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Recurrent Desmoid-Type Fibromatosis of the Breast in a Young Woman With a Monoallelic MUTYH Germline Variant

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Abstract

Desmoid-type fibromatosis of the breast is an uncommon, low-grade mesenchymal neoplasm with benign histologic features, characterized by locally aggressive growth and a notable tendency for local recurrence. Clinical management is challenging owing to frequent positive surgical margins, a propensity for recurrence, and the absence of standardized treatment guidelines. We report the case of a 34-year-old woman who underwent mastectomy for recurrent breast desmoid-type fibromatosis, in whom a heterozygous germline *MUTYH* (MutY DNA glycosylase) variant was identified through hereditary cancer panel testing. This case highlights the potential value of comprehensive pathologic assessment and selective germline genetic testing in patients with recurrent breast fibromatosis, which may offer further insight into tumor biology and inform individualized management strategies.

Categories: General Surgery
Keywords: breast disease, desmoid-type fibromatosis, germline mutation, germline testing, mutyh mutation

Introduction

Desmoid-type fibromatosis, historically referred to as aggressive fibromatosis, is a rare mesenchymal neoplasm originating from musculoaponeurotic tissues and characterized by locally infiltrative growth in the absence of metastatic potential [1]. Although desmoid-type fibromatosis most commonly affects the mesentery, abdominal wall, and extremities, breast involvement is uncommon, accounting for approximately 4% of extra-abdominal cases [2,3]. It typically affects young adults aged between 20 and 44 years and demonstrates a marked female predominance, with an estimated incidence ranging from two to six cases per million individuals [4]. Overall, it represents only about 0.2% of all solid breast tumors, presenting a significant diagnostic challenge because of its close clinical and radiologic resemblance to breast carcinoma [3]. This diagnostic challenge is further compounded by the high rate of incorrect initial diagnoses, as the lesion is frequently misinterpreted as invasive carcinoma owing to its spiculated mammographic appearance, hypoechoic sonographic features with posterior acoustic shadowing, and morphologic overlap with spindle cell neoplasms [5,6]. At the molecular level, desmoid-type fibromatosis is characterized by dysregulation of the adenomatous polyposis coli (APC)/β-catenin signaling pathway, most commonly associated with somatic *CTNNB1* mutations and *APC* gene alterations in both sporadic and familial adenomatous polyposis (FAP)-related cases [2]. However, high-quality evidence correlating specific mutational profiles with local recurrence risk, surgical margin status, or adjuvant treatment decision-making remains limited and heterogeneous. Recent studies suggest that germline variants beyond *APC*, including *MUTYH*, may contribute to tumor susceptibility through indirect modulation of the Wnt/β-catenin pathway, although their role remains poorly defined [7-9].

In this report, we describe the clinical course, mutational findings, and management of a patient with recurrent desmoid-type fibromatosis of the breast, discussed in the context of the current literature.

Case Presentation

A 34-year-old woman, gravida 1 para 1, presented with a painless, recurrent mass occupying most of the right breast that had been progressively enlarging over the preceding four months. The patient's medical history was notable for prior breast surgery performed three years earlier for a mass in the same breast. Histopathologic evaluation at that time described the lesion as a stromal tumor without a definitive diagnosis. Detailed information regarding previous diagnostic assessments, therapeutic decisions, and follow-up could not be fully retrieved.

The patient's family history was notable for malignancy, including a paternal aunt who was diagnosed as having breast cancer at age 42 years and another paternal aunt with a history of a head and neck tumor of

unclear etiology. Her medical history was otherwise unremarkable. Notably, the patient reported a nonsurgical blunt traumatic event involving the same breast during exercise before the initial surgery.

The patient had previously undergone a segmental mastectomy due to discordant radiologic, clinical, and pathologic findings. A histopathologic examination revealed a mesenchymal tumor of uncertain subtype with close surgical margins (<1 mm), and no adjuvant therapy was administered. Three years later, the patient presented again with a recurrent mass localized to the site of the prior incision.

On physical examination, a firm, immobile, painless mass measuring approximately 10 × 8 cm was palpated, extending toward the nipple, and associated with overlying skin retraction (Figure *1*).



