Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Desmoid-Type Fibromatosis of the Foot: A Diagnostic Challenge — Case Report and Literature Review

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DOI: https://doi.org/10.36347/sjmcr.2026.v14i05.097 | **Received:** 13.04.2026 | **Accepted:** 21.05.2026 | **Published:** 26.05.2026

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Abstract | **Case Report**

Background: Desmoid-type fibromatosis (DTF) is a rare, locally aggressive but non-metastasizing soft tissue tumor arising from musculoaponeurotic structures. Its occurrence in the foot is exceedingly uncommon, and its varied imaging appearance frequently poses diagnostic difficulties. ***Case Presentation:*** We report the case of a 47-year-old woman with no relevant medical history who presented with a painless swelling of the left foot evolving over several years. Magnetic resonance imaging (MRI) revealed an intra- and supra-aponeurotic mass tracking through the inter-metatarsal spaces, measuring approximately 85 mm in its greatest dimension, demonstrating low signal on T1-weighted sequences, heterogeneous signal on T2-weighted sequences, hyperintensity on STIR sequences, and avid enhancement following gadolinium administration. The initial differential diagnosis included synovial sarcoma and Morton's neuroma. Biopsy followed by surgical excision confirmed the diagnosis of desmoid-type fibromatosis on histopathological examination. The postoperative course was notable for local recurrence, without radiological evidence of aggressive behavior. ***Conclusion:*** DTF of the foot is a rare entity with non-specific imaging features that can mimic more aggressive tumors. A thorough histopathological analysis is mandatory for definitive diagnosis. Given its propensity for local recurrence despite negative surgical margins, long-term follow-up is essential. This case highlights the importance of including desmoid fibromatosis in the differential diagnosis of soft tissue foot masses.

Keywords: desmoid fibromatosis; foot tumor; MRI; soft tissue tumor; local recurrence; musculoaponeurotic neoplasm.

1. INTRODUCTION

Desmoid-type fibromatosis (DTF), also referred to as aggressive fibromatosis or musculoaponeurotic fibromatosis, is a rare, clonally proliferating fibroblastic neoplasm arising from fascial and musculoaponeurotic structures [1]. Despite its histologically benign appearance and the absence of metastatic potential, DTF is characterized by an infiltrative growth pattern and a high propensity for local recurrence, making it a clinically challenging condition [2,3].

DTF has an estimated annual incidence of 2–4 cases per million population and can arise at virtually any anatomical site, with the shoulder girdle, mesentery, and abdominal wall being the most commonly reported locations [4]. Involvement of the foot and ankle is exceedingly rare, accounting for fewer than 5% of all extra-abdominal desmoid tumors, and fewer than 100 cases have been documented in the literature to date [5,6].

The diagnosis of DTF is particularly challenging when it occurs in atypical locations such as the foot. Its imaging characteristics on MRI overlap with those of more aggressive soft tissue neoplasms, including synovial sarcoma, and benign neurogenic tumors such as Morton's neuroma. Histopathological examination with immunohistochemistry remains the gold standard for definitive diagnosis [7].

2. CASE PRESENTATION

A 47-year-old woman with no significant past medical or family history presented to the orthopedic surgery outpatient clinic with a three-year history of a progressive, painless swelling of the left foot. She denied any history of trauma, prior surgery, or use of medications known to predispose to fibromatosis, such as hormonal therapies. There was no family history of familial adenomatous polyposis (FAP) or Gardner syndrome.

Physical examination revealed a firm, non-tender, non-mobile soft tissue mass localized to the dorsal aspect of the left foot, with no skin changes or local erythema. Peripheral pulses were intact, and neurological examination of the lower extremity was normal. No regional lymphadenopathy was detected.

Plain radiographs of the left foot demonstrated soft tissue swelling without cortical erosion or periosteal reaction of the metatarsal bones. No calcifications were identified within the mass.

MRI of the left foot was performed using a 1.5 Tesla system with a dedicated foot coil. The examination revealed an intra- and supra-aponeurotic mass infiltrating along the inter-metatarsal spaces, measuring approximately 85 mm in its greatest axial dimension (Figures 1–4). The lesion demonstrated:

- Iso to low intense signal relative to adjacent skeletal muscle on T1-weighted sequences
- Heterogeneous signal intensity on T2-weighted sequences, with alternating areas of moderate and low signal
- Marked hyperintensity on STIR sequences
- Avid, heterogeneous enhancement following intravenous gadolinium-based contrast agent administration

The mass was seen to infiltrate between the 2nd, 3rd, and 4th inter-metatarsal spaces, with no evidence of osseous destruction or neurovascular encasement. The lesion's insinuating growth pattern and ill-defined margins were the most striking imaging features.

Based on the imaging findings, the primary differential diagnosis included: (1) synovial sarcoma, given the biphasic signal behavior on T2 sequences, large size, and periarticular location; and (2) Morton's neuroma (intermetatarsal plantar neuroma), considered due to the inter-metatarsal distribution of the lesion.

