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Case Report

A Rare Case of Desmoid Fibromatosis in a Pediatric Patient: Surgical Management and Outcomes ☆

Boon Tat Yeap, MD, MMed[a,b], *Ikhwan Nasir, MD, MMed*[a], *Thai Hau Koo, MD*[c], *Aaron Gerarde Paul, MD, MMed*[d], *Mohd Hazeman Zakaria, MD, MMed*[e,*]

[a] *Department of Anaesthesiology and Intensive Care, Hospital Universiti Malaysia Sabah, Kota Kinabalu, Sabah, Malaysia*
[b] *Faculty of Medicine and Health Sciences, Universiti Malaysia Sabah, Kota Kinabalu, Sabah, Malaysia*
[c] *Department of Internal Medicine, School of Medical Sciences, Hospital Universiti Sains Malaysia, Kubang Kerian, Kelantan, Malaysia*
[d] *Department of Orthopaedics, Hospital Queen Elizabeth, Kota Kinabalu, Sabah, Malaysia*
[e] *Department of Radiology, Faculty of Medicine and Health Sciences, Universiti Putra Malaysia, Selangor, Malaysia*

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ABSTRACT

Desmoid fibromatosis (DF) is a rare, locally aggressive tumor arising from the abdominal fascia or musculoaponeurosis, commonly affecting individuals between 15 and 60 years of age. We present a case of a 13-year-old boy with a 9-month history of progressive swelling in the left posterior thigh. Magnetic resonance imaging (MRI) revealed a soft tissue tumor encasing the sciatic nerve. A biopsy confirmed the diagnosis of DF, showing spindle cells arranged in fascicles. The patient underwent wide local excision of the tumor with preservation of the sciatic nerve. Postoperative recovery was smooth, and 1-year follow-up MRI showed no recurrence. Surgical excision remains the primary treatment, especially in symptomatic patients, although recurrence is common even with negative margins. This case underscores the importance of regular follow-up for DF and a multidisciplinary approach to optimize management and surveillance.

Introduction

Desmoid fibromatosis (DF) is a nonmetastasizing fibrous tumor that commonly originates in the abdominal fascia and musculoaponeurosis. The incidence of DF is reported as 2-4 cases per one million population per year, affecting individuals between the ages of 15 and 60 years [1]. The most common extra-abdominal locations are the shoulders, chest wall, back, thighs and neck. It is characterized by infiltrative invasion of soft tissues and has a high propensity for local recurrence after surgical excision [2]. Here, we present a rare case of DF in

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* Corresponding author.
E-mail address: hazeman@upm.edu.my (M.H. Zakaria).

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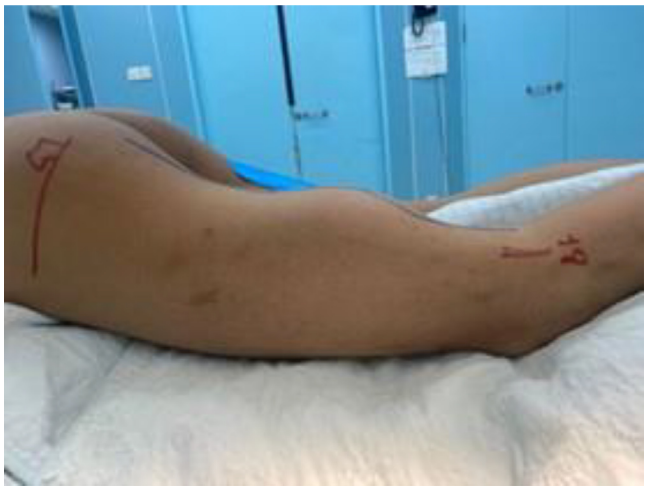

Fig. 1 – Clinical aspect of the tumor at the left thigh when the patient was proned.

a young teenage boy who presented with progressive swelling of the left posterior thigh. Herein, we discuss the epidemiology, management, and prognosis of patients with DF.

Case presentation

A 13-year-old boy presented to our center with a 9-months history of swelling of the posterior aspect of his left thigh. The swelling started with a ping-pong ball that gradually increased in size. He described the swelling as painless and immobile. The swelling did not disturb his activities of daily living such as walking or running. However, approximately a month prior to seeking treatment at our center, he complained of increasing pain and a limited range of movement of his left thigh. He denied having fever, weight loss, appetite, or any family history of malignancy. He had no history of trauma to the lower limbs.

