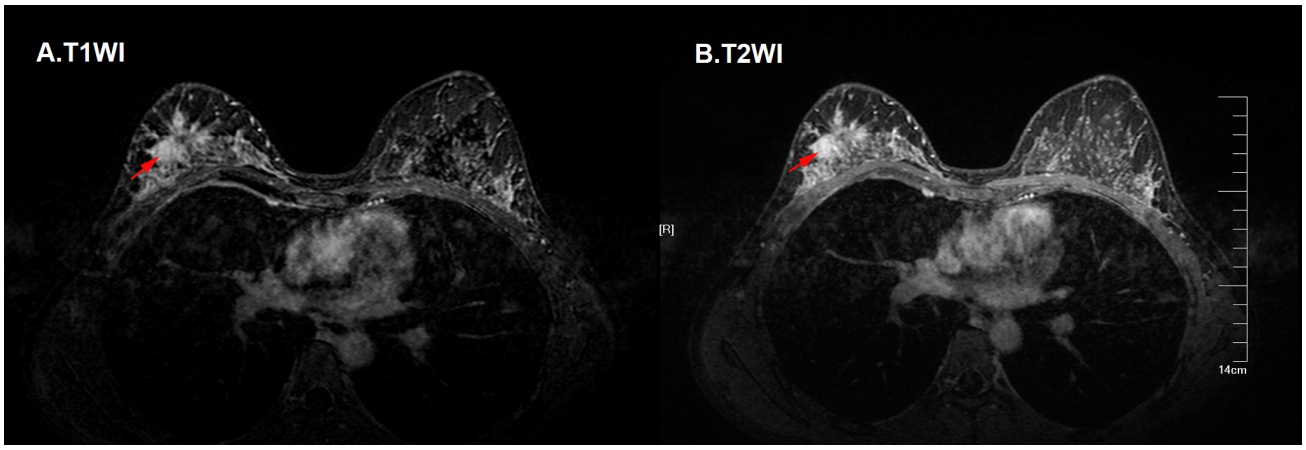

Fig. 4. (A) T1- and (B) T2-weighted magnetic resonance imaging indicated an approximately 1.2 × 0.9 cm nodule in the upper outer quadrant of the right breast, with irregular edges and uneven enhancement. The arrow indicates the location of the lesion. The time-signal curve showed a rapid-rising plateau pattern. T1WI, T1-weighted images; T2WI, T2-weighted images.

