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Case Report

Successful Limb-Sparing Wide Excision of a Giant Lipofibromatosis in a 5-Year-Old Girl

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Keywords
Lipofibromatosis · Limb salvage · Amputation

Abstract
A 5-year-old girl presented with a big painless mass, sized 24 × 37 × 35 cm, in her lower left limb. MRI revealed a huge heterogeneous mass splaying from the left distal femur to the calcaneal region without bony erosion but compressing the arteries and causing bowing of the left tibia and fibula bones. The difficulty was to determine the best course of action taken which would either be limb salvation or amputation. Considering that only a few muscles could be saved, the author initially recommended amputation but still considered a limb-sparing procedure. After a double set-up examination in the operating room, the author ultimately decided to save the affected limb. The salvaged limb was found to be viable after the surgery, and there was no further recurrence over a subsequent 6-month follow-up period. The careful surgical decision is vital in giving the best possible care to the patient.

KARGER

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Introduction

Lipofibromatosis is a rare benign pediatric tumor, generally presenting as a slow-growing, painless mass in infants and young children [1]. Males are mostly affected, with a male to female ratio of 2:1 [2]. A hypothesis of complex translocation of chromosome 4, 6, and 9 was shown to be the culprit in pediatric liposarcoma [3].

Management of the tumor includes observation or excision. Until now, choosing between treatments for lipofibromatosis has remained controversial, although many authors have recommended complete surgical excision [4]. In a report by Teo et al. [1] in 2005, wide excision of extensive upper limb lipofibromatosis in a 1-day-old female caused nonviable upper limb after surgery. As wide excision may cause further comorbidity, treatment needs to be individualized based on the clinical presentation [5].

This case report presents the difficulty in determining whether to amputate or salvage the affected limb. A double set-up examination was carried out to ensure the best surgical option for the patient. During surgery, vessels, nerves, muscles, and bone could still be saved, rendering the choice to perform a limb-sparing procedure. As hoped, the saved limb was still functional after several courses of physiotherapy.

Case Report

A 5-year-old girl presented to an outpatient clinic with a painless mass in her left lower limb that had slowly enlarged since she was 3 months old (Fig. 1). She was otherwise healthy and the limb was still functioning normally. Nevertheless, the patient walked with foot-dragging gait due to the large tumor size. She had no prior history of surgery or medication. Physical examination demonstrated a firm tumor arising from the left distal femur to calcaneus bone, well-delineated, and not tender with palpation. The patient's neurological status was not compromised. Both the knee and ankle joints were clinically normal. There were no skin lesions or discoloration seen. The initial hematological investigation was within normal limits.

Plain left lower leg radiographs revealed a soft-tissue mass arising from the left distal femur to the calcaneal region. The tibia and fibula bones were bowed, but no bone destruction was shown. To examine the condition of the soft tissues and vascularity, magnetic resonance imaging (MRI) was obtained. MRI revealed a 24 × 37 × 35 cm soft-tissue mass located at the lower left leg (Fig. 2). On T1-weighted image, the intensity of the large lesion was found to be heterogeneous, corresponding to fat and fibrous signal intensity. There were enlargements of gastrocnemius, long digital extensor, long digital flexor, extensor hallucis longus, flexor hallucis longus, peroneus brevis, long peroneal, soleus, anterior tibial, posterior tibial, and semimembranous muscles. This giant mass compressed the arteries around it and caused the bowing of the tibia and fibula bones.

She underwent incisional biopsy under general anesthesia. The histopathological diagnosis was lipofibromatosis. No malignancy feature was found. Due to the rarity of similar cases, the treatment had to be approached with caution as there are no standard models of treatment to be followed.

Considering that only a few muscles could be saved, the author initially recommended amputation but still considered a limb-sparing procedure. The final decision was made after a double set-up examination in the operating room. After evaluation of intact vital struc-

tures, during surgery, the author found that the tumor compressed the muscles but did not adhere to the skin. Finally, the author decided to carry out wide excision, sparing the limb.

The tumor was successfully removed completely by separating the healthy unaffected muscles that surrounded the bones without any significant functional compromise. The deep popliteal veins and their branches were anatomically intact. The superficial veins were fully damaged. The tibial, peroneal, and sural nerves were intact. Bleeding occurred in the patient (450 mL; weight 19.2 kg including the weight of tumor), resulting in anemia (Hb 6.8 g/dL). After the surgery, she was managed in an intensive care unit for 2 days for anemia correction and monitoring due to bleeding. After 5 days in hospital, she was discharged. In order to observe and avoid any possibility of a recurrence, regular follow-up in an outpatient clinic was recommended.

