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Imaging Features of Desmoid-type Fibromatosis in the Teres Major Muscle

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Abstract. *Desmoid-type fibromatosis is a locally aggressive fibroblastic neoplasm with a tendency for local recurrence, despite adequate surgical resection. Its clinical presentation, biological behavior, and natural history can vary considerably. We present a unique case of desmoid-type fibromatosis arising in the left teres major muscle of a 62-year-old female. Physical examination showed a 7-cm, elastic-hard, immobile, tender mass. Magnetic resonance imaging (MRI) revealed a partially ill-defined mass, with intermediate signal intensity on T1-weighted sequences and heterogenous high signal intensity on T2-weighted sequences. Contrast-enhanced fat-suppressed T1-weighted sequences demonstrated intense and homogenous enhancement throughout the mass. Integrated positron-emission tomographic (PET)/computed tomographic (CT) images showed moderate focal ^{18}F-fluorodeoxyglucose uptake corresponding to the clinically palpable and MRI-described soft tissue mass, with a maximal standardized uptake value of 4.85. The possibility of a malignant lesion was raised. Following an open biopsy, wide resection of the tumor was performed. Histological examination confirmed the diagnosis of desmoid-type fibromatosis. Finally, we discuss the imaging features of this peculiar neoplasm on MRI and PET/CT.*

Desmoid-type fibromatosis is a locally aggressive soft tissue tumor, which does not metastasize. It belongs to the fibroblastic/myofibroblastic tumor group according to the 2013 World Health Organization Classification of Soft Tissue Tumors (1). Young adults are most commonly affected, with a peak prevalence between the ages of 25 and 35 years. Desmoid-type fibromatosis usually presents as a firm, poorly-circumscribed mass in the deep soft tissues of shoulder, chest wall, thigh, and neck. Local control is the main goal of treatment. The exact etiology of this tumor is unknown, but genetic, endocrine, and physical factors seem to play an important role in its development and growth (2). We herein describe the magnetic resonance imaging (MRI) and ^{18}F-fluorodeoxyglucose (FDG) positron-emission tomographic (PET)/computed tomographic (CT) appearances of a pathologically-proven desmoid-type fibromatosis. We also discuss the differential diagnosis of this rare condition.

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Case Report

A 62-year-old female was referred to Fukuoka University Hospital with a painless mass of unknown duration in the left axillary region. There was no history of antecedent trauma. Physical examination showed a 7-cm, elastic-hard, immobile, tender mass. Neurological and vascular examinations were unremarkable. The patient's medical history was non-contributory. MRI showed a partially ill-defined soft tissue mass in the left teres major muscle. The mass exhibited intermediate signal intensity on T1-weighted sequences (Figure 1A) and heterogenous high signal intensity on T2-weighted sequences (Figure 1B). Contrast-enhanced fat-suppressed T1-weighted sequences demonstrated intense and homogenous enhancement of the mass (Figure 1C). FDG PET images demonstrated moderate focal uptake in the left axillary region (Figure 2A). CT showed a homogeneous low-attenuating mass, with corresponding tracer uptake (Figure 2B). The maximal standardized uptake value (SUV_{max}) was 4.85 (Figure 2C). The differential diagnosis at this point included neurofibroma, desmoid-type fibromatosis, fibrosarcoma, and low-grade fibromyxoid sarcoma.

An open biopsy was performed to make an accurate diagnosis. Microscopically, the tumor consisted of elongated, slender, spindle-shaped cells forming a fascicular pattern in a collagenous and focally fibromyxomatous stroma (Figure 3A). Immunohistochemically, the tumor cells were positive for vimentin, β-catenin (Figure 3B), and smooth muscle actin (SMA) (Figure 3C), and focally positive for desmin (Figure 3D). Immunostaining for CD34 and S-100 protein was negative. The MIB-1 labeling index was approximately 8.1%. Based on these findings, the tumor was diagnosed as a

