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GIANT RECURRENT DESMOID FIBROMATOSIS INVOLVING A BELOW-KNEE AMPUTATION STUMP WITH EXTENSION TO THE THIGH AND GLUTEAL REGION: A CASE REPORT

KEY WORDS: Desmoid fibromatosis; aggressive fibromatosis; amputation stump; recurrent soft tissue tumor; radiotherapy

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ABSTRACT

Introduction: Desmoid fibromatosis is a rare, locally aggressive fibroblastic tumor with no metastatic potential but a significant tendency for local recurrence. We report an unusual case of giant recurrent desmoid fibromatosis involving a right below-knee amputation stump with proximal extension to the thigh and gluteal region in a 35-year-old man. **Case Presentation:** The patient presented with pain, foul-smelling discharge, ulceration, severe anemia, nutritional compromise, and wheelchair dependence. Contrast-enhanced computed tomography angiography demonstrated a large infiltrative soft tissue mass measuring approximately 54 × 40 × 19.4 cm with prominent vascularity. Complete wide excision was not feasible because of the size, infiltrative nature, and vascularity of the lesion. Surgical debulking was performed to reduce tumor burden and improve function, followed by adjuvant radiotherapy. Histopathology confirmed desmoid fibromatosis with nuclear β-catenin positivity. At one-year follow-up, the patient remained free of clinically evident recurrence and had improved from wheelchair dependence to crutch-assisted ambulation. **Conclusion:** This case highlights the role of individualized multimodal management in advanced recurrent desmoid fibromatosis with severe functional morbidity.

Case Report

A 35-year-old man presented with a progressively enlarging mass over the right lower limb stump and proximal thigh for one year. He had undergone right below-knee amputation at the age of 14 years for a clinically similar lesion involving the right foot and leg; however, previous histopathology records were not available. The present swelling had gradually increased in size and was associated with pain, foul-smelling discharge, and severe restriction of mobility. At presentation, the patient was wheelchair-bound and appeared severely anemic and nutritionally depleted.

On clinical examination, there was a giant, circumferential, irregular, lobulated soft tissue mass involving the right gluteal region, entire thigh, and below-knee amputation stump. The overlying skin was thickened and showed areas of ulceration with discharging sinuses, consistent with chronic pressure and secondary infection-related changes. (Fig. 1)

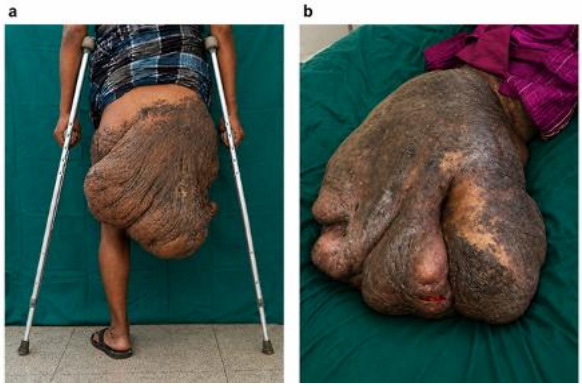

Fig. 1: Preoperative clinical photographs showing a giant lobulated soft tissue mass involving the right gluteal region, thigh, and below-knee amputation stump, with thickened skin, ulceration, and discharging sinuses.

Laboratory evaluation revealed severe anemia with a hemoglobin level of 6.5 g/dL. The patient was optimized preoperatively with transfusion of three units of packed red blood cells. Contrast-enhanced computed tomography angiography (CTA) demonstrated a large infiltrative soft tissue mass involving the skin, subcutaneous tissue, and right gluteus maximus muscle, measuring approximately 54 × 40 × 19.4 cm, with no evidence of intra-abdominal, pelvic, or osseous extension. Cross-sectional imaging and angiographic assessment further demonstrated extensive soft tissue involvement with prominent internal vascularity. The lesion showed vascular supply from hypertrophied branches of the superior and inferior gluteal arteries, with venous drainage into tributaries of the internal iliac venous system, explaining the anticipated risk of intraoperative bleeding. (Fig. 2)

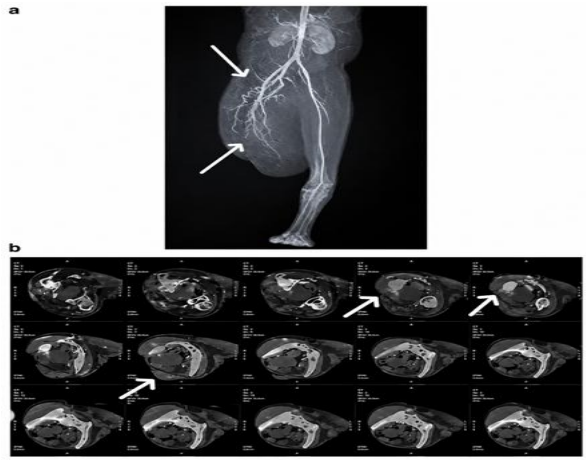

Fig. 2: Contrast-enhanced computed tomography angiography images showing a large infiltrative soft tissue lesion involving the right gluteal region and proximal thigh,

with extensive subcutaneous and deep soft tissue involvement. Prominent vascular channels and arterial supply from hypertrophied gluteal artery branches are marked with arrows.