FIGURE 1: Clinical appearance of the breast lesion.

Preoperative clinical photograph demonstrating a visible contour deformity and skin retraction of the right breast. The lesion margins are marked on the skin to delineate the extent of the palpable mass before the surgical intervention.

No palpable axillary or supraclavicular lymphadenopathy was detected bilaterally. Bilateral mammography demonstrated moderately dense breasts with heterogeneous fibroglandular tissue distribution (pattern C). An asymmetric density was identified in the right breast on mediolateral oblique views, and an incidental asymmetric density in the outer quadrant of the left breast on craniocaudal views; neither finding corresponded to the known tumor, and neither was confirmed as suspicious on ultrasonography. Notably, the primary mass in the right inferomedial quadrant was not visualized on mammography but was clearly identified on ultrasonography and magnetic resonance imaging (Figures *2A*-*2D*).

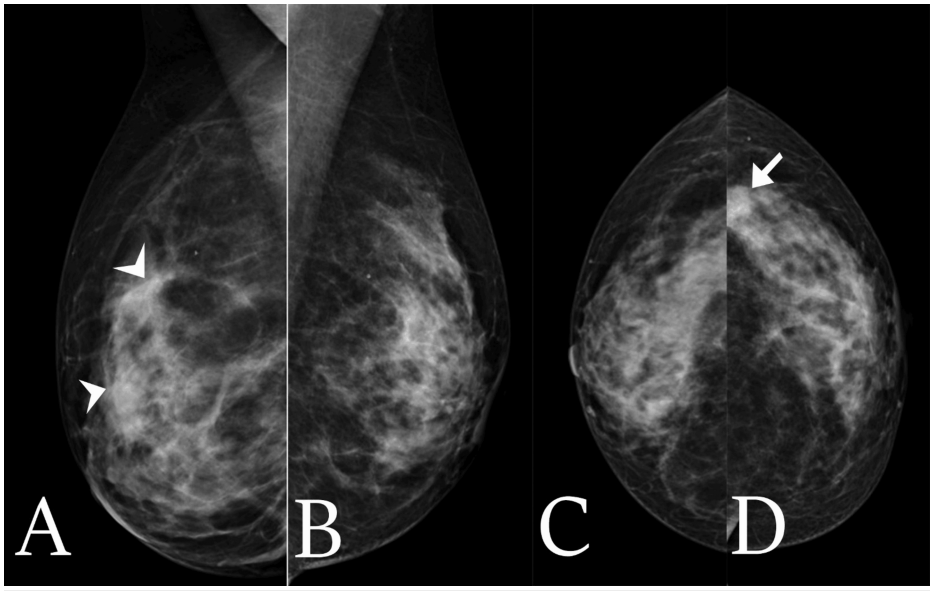


FIGURE 2: Mammographic findings.

Bilateral mammography demonstrating moderately dense breasts with heterogeneous fibroglandular tissue distribution. Mediolateral oblique views (A, B) show an asymmetrical density in the right breast (arrowheads), whereas craniocaudal views (C, D) reveal an incidental asymmetrical density in the left breast (arrow). Neither finding corresponded to the known tumor, and targeted ultrasonography of the corresponding regions showed no suspicious abnormality. The primary mass located in the right inferomedial quadrant was not visualized on mammography but was clearly identified on ultrasonography and magnetic resonance imaging.

Breast ultrasonography revealed a heterogeneous hypoechoic solid mass measuring approximately 110 × 30 mm in the lower portion of the right breast, extending along the pectoral fascia and demonstrating increased vascularity on Doppler examination. Magnetic resonance imaging revealed a heterogeneously enhancing mass lesion measuring approximately 77 × 41 mm in the lower quadrant of the mid-right breast, located in close anterior proximity to the pectoral muscle with indistinct margins, suggestive of residual or recurrent disease (Figures *3A*-*3C*).