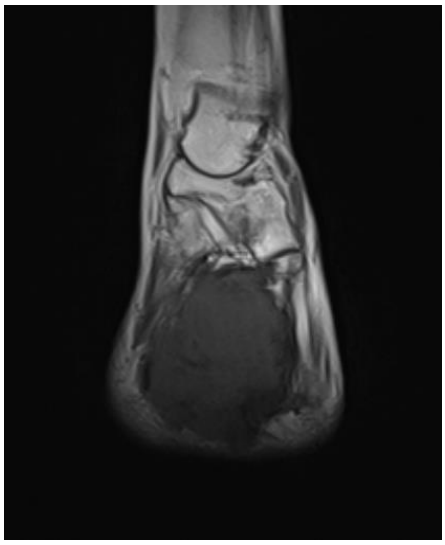

Figure 1: Coronal T1-weighted MRI of the left foot demonstrating a hypointense intra- and supra-aponeurotic mass (arrows) relative to adjacent muscle, occupying the inter-metatarsal spaces. No intrinsic fat signal is identified

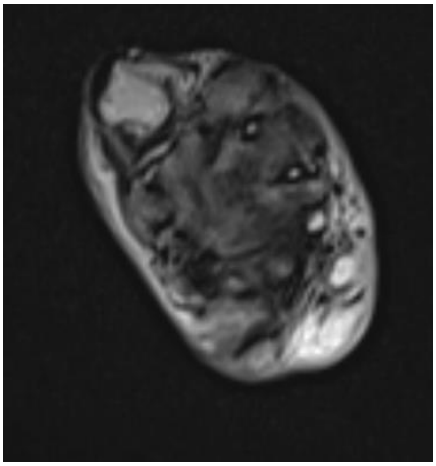

Figure 2: Axial T2-weighted MRI sequence showing the heterogeneous signal of the mass with alternating areas of moderate and low signal intensity (asterisk), consistent with fibrous tissue content interspersed with cellular components, and enlargement of inter metatarsal spaces

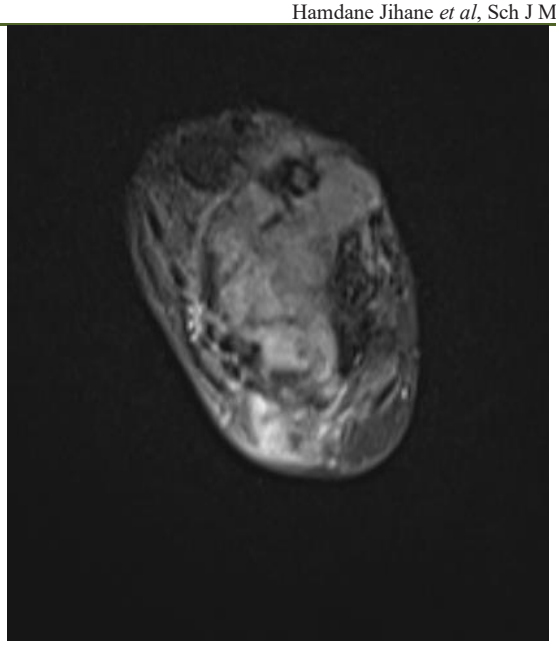

Figure 3: AXIAL STIR MRI sequence demonstrating marked hyperintensity of the lesion, reflecting high free water content associated with edema and myxoid stroma. The lesion measures approximately 85 mm in greatest dimension

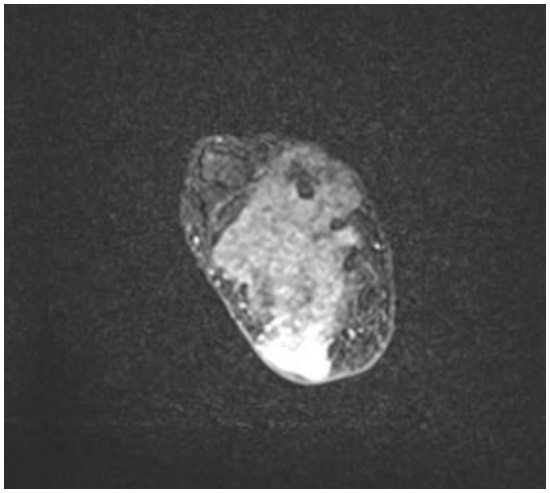

Figure 4: Post-contrast fat-suppressed T1-weighted MRI sequence demonstrating avid and heterogeneous enhancement of the mass following intravenous gadolinium administration (arrows), suggesting high vascularity and active proliferating components

Core needle biopsy under ultrasound guidance was performed. The histopathological analysis of the biopsy specimen revealed a hypocellular proliferation of bland spindle-shaped fibroblasts embedded within an abundant collagenous stroma, with long sweeping fascicles and no significant nuclear atypia or mitotic activity. These features were suggestive of a fibromatosis-type lesion, prompting surgical excision.