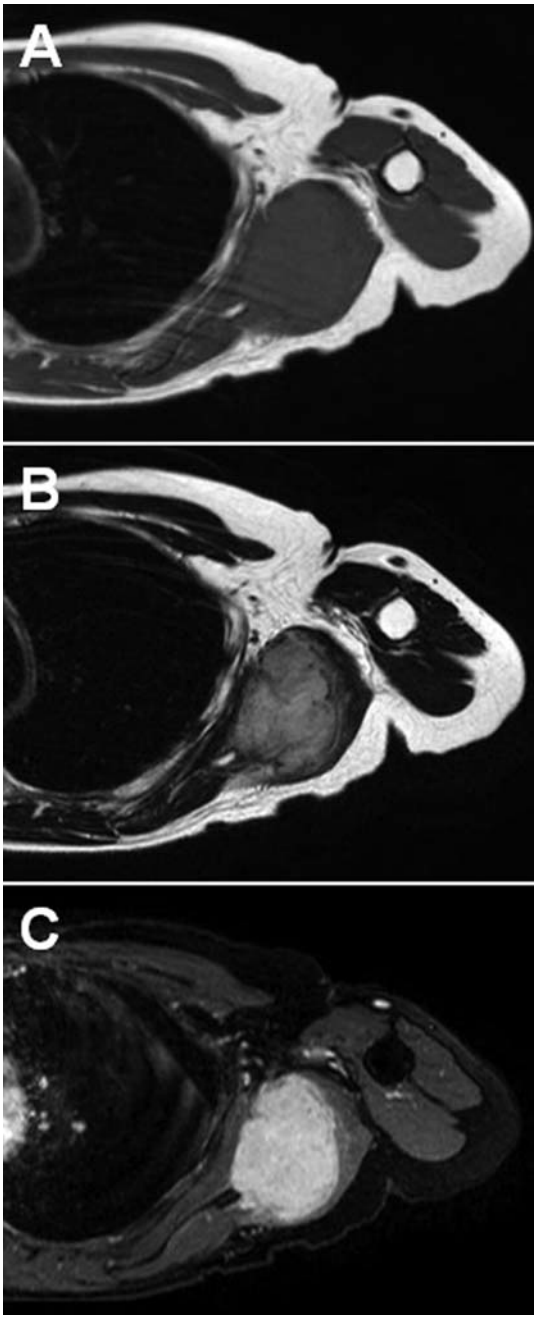

Figure 1. *Axial magnetic resonance images of desmoid-type fibromatosis in the left axillary region. A: T1-Weighted sequence shows that the mass has intermediate signal intensity. B: T2-Weighted sequence shows that the mass has heterogenous high signal intensity. C: Contrast-enhanced fat-suppressed T1-weighted sequence shows intense and homogenous enhancement of the mass.*

desmoid-type fibromatosis. We then performed a wide resection of the tumor. The postoperative course was uneventful. At the four-month follow-up, the patient was doing well without evidence of local recurrence.

Discussion

Wide surgical resection is the mainstay of treatment for desmoid-type fibromatosis when functionally and cosmetically acceptable. Radiation may be indicated after incomplete resection or if the tumor is unresectable with impeding functional problems (2). Systemic therapies can be considered in unresectable or recurrent disease, including hormone therapy, non-steroidal anti-inflammatory drugs, chemotherapy, and tyrosine kinase inhibitors (2-4). On the other hand, several recent studies have shown that a 'wait and watch' attitude is reasonable for asymptomatic or non-progressive desmoid-type fibromatoses (5, 6).

Histologically, the lesion is poorly-circumscribed and composed of elongated, slender, spindle-shaped cells in a prominent collagenous stroma. Microhemorrhages and focal aggregates of lymphocytes may be seen. The cells lack nuclear hyperchromasia or cytological atypia. Mitotic activity is typically low but can be higher in some cases. Immunohistochemically, the tumor cells are positive for vimentin, SMA, and muscle-specific actin, but typically negative for desmin, h-caldesmon, and S-100 protein. Approximately 70-75% of tumors exhibit nuclear positivity for β-catenin (1), as in our case.

The CT features of desmoid-type fibromatosis are usually non-specific. Lesions may be isodense or slightly hypodense relative to skeletal muscle (7, 8). On MRI, most tumors are either ovoid or infiltrative in shape (8, 9). Typically, desmoid-type fibromatoses exhibit intermediate signal intensity on T1-weighted sequences and heterogeneous high signal intensity on T2-weighted sequences (7-9). Bands of low signal intensity within the lesion on all pulse sequences may be seen (7). With administration of intravenous contrast material, moderate to marked enhancement is observed, particularly in less collagenized and more cellular regions. Although these features are characteristic of desmoid-type fibromatosis, it is occasionally difficult to distinguish this tumor from soft tissue sarcomas on MRI, as shown in our case.

FDG PET is being increasingly used in the detection and management of soft tissue tumors. In the current case, moderate FDG uptake was observed in the intramuscular mass, suspicious for malignancy. There have been several reports describing FDG PET/CT findings for desmoid-type fibromatosis (10-15). Our case and others demonstrate that desmoid-type fibromatoses have mild to moderate FDG uptake (SUV_{max}: 1.7-8.1). These findings suggest that FDG PET is an insufficient screening method for differential diagnosis between this tumor and soft tissue sarcomas. On the other hand, it has been shown that FDG PET is helpful for monitoring therapy with imatinib in patients with desmoid-type fibromatosis (13).