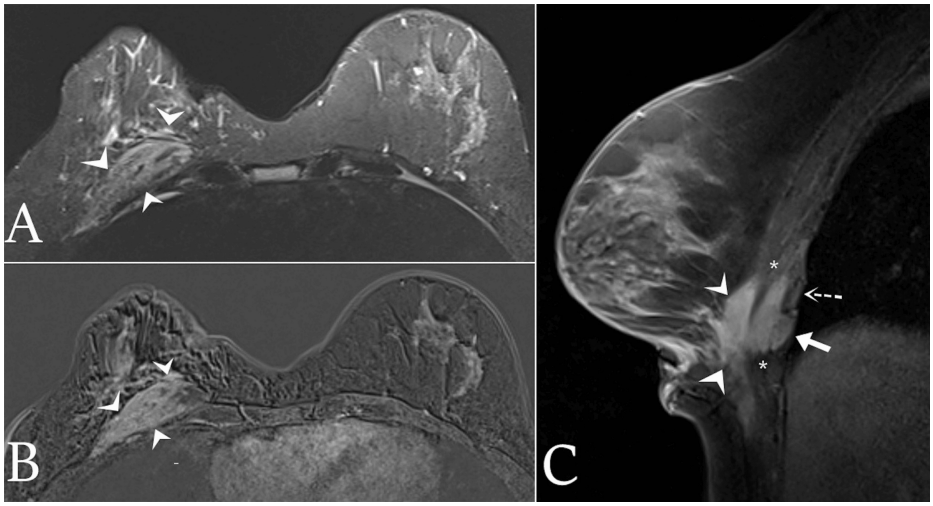


FIGURE 3: Breast magnetic resonance imaging findings.

(A) Axial fat-suppressed T2-weighted image demonstrating a hyperintense, irregular lesion in the right breast (arrowheads).

(B) Axial contrast-enhanced T1-weighted image showing heterogeneous enhancement of the lesion (arrowheads) with infiltration of the anterior thoracic wall.

(C) Sagittal contrast-enhanced T1-weighted image demonstrating tumor extension (arrowhead) into the pectoralis muscles (asterisks) and intercostal muscle invasion (solid arrow). The dashed arrow indicates the fourth rib.

Ultrasound-guided core needle biopsy revealed a mesenchymal tumor with morphologic features identical to those of the previous specimen. Positron emission tomography-computed tomography revealed a hypermetabolic, irregular mass measuring 82 × 67 × 27 mm with a maximum standardized uptake value of 3.20, consistent with tumor recurrence. The variation in lesion dimensions across clinical examination and imaging modalities reflects differences in measurement planes, imaging techniques, and the characteristically infiltrative growth pattern of desmoid-type fibromatosis, rather than true inconsistency.

Subsequently, the patient underwent a total right mastectomy with partial pectoralis muscle excision. Histopathologic examination revealed a spindle cell proliferation arranged in long, infiltrative fascicles extending into the dermis and underlying skeletal muscle. The tumor cells were cytologically bland, with no significant atypia, mitotic activity, or necrosis, which is consistent with desmoid-type fibromatosis. The lesion involved multiple posterior surgical margins. Immunohistochemical analysis demonstrated weak nuclear positivity for β-catenin, diffuse positivity for smooth muscle actin and calponin, focal weak positivity for desmin, weak CD10 expression, and negativity for pan-cytokeratin, estrogen, and progesterone receptors. The Ki-67 proliferation index was low (1-2%). The lesion was classified as an extra-abdominal desmoid-type fibromatosis arising from the pectoral fascia based on the infiltrative growth pattern, characteristic spindle cell morphology, immunohistochemical profile, and low proliferative activity (Figures *4A*-*4D*).