En bloc partial surgical excision was performed confirming the diagnosis of desmoid-type fibromatosis.

The immediate postoperative period was uneventful. The patient was enrolled in a structured oncological follow-up program with clinical examination and MRI surveillance every 6 months. At 12 months postoperatively, MRI surveillance demonstrated a local recurrence at the surgical site, presenting as a new soft tissue mass with imaging characteristics similar to the primary lesion. However, no signs of increased aggressiveness — such as osseous invasion, neurovascular encasement, or rapid growth — were noted. Given the absence of symptoms and non-aggressive recurrence pattern, an active surveillance (wait-and-see) approach was adopted in multidisciplinary tumor board discussion.

3. DISCUSSION

Desmoid-type fibromatosis is a rare, locally aggressive mesenchymal neoplasm of fibroblastic/myofibroblastic origin. It is classified by the World Health Organization (WHO) as an intermediate soft tissue tumor — neither benign nor overtly malignant — given its infiltrative growth and tendency for local recurrence without metastatic potential [8]. Molecular

studies have established that approximately 85% of sporadic cases harbor activating mutations in the CTNNB1 gene encoding β-catenin, a key mediator of the Wnt signaling pathway, explaining the characteristic nuclear β-catenin immunoreactivity used in pathological diagnosis [9].

Extra-abdominal desmoid tumors most commonly affect the shoulder, chest wall, and extremities. Foot and ankle involvement, as illustrated in our case, is rare. A systematic review by Sbaraglia *et al.,* identified only 78 cases of plantar or foot fibromatosis in the published literature, with most representing the superficial plantar variant (Ledderhose disease) rather than the deep desmoid type [10]. The deep desmoid variant involving the inter-metatarsal spaces, as seen in our case, is therefore particularly unusual and poses specific diagnostic challenges.

The MRI characteristics of desmoid fibromatosis are well described in the literature and reflect the histological composition of the tumor. The isointense T1 signal relative to muscle, which was observed in our case, is a consistent feature across series and reflects the predominant collagenous stroma [11]. T2 signal heterogeneity — also present in our case — is attributed to the variable ratio of collagen to cellularity within different areas of the tumor, with hypointense foci corresponding to densely fibrous regions and hyperintense areas reflecting more cellular or myxoid zones [12]. STIR hyperintensity, as observed here, reflects the high-water content of the more cellular and edematous tumor regions [13].

The differential diagnosis in our case included synovial sarcoma, given the biphasic MRI signal pattern, large size (~85 mm), and periarticular location. Synovial sarcoma typically affects young adults, shows a triple signal pattern on T2 (the 'bowl of grapes' appearance), and frequently displays calcifications or fluid-fluid levels, features that were absent in our case [14]. Morton's neuroma was also considered due to the inter-metatarsal extension of the lesion; however, Morton's neuroma is typically much smaller (<1 cm), fusiform, and located at the plantar level of the inter-metatarsal space, without the extensive aponeurotic infiltration seen here [15].

Histopathology with nuclear β-catenin immunostaining remains the diagnostic gold standard, as was confirmed in our case. Molecular testing for CTNNB1 mutations can be used as an adjunct diagnostic tool, particularly in cases where β-catenin staining is equivocal [9]. The R0 resection achieved in our patient represents the primary therapeutic goal; however, local recurrence rates following surgery range from 20% to 77% across published series, even after histologically confirmed negative margins [3,16]. This underscores the importance of long-term follow-up, as also observed in our patient who developed local recurrence at 12 months.

Contemporary management of DTF has shifted from a mandatory surgical approach to a more individualized, often non-operative strategy. The multicenter ALTITUDES trial and the phase III DeCision study have demonstrated the efficacy of sorafenib and nirogacestat (a γ-secretase inhibitor targeting the Notch pathway) in progressive or recurrent DTF, respectively [17,18]. For stable or minimally symptomatic recurrences such as that observed in our patient, an active surveillance (watch-and-wait) policy is now recommended by expert guidelines, including the European Reference Network for Rare Adult Solid Cancers (EURACAN) [19].

4. CONCLUSION

This case illustrates the diagnostic and therapeutic challenges posed by desmoid-type fibromatosis occurring in the atypical location of the foot. Its MRI features — isosignal T1, heterogeneous T2, STIR hyperintensity, and avid post-contrast enhancement — while suggestive, are non-specific and overlap with more aggressive entities, particularly synovial sarcoma. The insinuating inter-metatarsal growth pattern adds to the diagnostic complexity. Histopathological examination with β-catenin immunostaining is essential for definitive diagnosis. The local recurrence observed in our patient, despite R0 resection, is consistent with the known behavior of this entity. A multidisciplinary approach, integrating surgery, active surveillance, and systemic therapies in selected cases, is paramount. Clinicians should maintain a high index of suspicion for DTF in the differential diagnosis of soft tissue masses of the foot.

Patient Consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Conflicts of Interest

The authors declare no conflicts of interest related to this manuscript.

Funding

This research received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

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