Upon examination, the patient was pink, alert, and conscious. His vital signs were stable. A well-circumscribed mass, measuring 15 cm × 10 cm, was observed on the posterior aspect of the left thigh (Fig. 1). It was firm in consistency, did not adhere to the underlying muscles or skin, and was nontender to touch. No redness was observed on the skin swelling. The range of motion of both the lower limbs was normal. The results of other systemic examinations were unremarkable.

Our patient's full blood count (FBC) showed normal levels of hemoglobin, white cell count, and platelet count. No abnormalities were observed in liver or renal function tests. Chest radiography did not reveal any significant opacity. Magnetic resonance imaging (MRI) of the left lower limb showed an aggressive soft tissue tumor measuring 4.9 cm x 6.1 cm x 13.2 cm located deep in the posterior compartment of the mid-thigh, partially encasing the sciatic nerve (Fig. 2A–D). It was isointense to muscle on T1 weighted sequence and was heterogeneously hyperintense on T2 (Fig. 2A–D). Patchy areas of low signal intensities are seen suggesting fibrous components. These features were favorable for fibrous soft tissue sarcoma. Preoperative true-cut biopsy revealed a hypocellular fragment composed of spindle-shaped cells arranged in fascicles with intervening thin-walled blood vessels. No nuclear hyperchromasia or areas of tumor necrosis were observed. These features are highly suggestive of desmoid tumor.

Our provisional diagnosis was soft tissue sarcoma, as it presented as a slowly enlarging mass in the deep tissues of our young teenage patient. Differential diagnoses included benign intramuscular lipoma, lymphomas, and giant cell tumors. Lipomas are usually soft, mobile, and are less likely to cause significant discomfort. Lymphomas and giant cell tumors may not be possible, given the young age of our patient and the location of the mass in the posterior thigh.

Due to the huge size of the mass, which caused discomfort to our patient, he was counselled for wide local excision of the mass. The patient and his parents consented to this surgery, general anesthesia (GA), and for publication in this journal. Wide excision of the tumor was performed under GA, with the patient in the prone position. An incision was made along the axis of the tumor. The involved muscles, biceps femoris and semimembranosus, were excised with a grossly wide margin, preserving the entire sciatic nerve (Fig. 3). The mass measured 15 cm × 4 cm (Fig. 4) and was sent for histopathological examination (HPE).

The patient was discharged on the third postoperative day. He was examined at the outpatient orthopaedic clinic 2 weeks later. He was able to ambulate normally without any difficulties. The wound healed well without any infective changes. Postoperative HPE of the excised mass revealed spindle cells composed of fascicles of various lengths, with a uniform appearance. The neoplastic cells showed positive nuclear staining for beta-catenin but negative staining for Desmin and S100. The microscopic tumor margins were negative, and there was no evidence of malignant changes.

The patient underwent a 6 monthly review at our outpatient orthopaedic clinic. MRI of the left thigh, which was done 1 year later after surgery, did not show any evidence of tumor recurrence.

Discussion

Desmoid fibromatosis (DF), also known as aggressive fibromatosis or desmoid tumor, is a rare type of soft tissue tumor that arises from fibroblasts. They are slow-growing histologically benign tumors. However, they are locally aggressive and can infiltrate the surrounding tissues, including muscles, nerves, and organs. The most common extra-abdominal sites were the shoulders, chest wall, back, thigh, and neck. In the lower extremities, the quadriceps muscles in the anterior compartment and hamstring muscles in the posterior compartment are commonly affected [2,3]. The incidence of DF is reported as 2-4 cases per one million population per year, and it affects individuals between the ages of 15 and 60 years [1].