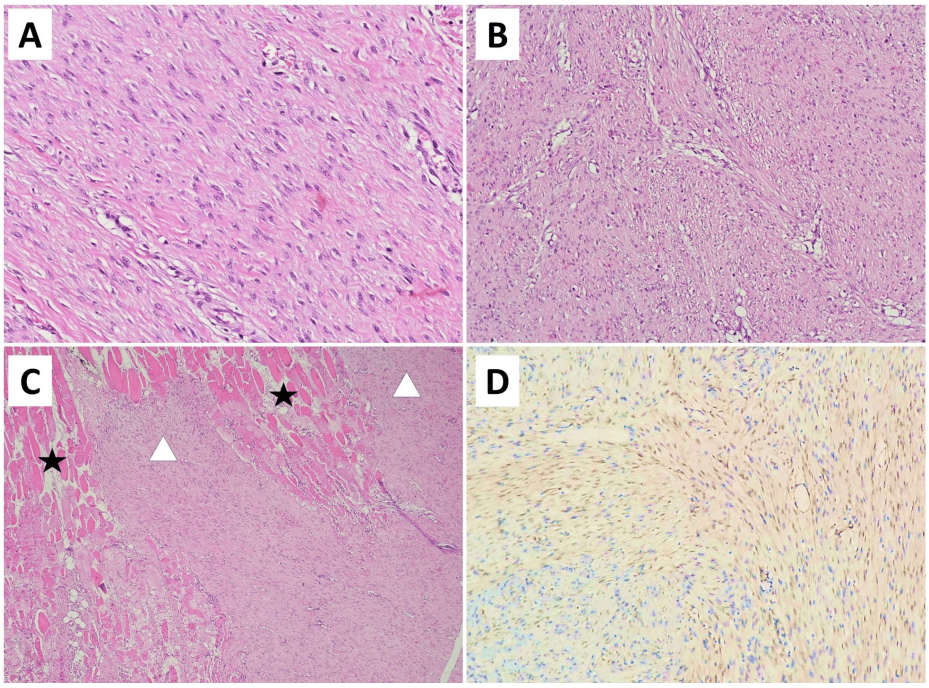


FIGURE 4: Histopathologic and immunohistochemical features of desmoid-type breast fibromatosis.

(A) The lesion comprises cytologically bland spindle cells arranged in long fascicles within a collagenous stroma, without significant cytologic atypia or mitotic activity (hematoxylin and eosin, ×400).

(B) Low-power view illustrating the characteristic infiltrative growth pattern of the tumor (hematoxylin and eosin, ×200).

(C) Tumor infiltration (white triangles) into adjacent skeletal muscle fibers (black stars) (hematoxylin and eosin, ×100).

(D) Immunohistochemical staining demonstrating weak nuclear positivity for β-catenin in tumor cells, supporting the diagnosis of desmoid-type fibromatosis (×400).

Germline genetic testing using a hereditary cancer panel with whole-exome sequencing (GeneMind WES solution) identified a heterozygous protein-truncating variant in exon 13 of the *MUTYH* gene (NM_001048174.2; c.1160_1167dup; p.Lys390PhefsTer37). This was the sole pathogenic variant detected among the cancer-predisposition genes analyzed. The variant allele frequency was 50%, consistent with heterozygous carriage. The variant resulted in a frameshift. Although it is listed in ClinVar (Variation ID: 1452003), it has not been previously reported in association with desmoid-type fibromatosis, and detailed phenotypic data remain lacking. To our knowledge, this variant has not been previously described in this clinical context in either germline or somatic cancer genomic studies. The finding was further confirmed through visual inspection using Integrative Genomics Viewer (Figure *5*).

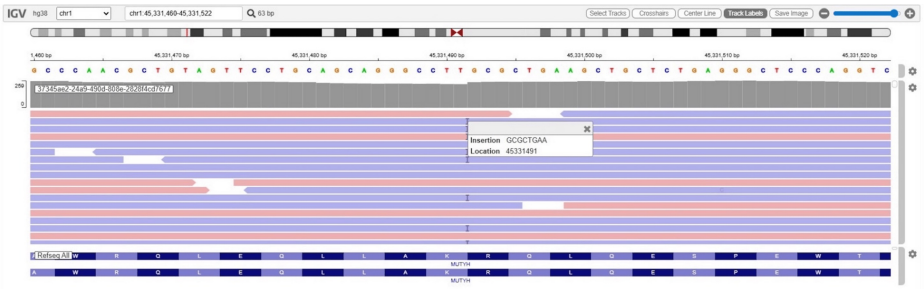


FIGURE 5: Integrative Genomics Viewer (IGV) visualization of the identified MUTYH mutation.

IGV screenshot demonstrating the heterozygous *MUTYH* c.1160_1167dup (p.Lys390PhefsTer37) variant, with consistent variant-supporting reads in both forward and reverse directions, confirming the presence of the mutation.

