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Desmoplastic Fibroblastoma (Collagenous Fibroma) of the Knee: A Case Report and Literature Review

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Abstract. *Background/Aim: Desmoplastic fibroblastoma, also known as collagenous fibroma, is a rare benign mesenchymal neoplasm that primarily arises in the subcutaneous tissue of upper extremities and limb girdles. The knee is an uncommon location for desmoplastic fibroblastoma. Recent studies have demonstrated the presence of immunoreactivity for FOS like 1 (FOSL1) and rearrangements of FOSL1. Case Report: A 70-year-old woman presented with a 1-year history of a palpable mass in the medial aspect of the right knee. Physical examination revealed a 4-cm, elastic hard, mobile, nontender mass. Magnetic resonance imaging (MRI) showed a well-defined mass with prominent low signal intensity on all pulse sequences. Contrast-enhanced MRI demonstrated mild internal enhancement with rim enhancement. After an open biopsy, the lesion was successfully treated by complete excision. Histologically, the tumor was composed of bland spindle or stellate-shaped cells embedded in an abundant collagenous stroma. Immunohistochemically, the tumor cells showed diffuse nuclear positivity for FOSL1. These findings were consistent with a diagnosis of desmoplastic fibroblastoma. The patient was asymptomatic and there was no evidence of local recurrence eight months after surgery. Conclusion: Desmoplastic fibroblastoma is a distinctive benign soft-tissue tumor with FOSL1 immunoreactivity and should be clearly distinguished from more biologically aggressive mesenchymal neoplasms.*

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Desmoplastic fibroblastoma, also known as collagenous fibroma, is a rare benign soft-tissue tumor first described in a seven-case series by Evans in 1995 (1). It belongs to the fibroblastic/myofibroblastic tumor group according to the latest World Health Organization classification of soft tissue and bone tumors (2). The etiology of desmoplastic fibroblastoma remains unknown. Desmoplastic fibroblastoma shows a wide anatomical distribution but most frequently occurs in the upper arm, shoulder and upper back (2). Complete excision is the treatment of choice and local recurrence has not been reported (2). Herein, we describe an unusual case of desmoplastic fibroblastoma of the knee in an early old woman. We also provide a literature review about the clinicopathological, imaging and molecular features of desmoplastic fibroblastoma. Written informed consent was obtained from the patient to publish this case report and accompanying images.

Case Report

A 70-year-old woman presented with a 1-year history of a palpable mass in the medial aspect of the right knee. She had an arthroscopic partial meniscectomy for degenerative meniscus tears in the right knee two years ago. Physical examination revealed a 4-cm, elastic hard, mobile, nontender mass. Range of motion of the affected knee was normal. Neurological and vascular examinations were unremarkable. Plain radiographs showed a subtle soft-tissue shadowing without calcification. Magnetic resonance imaging (MRI) demonstrated a well-defined mass with low signal intensity on both T1- and T2-weighted sequences (Figure 1A and B). Contrast-enhanced fat-suppressed T1-weighted sequences (Figure 1C) showed mild heterogeneous internal enhancement with rim enhancement. Based on the MRI findings, initial differential considerations included desmoplastic fibroblastoma, fibroma of tendon sheath, tenosynovial giant cell tumor, giant cell tumor of soft tissue and desmoid fibromatosis.

An open biopsy was carried out, and the pathological diagnosis was desmoplastic fibroblastoma. A marginal excision of the tumor was performed. Grossly, the excised mass was well-circumscribed and the cut surface was white to pearl-gray in color (Figure 2). Microscopically, the lesion was composed of bland spindle or stellate-shaped cells in an abundant

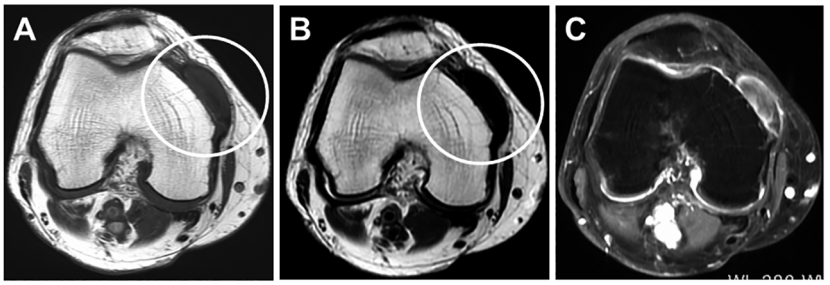

Figure 1. *Axial magnetic resonance images of desmoplastic fibroblastoma in the right knee. The mass (circle) reveals low signal intensity on both T1-weighted (A) and T2-weighted (B) sequences. Contrast-enhanced fat-suppressed T1-weighted sequence (C) demonstrates mild heterogeneous internal enhancement with rim enhancement.*

collagenous stroma (Figure 3A). Nuclear atypia and mitotic figures were absent. Immunohistochemically, the tumor cells were diffusely positive for FOS like 1 (FOSL1) (Figure 3B). Staining for CD34, smooth muscle actin, desmin, S-100 protein and beta-catenin was negative. These findings confirmed the diagnosis of desmoplastic fibroblastoma.

The postoperative course was uneventful. There was no clinical evidence of local recurrence at the 8-month follow-up.