The differential diagnosis for desmoid-type fibromatosis includes a variety of benign and malignant fibrous soft tissue

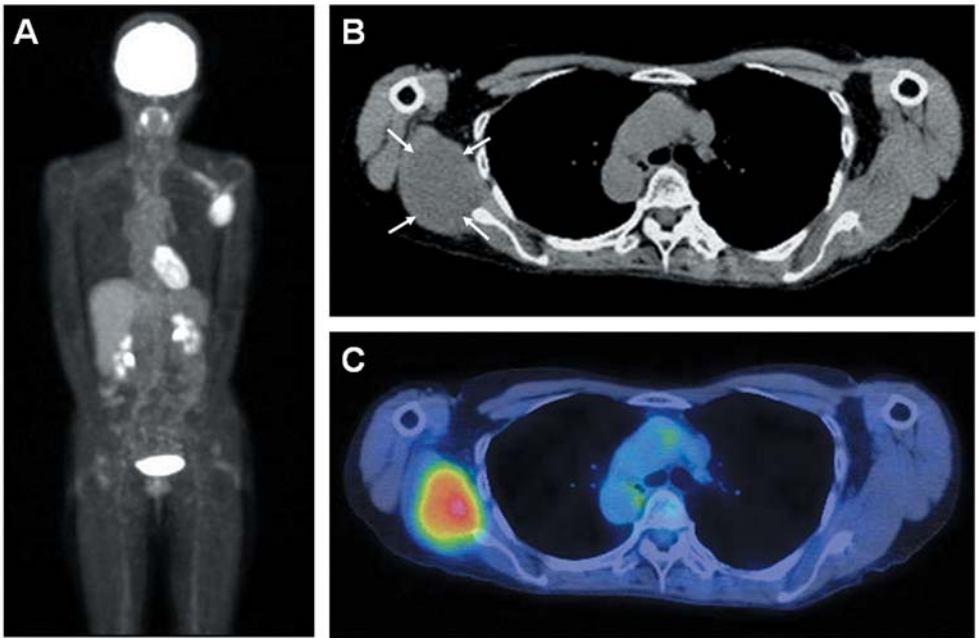

Figure 2. *A: Positron-emission tomography (PET) image demonstrates focal ^{18}F-fluorodeoxyglucose (FDG) uptake in the left axillary region. B: Axial computed tomography (CT) shows a homogenous low-attenuating mass (arrows). C: Integrated PET/CT image shows moderate focal FDG uptake in the deep soft tissue mass. The maximal standardized uptake value is 4.85.*