No additional pathogenic variants were identified in the remaining genes, and the patient was classified as a monoallelic *MUTYH* carrier.

The patient was evaluated by a multidisciplinary tumor board because of the presence of positive surgical margins, and adjuvant radiotherapy was recommended. Adjuvant radiotherapy was administered to the chest wall to a total dose of 50 Gy, followed by a 10 Gy boost to the tumor bed. The treatment was well tolerated, and at the most recent follow-up, 10 months after completion of radiotherapy, the patient had no clinical or radiological evidence of local recurrence (Figure *6*).

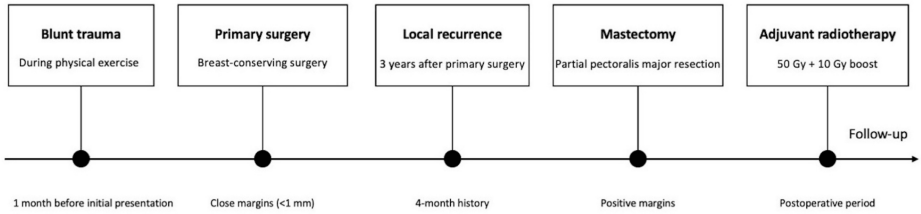


FIGURE 6: Clinical timeline of the patient's disease course.

Initial blunt trauma during exercise one month before the first clinical presentation (~3 years before the current presentation); primary segmental mastectomy with close margins (<1 mm); current presentation with a four-month history of recurrent breast mass; total mastectomy with partial pectoralis muscle excision (positive margins); and adjuvant chest wall radiotherapy (50 Gy + 10 Gy boost).

The figure was created using Microsoft PowerPoint.

Discussion

Desmoid tumors are rare fibroblastic neoplasms characterized by locally infiltrative growth and a high propensity for local recurrence [2]. Their clinical course is highly unpredictable, ranging from spontaneous regression to progressive, locally aggressive disease. Management of desmoid-type fibromatosis involves a spectrum of therapeutic strategies, including active surveillance, surgical resection, systemic therapy, and radiotherapy, with treatment decisions tailored to tumor location, biological behavior, and patient-specific factors [10,11].

In the present case, the initial surgical intervention before recurrence resulted in close surgical margins (<1 mm), while subsequent resection demonstrated positive margins. Margin positivity is common in desmoid-type fibromatosis and does not reliably predict recurrence [12]. Although conservative strategies such as active surveillance and watchful waiting have gained significant traction, no consensus has been reached regarding optimal management strategies. Treatment decisions are guided by symptom burden, tumor behavior, patient age, and the morbidity associated with extensive procedures, particularly in chest wall involvement.

Evidence supporting non-operative management has expanded substantially. In a French multicenter study, active surveillance yielded favorable outcomes even in recurrent disease, with disease stabilization observed

in approximately 53% of patients, spontaneous regression in 35%, and delayed surgery required in only 11.8% [13]. Similarly, in a large multicenter cohort, Salas et al. identified age, tumor size, and anatomical location as independent predictors of progression-free survival, whereas surgical margin status did not significantly affect outcomes [14]. These findings demonstrate that surgical radicality is not the primary determinant of recurrence. Nevertheless, achieving clear margins can be particularly challenging in large tumors with chest wall involvement. In such cases, additional surgery may be avoided due to substantial morbidity, as was the case with our patient.

Adjuvant radiotherapy is widely used to improve local control and reduce the need for reoperation, particularly in recurrent disease with positive or close margins, although concerns persist regarding long-term toxicity, especially in younger patients [4,12,15]. Despite increasing evidence supporting active surveillance in selected patients, adjuvant radiotherapy was favored in the present case. This decision was driven by multiple high-risk features, including young age, first recurrence, large tumor size, chest wall involvement, and positive surgical margins. Collectively, these factors indicated a substantial risk of further locoregional progression, supporting a more aggressive local control strategy in accordance with current management recommendations [12,14-16]. In this context, radiotherapy may be considered as a secondary or adjuvant option in selected high-risk cases of chest wall desmoid-type fibromatosis, particularly when re-excision would entail prohibitive morbidity [15,16]. The decision to administer radiotherapy was made following multidisciplinary tumor board discussion, based on the specific clinical context and current evidence. The patient was fully informed of the potential long-term sequelae and elected to proceed with adjuvant therapy.