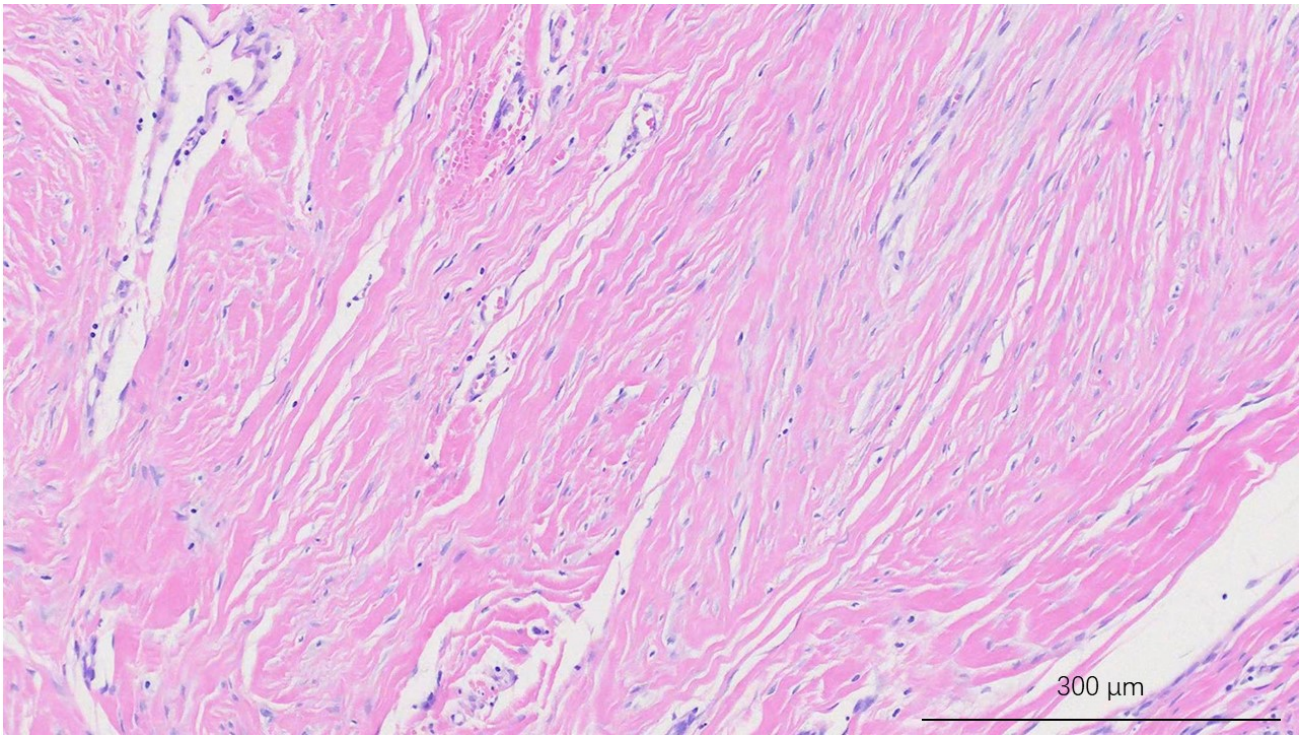

Fig. 5. Paraffin-embedded pathological sections showed that tumor cells were spindle-shaped and atypic, with abundant interstitial collagen fibers (Hematoxylin-eosin staining 200×).

strategies for DTF. A multicenter, retrospective study by Duazo-Cassin L *et al*. [4] included 63 cases of DTF, with a median follow-up of 24.9 months in the surgery group (surgery group) and 42.2 months in the active surveillance group (active surveillance group). Recurrence rates were 8.7% in surgery group and 11.8% in active surveillance group [4].

This study supports an initial nonsurgical approach to DTF, with surgery based on tumor growth, aligning with current ESMO recommendations [4]. However, differentiating DTF from breast cancer based on imaging can be challenging, as CNB may not provide an accurate diagnosis. As a result, inconsistencies between imaging results and pathological observations are common and often require surgery to rule out breast cancer. While DTF shows locally invasive growth, it does not lead to distant metastasis. Duazo-Cassin L *et al*. [4] observed no recurrence even when the surgical margin did not reach R0 resection. However, achieving negative margins (R0 resection) remains crucial to reduce the risk of local recurrence. In this case, the possibility of malignant tumor was considered due to the results of various imaging examinations. Although the results of