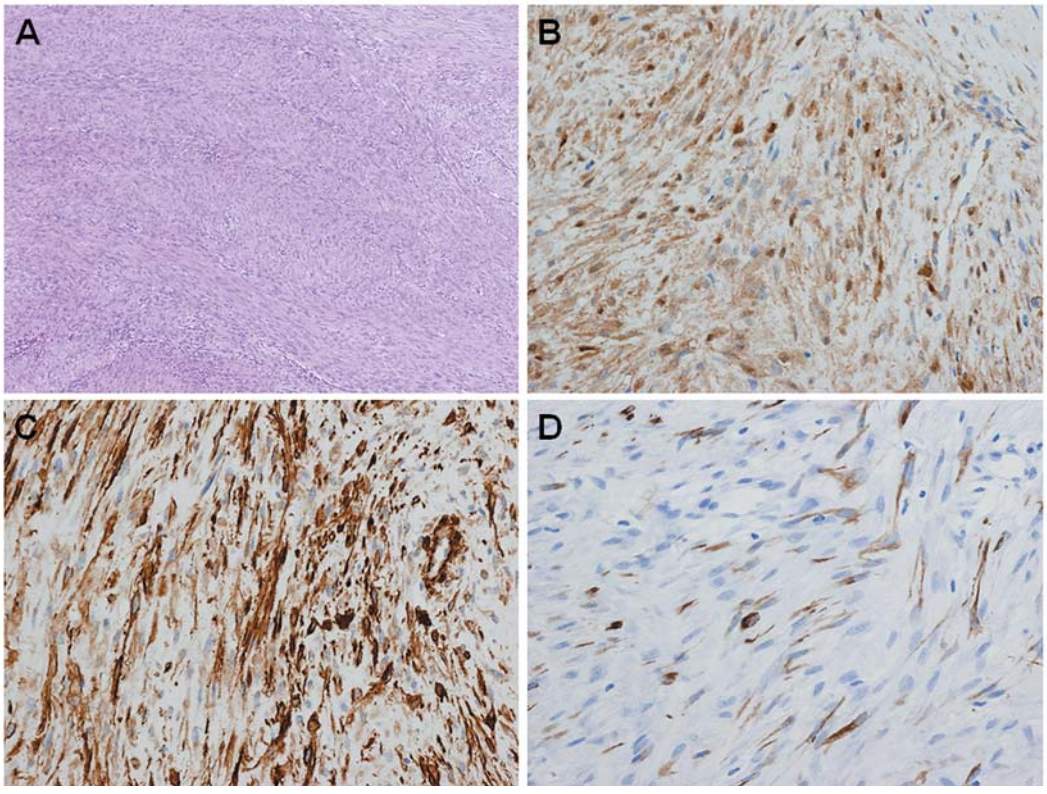

Figure 3. *Histological and immunohistochemical findings of desmoid-type fibromatosis. A: The tumor is composed of elongated, slender, spindle-shaped cells forming fascicular pattern in a collagenous stroma (hematoxylin and eosin staining, original magnification ×40). The tumor cells are positive for β-catenin (B) and smooth muscle actin (C), and focally positive for desmin (D) (B, C, and D, original magnification ×100).*

tumors, such as collagenous fibroma, fibrosarcoma, and low-grade fibromyxoid sarcoma.

Collagenous fibroma, also known as desmoplastic fibroblastoma, is a distinct benign fibroblastic/myofibroblastic tumor. It usually presents as a firm, slow-growing, painless mass in the subcutaneous tissues or skeletal muscle of the upper extremities (16). Collagenous fibroma and desmoid-type fibromatosis have similar signal intensity on T1- and T2-weighted sequences. However, the presence of rim enhancement on postcontrast T1-weighted sequences with fat suppression is strongly suggestive of collagenous fibroma (17). Histologically, collagenous fibroma consists of spindle- to stellate-shaped cells embedded in a collagenous or myxocollagenous stroma and is less cellular than desmoid-type fibromatosis. Re-arrangements of chromosomal band 11q12 are characteristic of collagenous fibroma, with the presence of an identical t(2;11)(q31;q12) translocation (16, 18).

Fibrosarcoma is a malignant tumor composed of fibroblasts with variable collagen production. It usually presents as a slow-growing, painless mass in the deep soft tissues of the lower extremities (19). Fibrosarcoma is difficult to diagnose clinically and has become, in large part, a diagnosis of exclusion (20). Histologically, fibrosarcoma consists of fusiform or spindle-shaped cells arranged in a herringbone growth pattern and is more uniformly cellular than desmoid-type fibromatosis. Unlike desmoid-type fibromatosis, the tumor cells are often overlapping and separated by less collagen. In our experience, however, it is often difficult to distinguish between these two tumor types, particularly when evaluating limited tissue samples.

Low-grade fibromyxoid sarcoma is a distinct malignant fibroblastic tumor with a bland histological appearance. It usually presents as a slow-growing, painless mass in the deep soft tissues of the lower extremities or trunk (21). Lesions tend to be hypodense relative to skeletal muscle with focal isodensity on CT. Low-grade fibromyxoid sarcomas typically have low to intermediate signal intensity on T1-weighted sequences and heterogeneous signal intensity on T2-weighted sequences. Moreover, a gyriform pattern may be seen on fluid-sensitive sequences (22). Histologically, low-grade fibromyxoid sarcoma consists of alternating fibrous and myxoid areas with bland spindle-shaped cells arranged in a whorled growth pattern. Compared with desmoid-type fibromatosis, low-grade fibromyxoid sarcoma is usually more cellular and less fascicular, and displays alternating fibrous and myxoid areas (23). Immunohistochemically, the tumor cells are positive for MUC4 (mucin 4) and focally positive for epithelial membrane antigen (24). Cytogenetically, low-grade fibromyxoid sarcoma is characterized by a balanced t(7;16)(q34;p11) translocation resulting in a *FUS* (fused in sarcoma)-*CREB3L2* (cAMP responsive element binding protein 3-like 2) fusion gene (21).

In summary, we report on the imaging features of an intramuscular desmoid-type fibromatosis with histological correlation. Our case indicates that a relatively high accumulation of FDG can be observed in desmoid-type fibromatosis.

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