Breast desmoid-type fibromatosis is associated with surgical trauma, hormonal influences, and genetic susceptibility [2]. A well-established association exists with FAP, particularly the Gardner variant, in which desmoid tumors represent a prominent extracolonic manifestation [4]. In the present case, there was no personal or family history suggestive of FAP or *MUTYH*-associated polyposis. Importantly, monoallelic *MUTYH* carriage alone does not imply a diagnosis of *MUTYH*-associated polyposis. Non-surgical blunt trauma has been described as a potential triggering factor in approximately 22% of trauma-associated cases, including sports-related injuries, whereas antecedent trauma of any type has been reported in approximately one-third of sporadic desmoid tumors [17]. Consistent with these observations, the patient reported a history of nonsurgical blunt trauma to the affected breast sustained during exercise before the initial presentation.

The accurate diagnosis of desmoid-type fibromatosis relies on thorough pathologic evaluation; however, it remains challenging because of interpretative variability and pre-analytical, technical, and clinical factors. The key clinical, radiologic, histopathologic, and genetic features distinguishing desmoid-type fibromatosis from invasive ductal carcinoma are summarized in Table *1*.

Feature	Desmoid-type fibromatosis	Invasive ductal carcinoma
Age and sex	Young women (20–44 years) [4]	Any age; 50–70 years [5]
Clinical presentation	Firm, painless mass; skin retraction; typically no lymph node involvement [1]	Firm mass ± skin/nipple changes; lymph node involvement possible [5]
Mammography	Ill-defined density; may be occult; typically lacks suspicious microcalcifications [1,3]	Spiculated/irregular mass; microcalcifications common [5]
Ultrasonography	Hypoechoic, heterogeneous mass with posterior shadowing; infiltrative margins; increased vascularity on Doppler [1,2]	Hypoechoic irregular mass with angular margins; posterior shadowing; variable vascularity [5]
MRI features	Heterogeneous enhancement; infiltrative margins; fascial involvement; variable T2 signal intensity (low to high), fascial tail sign [2]	Variable enhancement; mass effect; high T2 signal [5]
Histology	Bland spindle cells; no atypia, mitotic activity, or necrosis [2]	Neoplastic epithelial cells with nuclear pleomorphism; mitoses and necrosis possible [5]
Immunohistochemistry	Nuclear β-catenin, SMA positive; ER/PR/CK negative [3]	CK positive; ER/PR/HER2 variable; β-catenin negative [5]
Metastatic potential	Locally aggressive, infiltrative; no metastatic potential [4]	Yes (regional and distant) [5]
Genetic associations	Somatic *CTNNB1* mutations; germline *APC* mutations (FAP-associated cases) [4]	*BRCA1/2*, *TP53* alterations; *HER2* amplification [5]

TABLE 1: Comparison of clinical, radiologic, histopathologic, and genetic features between desmoid-type fibromatosis and invasive ductal carcinoma.

SMA: smooth muscle actin; ER: estrogen receptor; PR: progesterone receptor; CK: cytokeratin; HER2: human epidermal growth factor receptor 2; FAP: familial adenomatous polyposis; MRI: magnetic resonance imaging

In our case, neither the initial core needle biopsy nor the primary excision specimen allowed definitive diagnosis, which was ultimately established following mastectomy for recurrent disease. Although the morphologic features were suggestive, only weak nuclear β-catenin positivity was demonstrated, which potentially reflects biologic variability or technical limitations. Importantly, fibromatosis-like metaplastic carcinoma should be considered in the differential diagnosis of spindle-cell lesions of the breast; its exclusion in the present case was supported by cytokeratin negativity. The rarity of this entity, low clinical suspicion, and limited access to specialized sarcoma centers may further contribute to diagnostic delays [4].