Macroscopic findings of excised tumors are generally described as yellow or brown/white with a firm texture. The microscopic histopathology finding showed an admixture of mature adipose tissue and fibroblastic elements, revealing lipofibromatosis (Fig. 3). No immunohistochemistry or molecular analysis was performed.

Postoperative follow-up was uneventful. This patient was referred to physiotherapy to avoid any gait difficulties. Cosmetically, the bone structure was not completely normal. Two weeks after surgery, the child was able to commence walking without any gait difficulties. During the 6-month follow-up, the patient's condition was still favorable without any signs of relapse or complication. The patient was asked to do regular follow-up after discharge. No recurrence or lesion was reported 6 months after surgery.

Discussion

Lipofibromatosis was first described as a separate entity by Fetsch et al. [6] in 2000 in 45 infants. The lesion is usually sized 1–3 cm, with an average size of 2 cm. Most lipofibromatosis cases usually involve subcutaneous or deep soft-tissue infiltration with possible entrapment of vessel, nerves, skin adnexa, and skeletal muscle. The predilection was found mostly in the trunk and distal extremities [4]. Rare locations have been reported in case series, such as intraoral [7] and retroclavicular [8]. Our patient's MRI scan showed the finding of a lesion consisting almost entirely of fat, consistent with the common histological findings, which is a lesion composed predominantly of adipose lobules [9]. Enlargement of the muscles compressed the surrounding arteries.

Upon physical examination, a solid, well-defined mass was noted arising from the distal left femur bone to the calcaneus bone without neurological compromise. In this patient, the presented giant mass measured a 24 × 37 × 35 cm size and is probably the first case of this size. In this case, delay in seeking treatment caused the huge enlargement of the mass. This happened due to difficulty of health facility access as well as lack of education of the patient's family. The lesion compressed its surroundings, resulting in the bowing of the tibia and fibula bones. Surprisingly, the patient could walk well with the exception of the exaggerated lifting of the affected limb due to the progressively increasing mass of the tumor. In 2014, Joseph et al. [9] also reported a lipofibromatosis mass causing bowing of the tibial midshaft and gait abnormalities.

Imaging is an adjunct to diagnosing this rare tumor. In this case report, a plain radiograph was obtained to evaluate the tibial and fibular bone condition and MRI was obtained to ensure the characteristic of the mass and evaluate the entrapment of nerves and vascularity [10]. The definitive diagnosis of lipofibromatosis is made with histopathology examina-

tion. Typically, lipofibromatosis features are abundant adipose tissue with a spindled fibrous tissue element traversing the adipose tissue in the form of septa [4]. In this case, the specimen showed the admixture of mature adipose tissue and fibroblastic elements, suggesting lipofibromatosis. Some authors performed immunohistochemistry examination to find CD34, CD99, SMA, and BCL-2 that were positive markers for lipofibromatosis [6]. In spite of this, the authors stated that immunohistochemistry examination was nonspecific and generally not useful in diagnosing lipofibromatosis [11]. The immune profile of the patient was not observed in this case.

In this case, the author was confronted with the difficulty in determining whether to amputate or salvage the affected limb. Since only a few muscles could be saved, the author initially recommended amputation but still considered the possibility of limb-sparing surgery because the patient was still able to walk, with no bone destruction and nerve damage. During surgery, vessels, nerves, muscles, and bone could be saved rendering the choice to perform a limb-sparing procedure. As hoped, the saved limb was still functional after several courses of physiotherapy.

In the only 6 patients with lipofibromatosis reported from 2000 to 2012, Boos et al. [12] showed that the tumor sizes ranged from 1 to 2 cm in diameter, and after managing the tumor with excision the recurrence rate was 33%. As a high recurrence rate was reported due to the difficulty of obtaining clear margins in wide excision, a follow-up needed to be carried out. According to Fetsch et al. [6], risk factors for recurrence included congenital onset, male sex, tumor location in the hands and feet, incomplete excision, and mitotic activity in the fibroblastic component on histology. In this case report, a 6-month follow-up was favorable and no recurrence lesion was shown on MRI examination.

In summary, although many documented cases of this entity have been published, insufficient clinical experience in treatment is still the main concern. Hence, with publishing this case, the author hopes to broaden the knowledge in lipofibromatosis and provide the data of successful limb-sparing excision for giant lipofibromatosis of the lower limb. The careful surgical decision is vital in offering the best possible care to the patient. Finally, a routine follow-up is important in monitoring and preventing a recurrence in this patient.

