Case Report

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Lower li1mb mesenchymal hamartoma masquerading as hemangioma: a case report of AA2 atypical presentation and diagnostic challenges

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Introduction and importance: Hamartomas are abnormal growths that consist of various types of mesenchymal tissues, including cartilage, fat, connective tissue, and smooth muscle. These tumors can occur in almost any organ system within the body. While head and neck hamartomas have been frequently documented, cases involving the lower limbs are relatively rare. However, a unique instance of a mesenchymal hamartoma located in the leg has recently emerged.

Case presentation: A 30-year-old woman presented with a painless, gradually growing swelling on her left leg, diagnosed as a subcutaneous hemangioma preoperatively. Despite unsuccessful propranolol treatment, surgical excision revealed a mesenchymal hamartoma. Postoperative examination confirmed the diagnosis, and the patient recovered smoothly without recurrence after a week of hospitalization.

Clinical discussion: Cutaneous mesenchymal hamartomas are benign tissue growths with unknown etiology, associated with syndromes like Cowden and Peutz-Jeghers. Smooth muscle hamartoma can be acquired after skin trauma. Hemartoma can be misdiagnosed as hemangiomas. Cutaneous mesenchymal hamartomas presents as painless swellings in various locations, occasionally in adulthood. Differential diagnoses include lipomas and fibromas. Surgical excision is recommended for symptomatic cases to prevent recurrence.

Conclusion: This case highlights the atypical presentation of the hamartoma and emphasizes the importance of accurate diagnosis and management.

Keywords: case reports, hemangioma, mesenchymal hamartoma, soft tissue mass

Introduction

Hamartomas are benign masses of disordered tissue. They are exclusive to the site from which they develop and grow at the same pace as the original tissue, as opposed to a cancerous tumor. Hamartomas rarely compress or invade surrounding structures. Hamartomas can appear sporadically in the body or as a part of a syndrome. They can occur in the lungs, colon, breast, and hypothalamus[1]. Hamartomas can develop in the skin from various cell lineages. Cutaneous hamartomas may look similar to masses and take on different shapes depending on the type of cell that predominates. Although most cases are noninvasive, hamartomas can cause complications such as infections, infarcts, pressure/obstructions, hemorrhage/anemia, and sometimes neoplastic transformation[2]. Therefore, it is important to diagnose and treat hamartomas so that quality of life can be improved and complications can be avoided. In this case, we present a patient with a mesenchymal hamartoma (hemangioma) over the middle third of the left leg. This case has been diagnosed in line with SCARE (Surgical Case Report) Criteria[3].

HIGHLIGHTS

- Hemartoma are rare tumors where the patient may have the only symptoms of swellings.
- Diagnosis of hamartoma in low-resource setting countries can be challenging and can be misidentified as hemangioma.
- Surgical biopsy and histological examination can lead to proper diagnosis.

Case presentation

A 30-year-old woman arrived at the surgical outpatient department complaining of a painless swelling on her left leg's anterior

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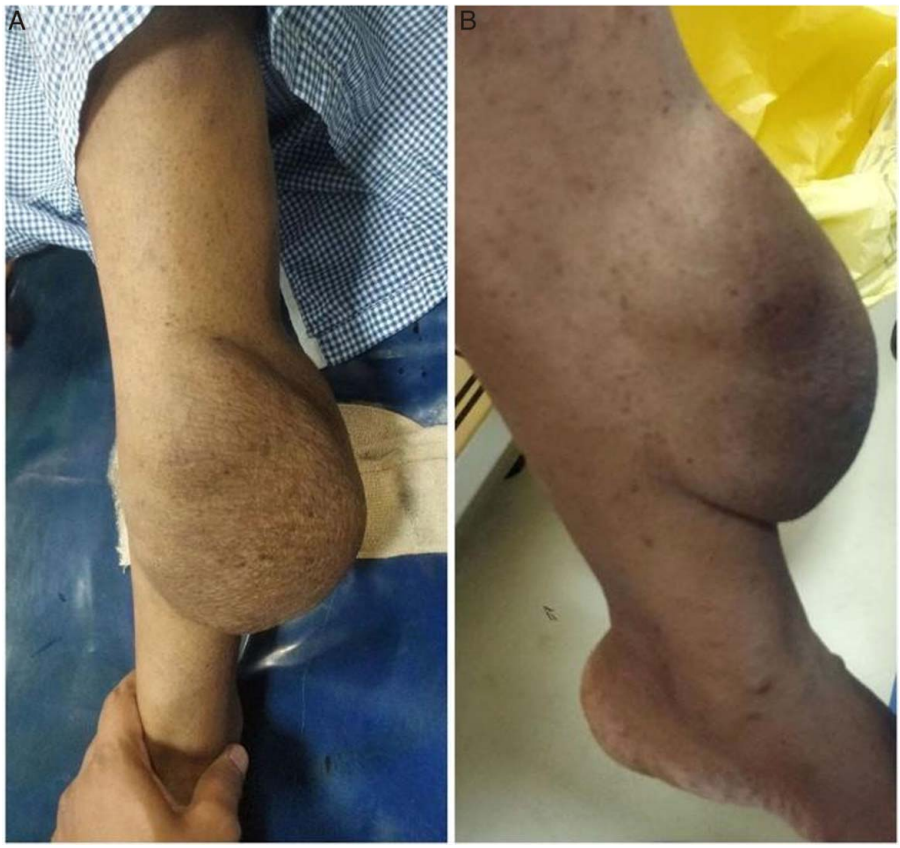