From a germline perspective, rare breast mesenchymal tumors, particularly those occurring at a young age or demonstrating aggressive local behavior, may warrant evaluation for an underlying hereditary cancer predisposition. Accordingly, germline genetic testing may be especially relevant in young patients and in cases in which a potential hereditary background could influence adjuvant treatment decisions. The mechanistic basis by which a monoallelic *MUTYH* variant might influence mesenchymal tumor behavior warrants consideration. *MUTYH* encodes a DNA adenine glycosylase in the base excision repair pathway, removing adenine misincorporated opposite oxidized guanine [8]. When this repair is compromised, G:C→T:A transversion mutations may accumulate in tumor suppressor genes such as *APC*, promoting β-catenin accumulation, nuclear translocation, and activation of pro-proliferative Wnt target genes [9]. A germline *MUTYH* variant may therefore lower the threshold for β-catenin activation in susceptible mesenchymal tissues [8,9]. Although several reports have described *MUTYH*-associated desmoid tumors, data regarding their anatomical distribution and tumor localization remain limited. Chan et al. described a patient with a monoallelic *MUTYH* mutation who developed multiple neoplasms, including a presacral desmoid tumor and recurrent breast fibroadenomas [18]. Caliendo et al. further demonstrated a higher prevalence of pathogenic *MUTYH* variants among patients with cancer compared with controls and documented cancer recurrence in heterozygous carriers within families harboring biallelic *MUTYH* mutations [7]. Collectively, these findings suggest that, beyond the established role of somatic *CTNNB1* mutations and germline APC alterations, additional germline variants may act as modifying factors that influence tumor behavior and recurrence risk in desmoid-type fibromatosis. In this case, the coexistence of a monoallelic *MUTYH* variant, young age, prior trauma, and recurrent disease supports a multifactorial model in which genetic susceptibility and local environmental factors jointly influence disease behavior. It should be noted that the clinical significance of monoallelic *MUTYH* carriage remains a subject of debate. While biallelic *MUTYH* variants are firmly established as the cause of *MUTYH*-associated polyposis, the

independent contribution of heterozygous carriage to cancer predisposition, particularly to mesenchymal tumor susceptibility, has not been definitively established. Furthermore, although the identified variant is classified as pathogenic based on its protein-truncating nature, its specific role in the pathogenesis of desmoid-type fibromatosis remains to be established. Therefore, the present finding should be interpreted with caution as a potential modifying factor in this individual case rather than as evidence of a generalizable causal relationship or a confirmed disease-causing alteration.

Several limitations of this report warrant acknowledgment. First, the retrospective nature of the initial surgical management prevented complete retrieval of prior imaging, operative, and pathological data, thereby precluding comprehensive reassessment of the primary disease course and initial margin status. Second, the clinical significance of the monoallelic *MUTYH* frameshift variant in desmoid-type fibromatosis cannot be established from a single observation. Third, somatic *CTNNB1* mutational analysis was not performed on the tumor specimen, which limits the ability to definitively characterize the molecular pathway underlying β-catenin activation in this case.

Conclusions

This case highlights the diagnostic and therapeutic challenges of breast desmoid-type fibromatosis, particularly in recurrent disease with positive surgical margins. The identification of a monoallelic *MUTYH* germline variant may represent a modifying factor in tumor behavior; however, a direct causal relationship cannot be established. Comprehensive pathologic evaluation and selective germline genetic testing may provide additional insights to inform individualized management strategies in recurrent or aggressive cases and underscore the need for prospective germline registries and collaborative molecular studies in patients with rare breast mesenchymal tumors.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

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Drafting of the manuscript: Gultekin Ozan Kucuk, Sultan Caliskan, Betul Turan, Mesut Ozturk, Necmiye Gul Temel

Critical review of the manuscript for important intellectual content: Gultekin Ozan Kucuk

Supervision: Gultekin Ozan Kucuk

Disclosures

Human subjects: Informed consent for treatment and open access publication was obtained or waived by all participants in this study. **Conflicts of interest:** In compliance with the ICMJE uniform disclosure form, all authors declare the following: **Payment/services info:** All authors have declared that no financial support was received from any organization for the submitted work. **Financial relationships:** All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work. **Other relationships:** All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

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