Conclusion

The management of lipofibromatosis was carried out after a double set-up examination in the operating room. The salvaged limb was found to be viable after the surgery, and there was no further recurrence over a subsequent 6-month follow-up period.

Statement of Ethics

The author has no ethical conflicts to disclose.

Disclosure Statement

The author declares that this study has no relevant financial interest.

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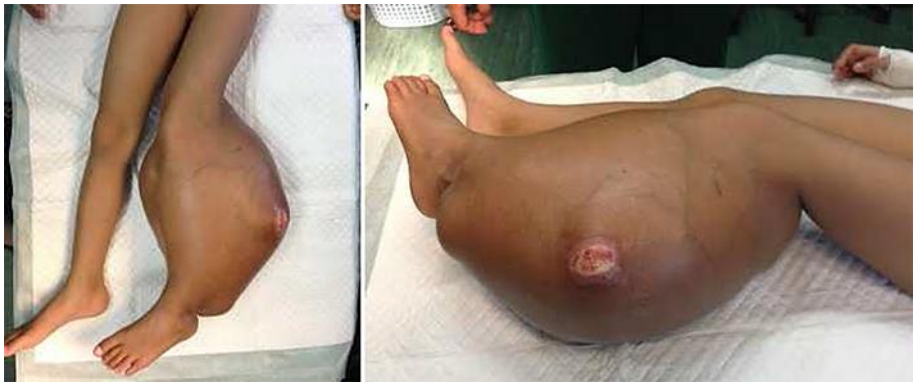

Fig. 1. The patient presented with a large painless soft-tissue mass. The picture was taken after incisional biopsy.

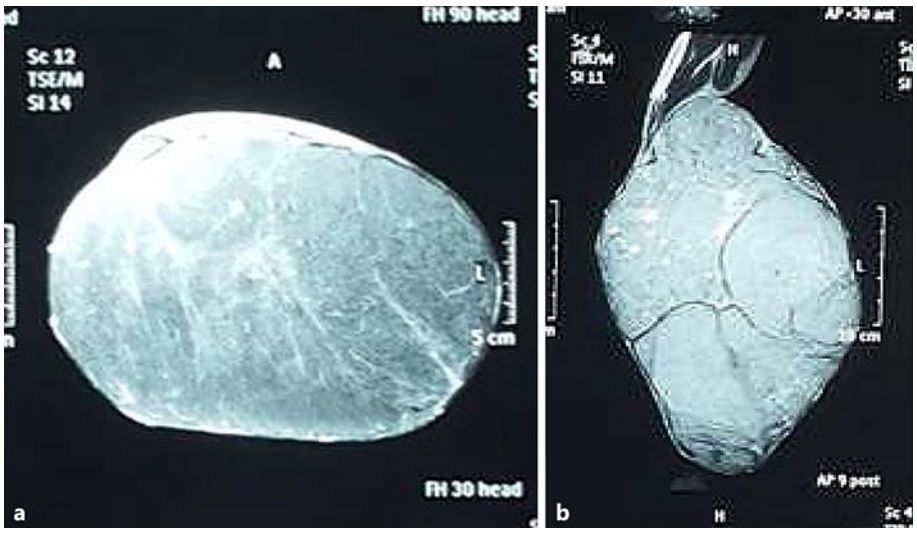

Fig. 2. T1-weighted axial (**a**) and STIR coronal (**b**) MR images showed heterogeneous intensity, containing mainly fat and also fibrous tissue.

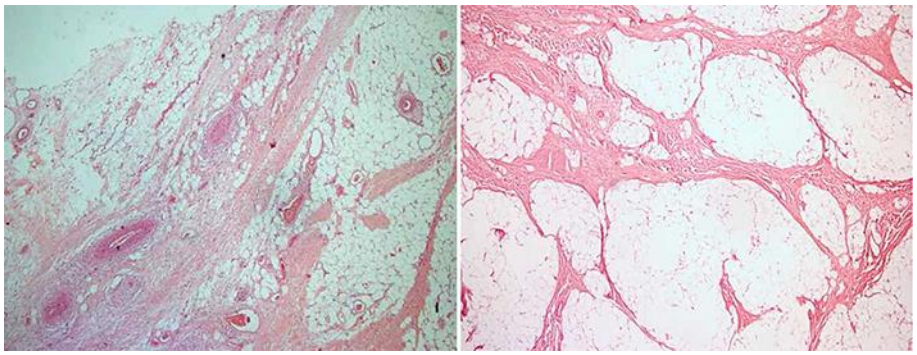

Fig. 3. Hematoxylin and eosin staining, high-power magnification (×100), showing a fibrous and fatty lesion infiltrating skeletal muscle.