Figure 1. Image of swelling on the anterior aspect of left leg below the knee A, B, C showing Intraoperative images – images of excision of the leg mass from subcutaneous planes.

aspect. Since birth, there has been a small oval-shaped bulge. It grew in size gradually and is currently around 10 × 6 cm in size (Fig. 1). No concomitant symptoms exist, such as fever, warmth locally, or any other obvious secondary changes in the area. There is no relevant family history, or drug and allergy history related to this illness.

On examination, the swelling is smooth and soft in consistency, with mobility in all directions. There was no fluctuation, translucency, reducibility, compressibility, or pulsatility. A left leg lateral view below the knee radiograph was taken (Fig. 2), followed by which MRI (Fig. 3) was also performed on the left leg.

The review of diagnostic reports showed a large solitary, relatively well-defined lesion noted on the anterior aspect of the diaphysis of the tibia, which was heterogeneously isointense. The measurement of 13 × 9 × 6 cm was observed in the subcutaneous planes, and a feeding vessel provided a suggestive diagnosis of hemangioma. Hemangioma in the subcutaneous plane was diagnosed preoperatively. Although propranolol was administered as part of the medical management, no improvement was observed. Thus, surgical management of excision and biopsy under general anesthesia was planned (Fig. 4).

The biopsy report of the specimen (Fig. 5A and B) revealed a lesion with vascular channels of varying caliber admixed with mature adipose tissue, neural elements in the form of bundles, and spindle cell loose stroma suggestive of mesenchymal hamartoma (Fig. 5C and D).

A final pathological postoperative examination confirmed that the lesion was a mesenchymal hamartoma of the left leg. The patient had a smooth postoperative recovery and was discharged in a week with no recurrence on follow-up for a month.

Discussion

Hamartomas are fundamentally normal tissue replicating in a disordered manner. They are usually benign and grow at the same rate as the original tissue but can sometimes turn malignant. The etiology of hamartomas is unknown but is most likely due to a developmental error[1]. Hamartomas can also be part of syndromes, such as Cowden syndrome and Peutz-Jeghers syndrome[1]. Cutaneous mesenchymal hamartomas (CMH) are usually congenital without a known cause. However, one type of CMH called smooth muscle hamartoma can be acquired, especially after a skin trauma[2].

CMH typically presents as a painless swelling and can emerge from any site, including the thigh, leg, foot, palm, and perianal area[4,5]. The prevalence of cutaneous hamartomas is unknown because of their rarity and has a potential for misinterpretation as other swellings. While cutaneous hamartomas usually present during infancy, our case came presenting with a CMH in her left leg at 30 years. This may be due to the innocuous nature of the swelling during that period, which did not prompt the patient to seek medical care until the hamartoma grew to a significant size and she was cosmetically unsatisfied. Jang and colleagues reported a similar case who