Discussion

Desmoplastic fibroblastoma is an uncommon mesenchymal tumor and has a peak incidence in the fifth to sixth decades of life, with a strong male predominance (3). It typically presents as a firm, mobile, slowly growing, painless, subcutaneous mass in the upper extremities. Fascial and skeletal muscle involvement can be seen (3). The diameter ranges from 1 to 20 cm (median of 3 cm) (4). The clinical presentation of desmoplastic fibroblastoma depends on the location of the tumor. Desmoplastic fibroblastoma has a benign clinical course and marginal excision is adequate (4).

Desmoplastic fibroblastoma can show a radiological overlap with a variety of benign, intermediate and malignant mesenchymal tumors, including fibroma of tendon sheath, tenosynovial giant cell tumor, desmoid fibromatosis and low-grade fibromyxoid sarcoma (4-6). The most important differential diagnosis for the current case is desmoid fibromatosis, which is a locally aggressive but non-metastasizing mesenchymal neoplasm with infiltrative growth. In general, desmoplastic fibroblastoma exhibits prominent low signal intensity on all plus sequences (4). Contrast-enhanced MRI demonstrates mild internal enhancement with rim enhancement, as in our case. Yamamoto *et al*. suggested that the presence of rim enhancement might be a primary indication of desmoplastic fibroblastoma (7). On the other hand, desmoid fibromatosis exhibits decreased signal intensity on all plus sequences similar to desmoplastic fibroblastoma but demonstrates more moderate to marked enhancement. Unlike desmoplastic fibroblastoma, low signal-intensity bands can be

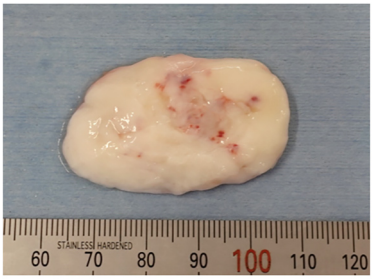

Figure 2. *Gross appearance displaying a well-circumscribed mass with a white to pearl-gray cut surface.*

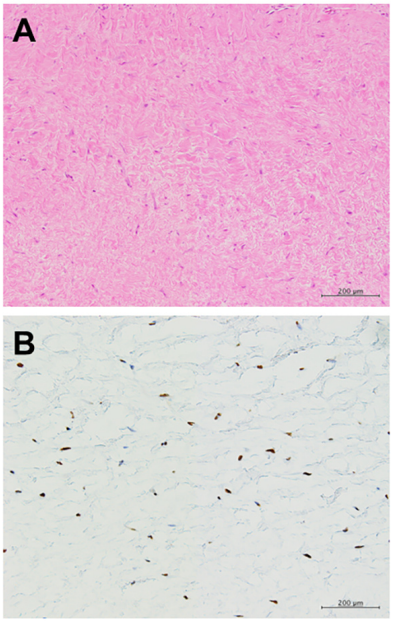

Figure 3. *Histological and immunohistochemical features of desmoplastic fibroblastoma. A: The tumor is hypocellular and consists of spindled and stellate-shaped cells in an abundant collagenous stroma (hematoxylin and eosin staining, original magnification ×100). B: The tumor cells show nuclear immunoreactivity for FOS like 1 (original magnification ×200).*

seen in desmoid fibromatosis (8). We previously suggested that the enhancement pattern and the presence of low signal-intensity bands would be helpful to distinguish desmoplastic fibroblastoma from desmoid fibromatosis (4).

The definitive diagnosis of desmoplastic fibroblastoma is made after excision and histopathological analysis. Histologically, desmoplastic fibroblastoma is hypocellular and consists of spindle to stellate-shaped cells in an abundant collagenous or myxocollagenous stroma with low vascularity, as in our case. Cytological atypia and nuclear hyperchromasia are absent (2). Mitotic figures are rare, and necrosis is absent. By immunohistochemistry, the tumor cells are diffusely positive for vimentin and focally positive for smooth muscle actin. Immunostains for desmin, CD34, S-100 protein, beta-catenin and epithelial membrane antigen are typically negative (3, 9). Notably, nuclear FOSL1 immunoreactivity, typically in a diffuse and strong manner, is highly sensitive and specific for desmoplastic fibroblastoma and is diagnostically helpful (10).

In current practice, cytogenetic and molecular genetic testing can serve as a useful diagnostic adjunct for soft-tissue neoplasms. Desmoplastic fibroblastoma is cytogenetically characterized by a t(2;11)(q31;q12) translocation (11, 12). Also, we previously reported the presence of a 2;11 translocation with slightly distal breakpoints (2q35 and 11q13) (13). In 2012, Macchia *et al*. reported that the functional outcome of 11q12 rearrangements was deregulated expression of *FOSL1* in desmoplastic fibroblastoma (14). Subsequently, *FOSL1* rearrangements were detected by whole-genome and targeted RNA sequencing in 10 (67%) of the 15 cases (15). The discovery of *FOSL1* rearrangements has recently led to more precise diagnosis of desmoplastic fibroblastoma.

Conclusion

Desmoplastic fibroblastoma is a distinctive benign soft-tissue tumor with FOSL1 immunoreactivity. Although rare, desmoplastic fibroblastoma should be considered in the differential diagnosis of a well-defined soft-tissue mass of the knee when prominent areas of low signal intensity are seen on all pulse sequences.

Conflicts of Interest

The Authors declare no conflicts of interest associated with this article.

Authors' Contributions

YS was a major contributor and collected the data. JN performed the operation and drafted the article. YC reviewed the article. All Authors read and approved the final article.

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