After preoperative optimization, the patient underwent surgical debulking under general anesthesia. Complete wide excision was not feasible because of the massive size, infiltrative nature, and vascularity of the lesion; therefore, debulking was performed with the aim of reducing tumor burden, controlling local symptoms, and improving functional status. The tumor was excised in stages, with meticulous dissection to minimize blood loss. As the procedure was intended as debulking rather than wide oncological excision, margin-negative resection was not attempted. Multiple large tumor fragments were removed. The excised fragments weighed approximately 22 kg in total, with the largest fragment weighing 14 kg. The wound was closed in layers over a drain with pressure dressing. (Fig. 3)

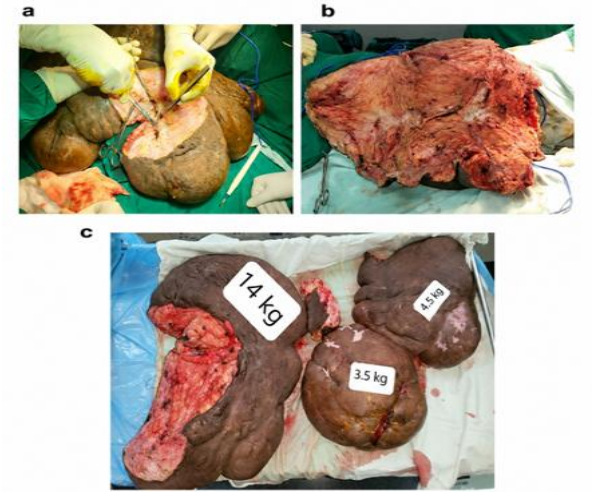

Fig. 3: Intraoperative photographs showing staged surgical debulking and the excised tumor specimen.

Histopathological examination showed a spindle cell lesion arranged in fascicles within collagenous stroma, consistent with desmoid fibromatosis. Immunohistochemistry demonstrated strong nuclear β-catenin positivity, confirming the diagnosis and distinguishing it from other spindle cell and neurofibromatous lesions.

The postoperative period was uneventful. In view of the recurrent nature, massive tumor burden, and high risk of local recurrence, the patient subsequently received adjuvant radiotherapy. At one-year follow-up, he remained free of clinically evident recurrence and demonstrated marked functional improvement, transitioning from wheelchair dependence to ambulation with crutch support. (Fig. 4)

Fig. 4: Follow-up clinical photograph showing improved functional status with crutch-assisted ambulation after surgical debulking and adjuvant radiotherapy.

DISCUSSION

Desmoid fibromatosis, also known as aggressive fibromatosis, is a rare intermediate-grade soft tissue tumor arising from fibroblastic or myofibroblastic proliferation. Although it does not metastasize, it is characterized by infiltrative local growth and a significant tendency for recurrence [1,2]. It accounts for approximately 0.03% of all tumors, with an estimated annual incidence of 2–4 cases per million population [1].

Desmoid fibromatosis can occur at almost any anatomical site, but it is most commonly reported in the abdominal wall, intra-abdominal region, and extremities [3,4]. Clinical presentation depends on the size and location of the lesion.

Management has evolved from routine surgical excision toward a more individualized approach. Active surveillance is now preferred for selected asymptomatic or stable lesions, whereas surgery, radiotherapy, and systemic therapy are reserved for progressive, symptomatic, recurrent, or function-limiting disease [5,6].

The pathogenesis of desmoid fibromatosis is multifactorial. Sporadic tumors are commonly associated with dysregulation of the Wnt/β-catenin pathway, while familial cases may be related to familial adenomatous polyposis [6,7]. Other factors such as previous trauma, surgery, hormonal influences, and genetic susceptibility have also been implicated [6,8]. In the present case, recurrence at the site of a previous below-knee amputation suggests a long-standing locally aggressive disease process..

The present case is unusual because of the involvement of an amputation stump with massive proximal extension to the thigh and gluteal region. Involvement of an amputation stump with massive proximal extension is an unusual presentation and appears to be rarely described in the available literature. The patient presented with a large dependent mass, ulceration, foul-smelling discharge, severe anemia, nutritional compromise, and wheelchair-bound status. These features made the case clinically challenging, not only because of tumor size, but also because of the need for preoperative optimization, vascular assessment, blood loss control, wound closure, and postoperative rehabilitation.

The management of desmoid fibromatosis has changed over time. Active surveillance is now recommended for selected asymptomatic or stable tumors because some lesions may remain indolent or regress spontaneously [5]. However, observation alone is not appropriate in patients with progressive, painful, ulcerated, infected, or functionally disabling disease. In this patient, the massive size of the tumor, recurrent nature, ulceration, discharge, anemia, and severe mobility limitation justified active intervention.

Surgery remains an important treatment option in carefully selected patients. However, complete resection with negative margins may be difficult because of the infiltrative nature of the tumor and its proximity to vital neurovascular and muscular structures. In the present case, radical excision was not feasible without significant morbidity. Surgical debulking was therefore performed with the practical aim of reducing tumor burden, controlling local symptoms, minimizing complications, and improving functional status.

Adjuvant radiotherapy has been reported to improve local control in selected cases, particularly in recurrent disease, residual disease, or tumors with a high risk of recurrence [8]. In this patient, postoperative radiotherapy was appropriate because of the recurrent presentation, massive tumor burden, and inability to achieve wide oncological clearance. Systemic therapies, including nonsteroidal anti-inflammatory drugs, hormonal therapy, chemotherapy, and tyrosine kinase inhibitors, have also been used in unresectable or

progressive disease [6]. However, in this case, the immediate priority was surgical reduction of a large symptomatic mass causing major functional disability.

This case demonstrates that desmoid fibromatosis, despite lacking metastatic potential, can cause major local morbidity when recurrent and advanced. In selected symptomatic patients with ulceration, infection, anemia, and functional disability, surgical debulking followed by adjuvant radiotherapy may provide meaningful local control and functional improvement. Long-term follow-up remains essential because of the risk of recurrence.

Statements and Declarations

Funding: No funding was received for this study.

Competing interests: The authors declare no competing interests.

Ethics approval: Institutional ethics approval was not required for this case report as per local policy.

Consent to publish: Written informed consent was obtained from the patient for publication of clinical details and accompanying images.

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