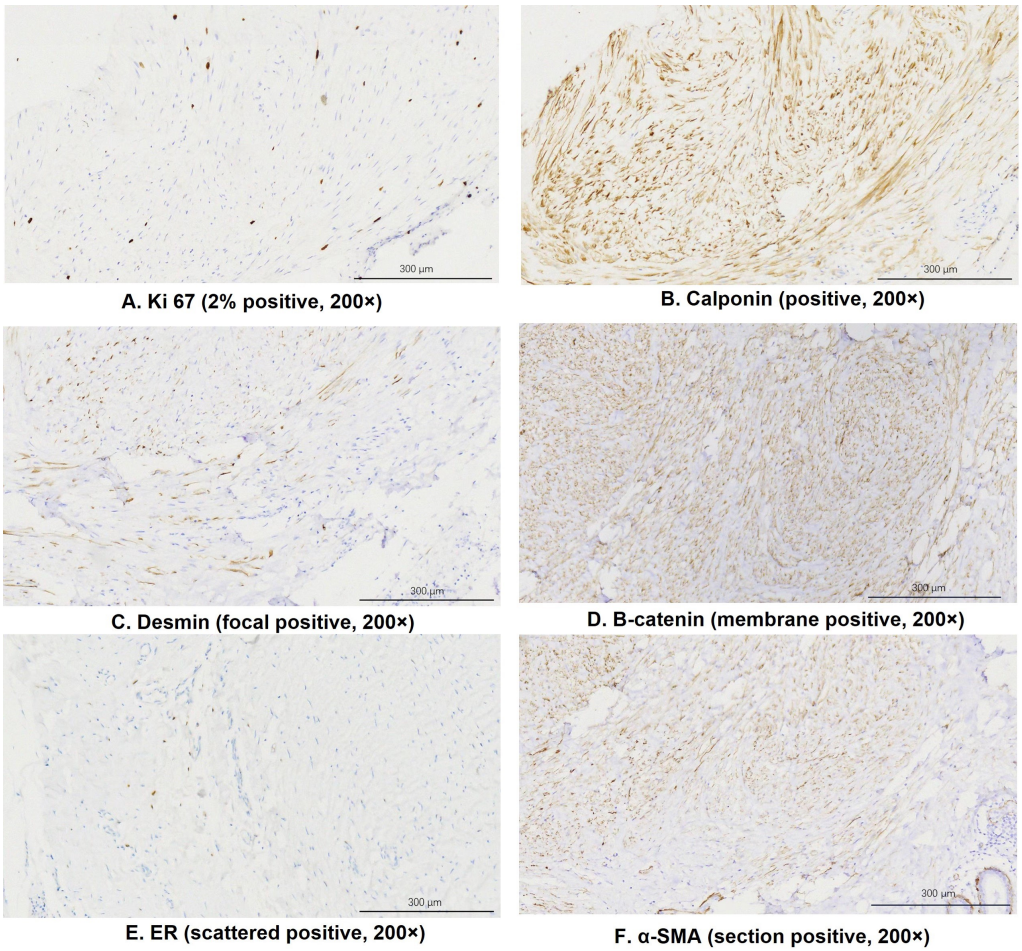

Fig. 6. Immunohistochemistry of desmoid-type fibromatosis of the breast: (A) Ki 67 (+2%), (B) Calponin (+), (C) Desmin (focal+), (D) B-catenin (membrane+), (E) ER (scattered+), (F) α-SMA (section+), CD34 (–), CKpan (–), S-100 (–), STAT6 (–). ER, Estrogen Receptor; SMA, Smooth Muscle Actin; STAT6, Signal Transducer and Activator of Transcription 6.

CNB suggested benign tumor, open surgical biopsy was performed due to the inconsistency between imaging examination and pathological results. Since the mammography indicated disordered gland structure and unclear lesion boundaries, local extended excision was performed. The intraoperative frozen section revealed negative margins, resulting in the conclusion of the procedure.

Although there are case reports of DTF cases treated with endocrine therapy, chemotherapy, radiotherapy, tyrosine-kinase inhibitors (TKIs) and interferon α2b, some of them have achieved good results [3,16]. However, these treatments are not supported by data from randomized trials. Therefore, postoperative adjuvant therapy was not performed in this case, and the patient was asked to review the breast ultrasound every 3 months for 2 years. The follow-up for the first year showed no recurrence.

Conclusion

In this case, preoperative imaging suggested malignancy, while biopsy pathology indicated a benign lesion. Subsequent surgical exploration confirmed DTF. This scenario underscores a clinical approach where complete resection is pursued when imaging and initial biopsy are inconclusive, with definitive treatment decisions based on post-resection pathology. Intraoperative frozen section analysis revealed negative margins, obviating the need for further extensive resection or adjuvant therapy. No recurrence or metastasis was observed during one-year follow-up post-surgery. This case highlights the significance of surgical management guided by thorough pathological evaluation in ambiguous breast lesions.

Availability of Data and Materials

The datasets used and analysed during the current study are available from the corresponding author.

Author Contributions

WYR: Designed the study, collected and analyzed data, and was primarily responsible for writing the manuscript. HS: Participated in data analysis, collected data and research discussions, and contributed to the writing of the manuscript. WG, XMF, YHD, JLJ and APS participated

in data collection and contributed significantly to the critical editing and revision of the manuscript. DW: Oversaw the entire research project, supervised and participated data collection and analysis, interpreted and discussed the results, and provided important contributions and revisions to the manuscript. All authors read and approved the final manuscript. All authors have participated sufficiently in the work and agreed to be accountable for all aspects of the work.

Ethics Approval and Consent to Participate

This study fully complies with the Declaration of Helsinki. This study uses a retrospective design, ensuring patient privacy by excluding any identifiable personal data. The analysis includes the basic clinical characteristics, imaging results, surgical data, and pathological findings of this patient. Informed consent was obtained from the study participant, and approval was granted by the Ethics Committee of the First Hospital of Jilin University (ethics approval number: 2024-999).

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Conflict of Interest

The authors declare no conflict of interest.

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