Risk factors for the development of DF include female sex, previous surgical history or trauma [4]. In addition, it is common in patients with familial adenomatous polyposis

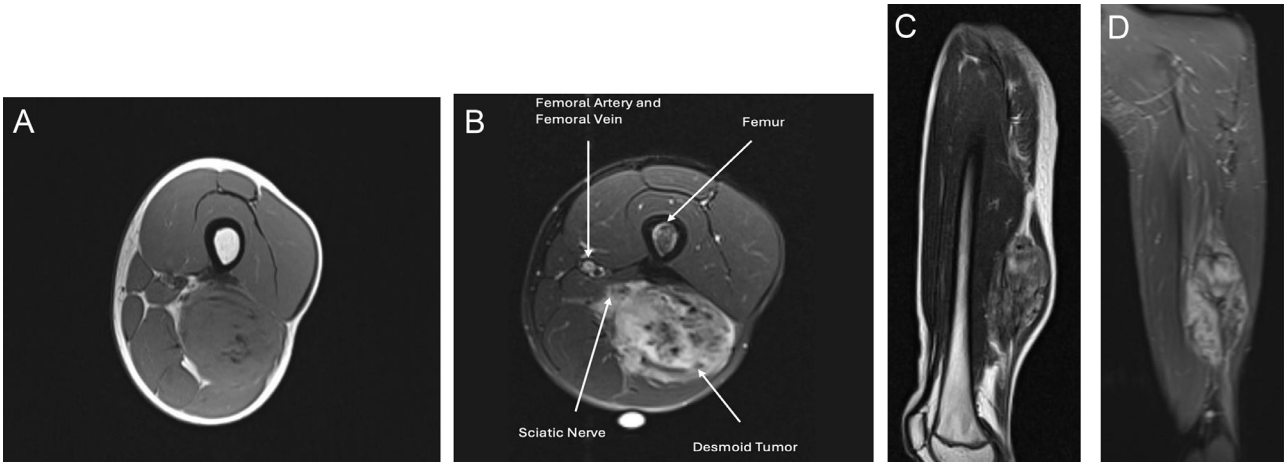

Fig. 2 – Magnetic Resonance Imaging (MRI) of Left Thigh at mid-thigh level. A: MRI-T1WI Axial; B: MRI-T1WI Axial fat saturation sequence; C: MRI-T1WI sagittal; D: MRI-T1WI sagittal fat saturation sequence. There is a large solitary mass arising from the posterior compartment of the mid-thigh. This mass is isointense to muscle in T1WI, heterogeneously hyperintense in T2WI, and shows no suppression in fat saturation sequence. The mass is partially encasing the sciatic nerve anteriorly. Contrast is not given in this case as the patient has contraindications for contrast agents.

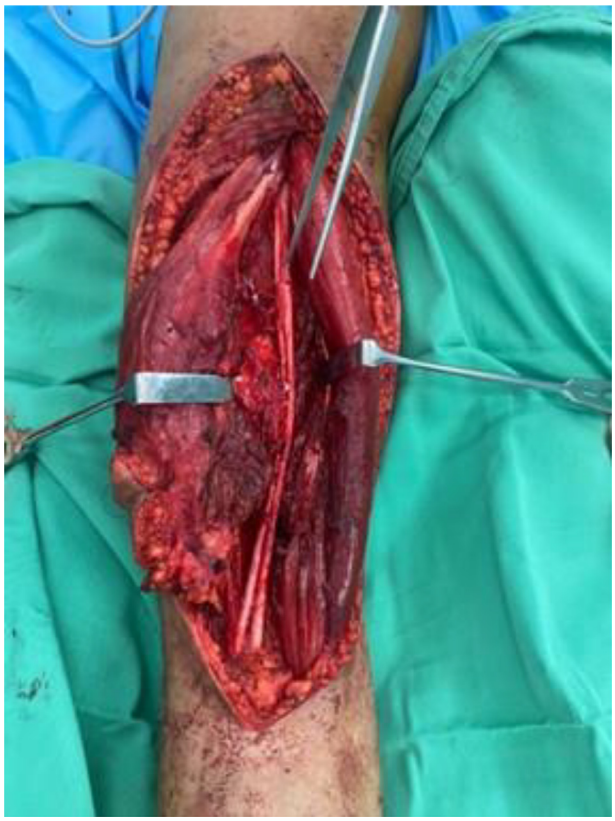

Fig. 3 – The sciatic nerve was preserved after being separated from the tumor.