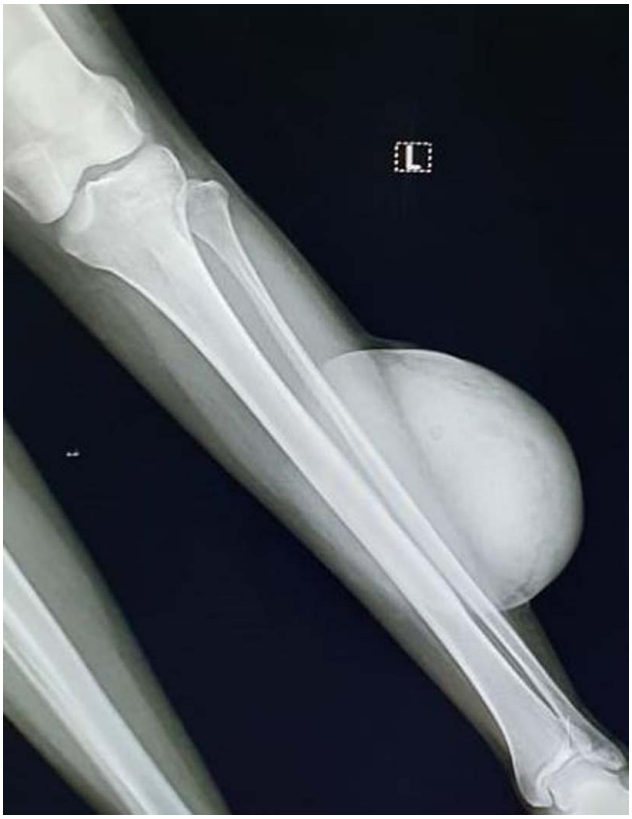

Figure 2. X-ray of left leg lateral view below the knee-revealing localized swelling.

came presenting with a CMH in the left foot at 16 years. In their presented case, the patient only had a few slightly elevated papules in his left foot during infancy. With age, the lesion grew until it started causing pain on walking[4] however, in our case, the patient had no complaints besides the swelling. Another case reported by Rosenberg and colleagues was a 71-year-old man with a rhabdomyomatous mesenchymal hamartoma (RMH) on his right temple. The patient sought treatment when he injured the swelling while cutting his hair[5].

Hamartomas can resemble numerous other swellings due to the unpredictability and variety of cell types seen and thus have a wide range of differential diagnoses. Lipomas, fibromas, hemangiomas, and rhabdomyoma are some of the swellings that cutaneous hamartomas resemble[2]. Some types of cutaneous hamartomas may have other swellings added to their list of differential diagnoses according to their appearance and site, such as RMH and fibro-lipomatous hamartomas[4]. Several authors in their studies have proposed oral propranolol as the treatment modality of choice in the case of capillary hemangiomas. A complete regression of facial infantile hemangioma in a child was reported, who was treated with propranolol[6].

Cutaneous hamartomas are divided into several types according to their appearance under a microscope and immunohistochemistry. Examples include fibrous hamartoma of infancy, fibro-lipomatous hamartoma, RMH, smooth muscle hamartoma, and vascular malformations[7]. Fibrous hamartoma of infancy shows mature fat cells, myofibroblasts haphazardly arranged in fascicles, and mesenchymal areas containing numerous vascular beds. In immunohistochemistry, the myofibroblasts express smooth muscle actin, and the mesenchymal areas express CD34[7]. Fibro-lipomatous hamartoma shows benign perineural fibroadipose proliferation and infiltration of the epineurium and perineurium with fat and fibrocartilaginous tissue that spreads the nerve bundles apart and thickens them[7]. RMH exhibits a diverse collection of disorganized proliferating cells, including mature fat