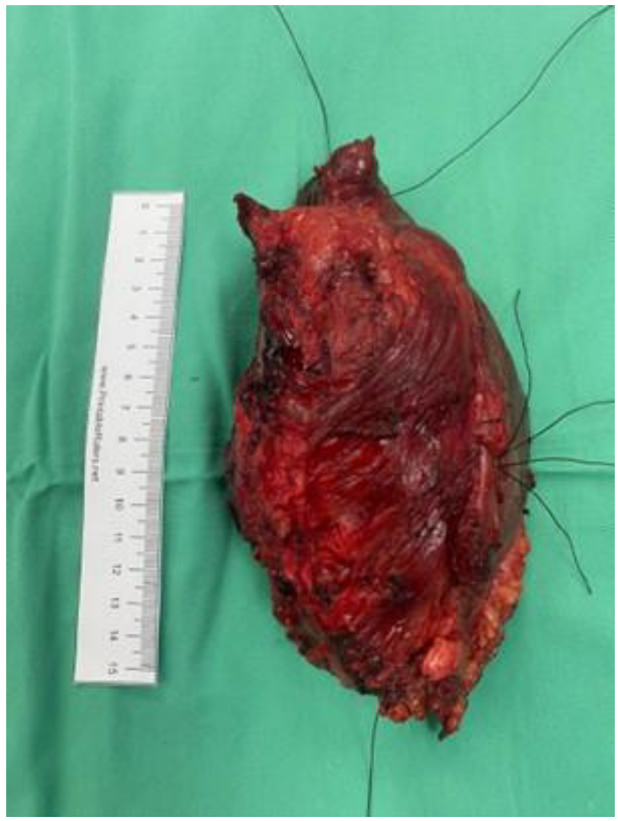

Fig. 4 – The appearance of the tumor measuring 15 cm × 4 cm after resection.

(FAP), with a prevalence of as high as 10% [5]. Our patient did not have any identifiable risk factors for the development of DF.

Considering its extreme rarity, DF is often misdiagnosed as a soft tissue and vascular tumor, such as fibrosarcoma or hemangioma. MRI is the gold standard for radiological investigation, as it allows detailed assessment of the extent of the

tumor, involvement of nearby structures, and planning for surgical interventions. The histopathological features of DF are characterized by proliferation of uniform spindle cells with an abundance of collagen. However, necrosis and pleomorphism were absent [6,7]. Based on our extensive literature review, B-catenin (CTNNB1) was detected in 80% of sporadic DF cases and 67% of tumors in patients with FAP. Thus, nuclear staining for B-catenin supports the diagnosis of DF [8].

The treatment options for patients with DF vary depending on the patient's symptoms, tumor location, size, and growth behavior. Management strategies are often individualized, ranging from active surveillance to aggressive multimodal therapy. Active surveillance is advocated for asymptomatic patients with slow-growing tumors [9]. This approach involves regular clinical monitoring using MRI.

Surgical excision is considered for tumors that progress rapidly and cause significant symptoms and functional impairment, as in the case of our patient. Complete surgical removal with clear margins is challenging because of the infiltrative nature of DF. Even with complete resection, there is a high risk of local recurrence of approximately 20-30% even when negative margins are achieved [10,11].

Radiation may be employed as an adjuvant therapy, particularly in cases where negative margins are not achieved or if the margins are contaminated [5]. Another treatment approach is targeted therapy using tyrosine kinase inhibitors, such as pazopanib, imatinib, or sorafenib, which have shown efficacy in managing DF. This is often used when the tumor is refractory to other treatments or when it shrinks prior to surgery [11]. However, this therapy was not used in our patient owing to its high cost.

Conclusion

Desmoid fibromatosis (DF) is a challenging clinical condition characterized by locally aggressive behavior and strong tendency for recurrence. Although imaging findings may be suggestive, histopathological confirmation is necessary before treatment. Management options include observation, surgical resection, radiotherapy, and the use of newer molecular-targeted agents. A multidisciplinary approach tailored to individual patients is usually needed depending on the location and local effects.

Data availability

Data not available / The data that has been used is confidential.

Patient consent

Written informed consent was obtained from the legal authorized representative of the patient for the publication of this case report.

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