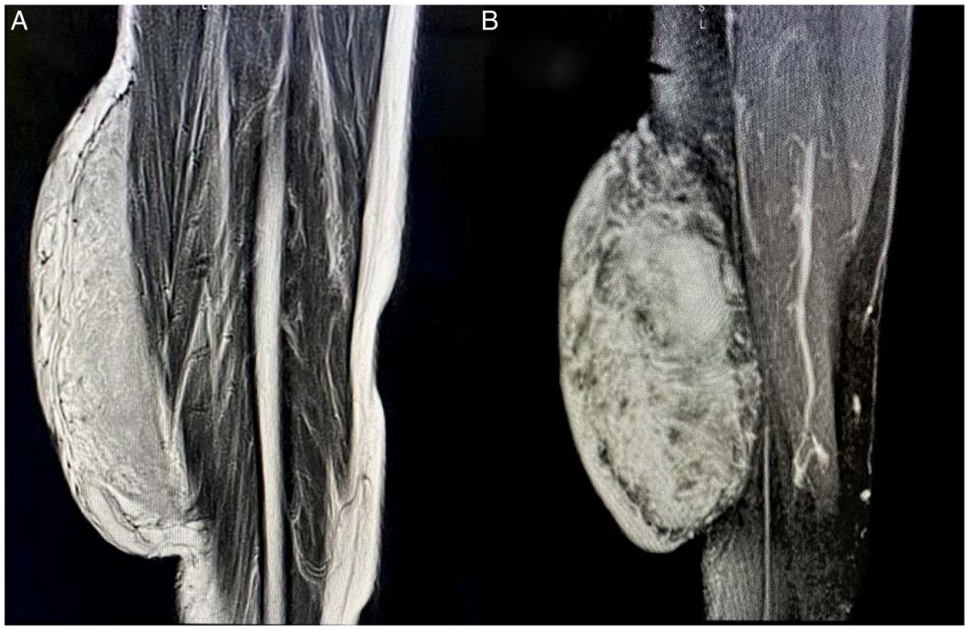

Figure 3. MRI left leg below knee sagittal view showcasing large solitary, well-defined, heterogeneously isointense lesion in the subcutaneous planes on the anterior aspect of the diaphysis of the tibia, measuring 13 × 9 × 6 cm with a feeding vessel A, B, C showing Intraoperative images – images of excision of the leg mass from subcutaneous planes.

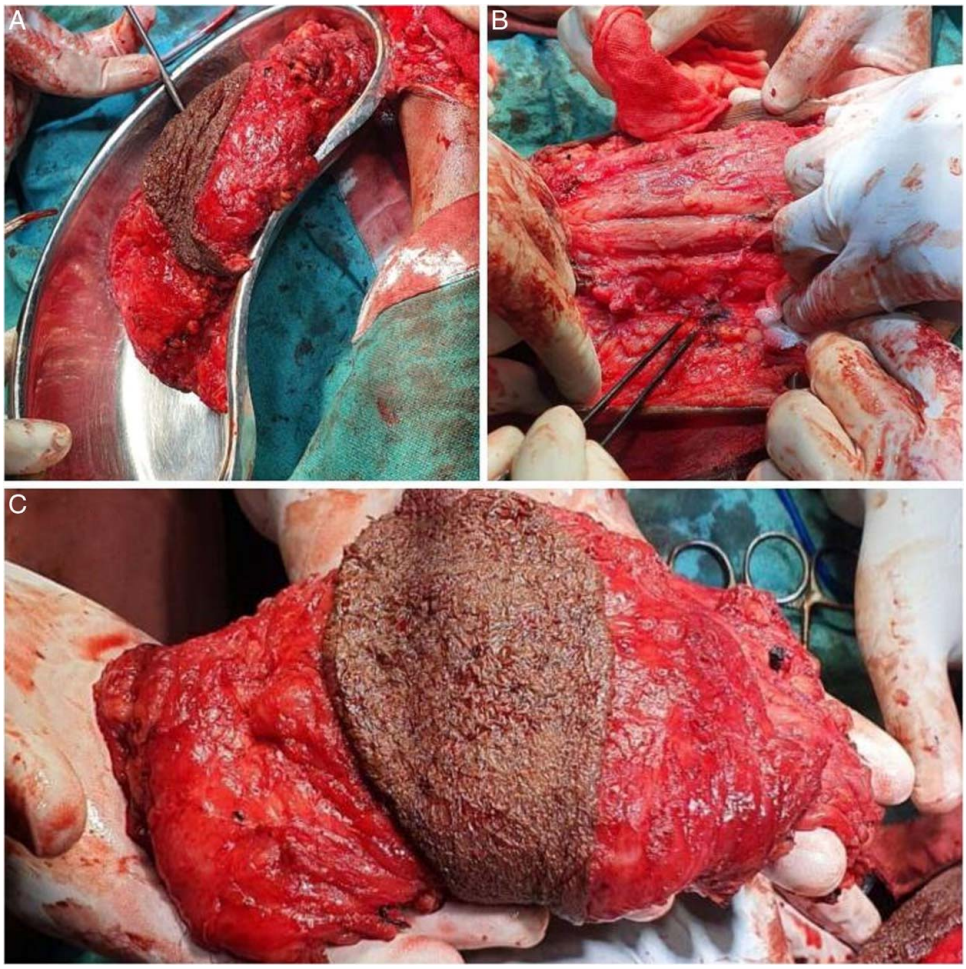

Figure 4. Intraoperative images – images of excision of the leg mass from subcutaneous planes A, B, C showing Intraoperative images – images of excision of the leg mass from subcutaneous planes.

cells, skeletal muscle, nerve fibers, fibrocartilaginous cells, and adnexal structures. One characteristic finding under the microscope in RMH is the presence of pilosebaceous units 'enveloped' by benign, haphazardly arranged skeletal muscle fibers[7].

Since they are benign and innocuous lesions, CMH can be left untreated. However, if the swelling causes any morbidity, disability, or cosmetic dissatisfaction, the hamartoma is surgically excised. Incomplete resection can lead to recurrence of hamartomas[8].

Conclusion

Hemangioma should be ruled out before arriving at the rare diagnosis of hamartoma. Failure to respond to oral propranolol can be a clue. Complete surgical excision is the only definite treatment available for hamartoma. There is very little literature available in line with CMH, which could be due to misdiagnosis, especially in low-resource setting countries, which also limited our case report with further discussion due to the lack of availability of reporting of such cases.

Ethical approval

Not applicable.

Consent

Written informed consent was obtained from the patient for the publication of this case report and accompanying images. A copy of the written consent form is available for review by the editor-in-chief of this journal upon request.

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Author contribution

All the authors individually contributed to manuscript writing, data collection, and reviewing and did the final proofreading of the manuscript before submission.

Conflicts of interest disclosure

The authors declare no conflicts of interest.

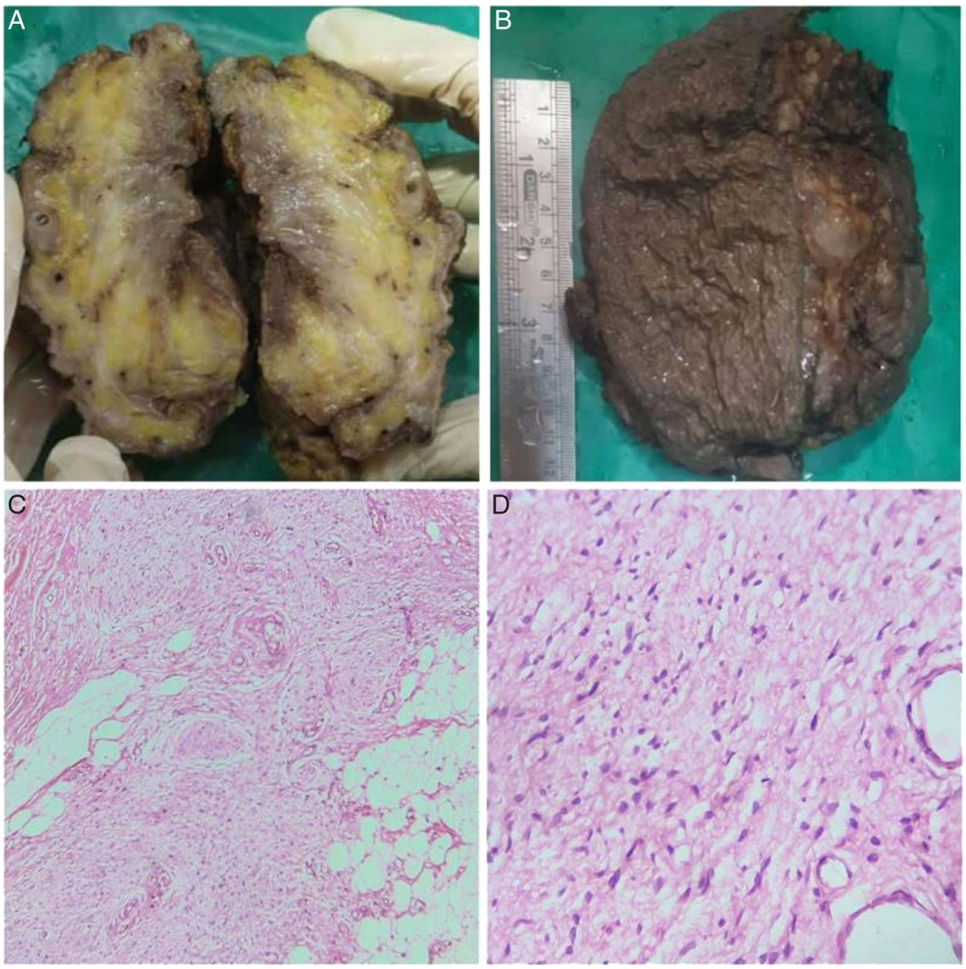

Figure 5. (A) Cut section of biopsy specimen, (B) biopsy specimen, (C and D) Histopathology images – vascular channels of varying caliber admixed with mature adipose tissue, neural elements in the form of bundles, and spindle cell loose stroma.

Research registration unique identifying number (UIN)

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Data availability statement

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