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Case Report

Macrodystrophia lipomatosa of the right foot: a case report and treatment

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Abstract

Macrodystrophia lipomatosa (MDL) is a rare congenital disorder characterized by localized gigantism of one or several digits. It may involve the entire limb due to progressive overgrowth of all mesenchymal elements with an excessive increase in the fibro-adipose tissues. It occurs most commonly in the lower limbs. It comes to clinical attention for cosmetic reasons, mechanical problems secondary to degenerative joint disease, or the development of neurovascular compression. We report a case of MDL of the right foot with difficulty walking and wearing footwear. With a complete radio-clinical workup and history review, a provisional diagnosis of MDL was made, confirmed by histopathology and during surgery.

Level of Evidence V; Therapeutic Studies; Expert Opinion.

Keywords: Diagnosis, differential; Foot deformitie; Lipomatosis; Ultrasonography.

Introduction

Macrodystrophia lipomatosa (MDL) is a very uncommon congenital condition that causes localized gigantism in one or more digits, or perhaps the entire limb, due to a gradual overgrowth of all mesenchymal elements and an excessive increase in the fibro-adipose tissues, associated with fibrosis[1]. Few cases of MDL have been reported till now. We describe another anomaly that only affects the right foot, emphasizing the clinical characteristics, alternative diagnosis, and management plan.

Case description

A 16-year-old boy presented progressive enlargement on the second and third toe of the right forefoot since birth. He complained of difficulty in walking and wearing footwear. However, he had no complaints of pain and or ulceration. History of removal of ipsilateral second toe three years back was reported. No record of similar complaints among the family members.

On physical examination (Figure 1)

1. Hypertrophy of the plantar aspect of the right forefoot (Discrepancy of 4 cm circumference compared to the left foot). The plantar swelling was soft to firm in consistency;
2. Hypertrophy of the third toe;
3. Scar mark of the previously amputated second toe;
5. No neurovascular deficit.

Investigations

Radiographs (Figure 2)

1. Increased translucency of plantar aspect suggestive of soft tissue hypertrophy;
2. Enlargement of the third toe phalanges and metatarsal;
3. Lateral deviation of the third digit at the metatarsophalangeal (MTP) joint;
4. Amputated second toe at metatarsal neck level

Study performed at the Vijayawada, Andhra Pradesh, India.

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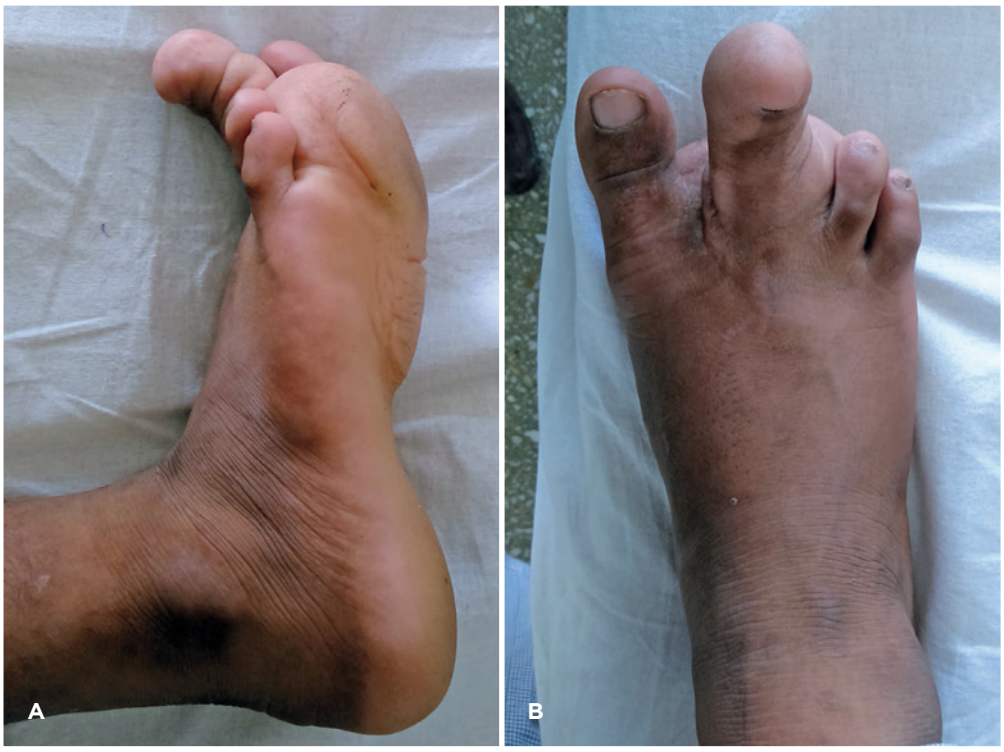

Figure 1. Preoperative clinical photos showing enlargement of the plantar aspect of the right forefoot (A) and enlargement of the third toe (A and B) of the right foot.

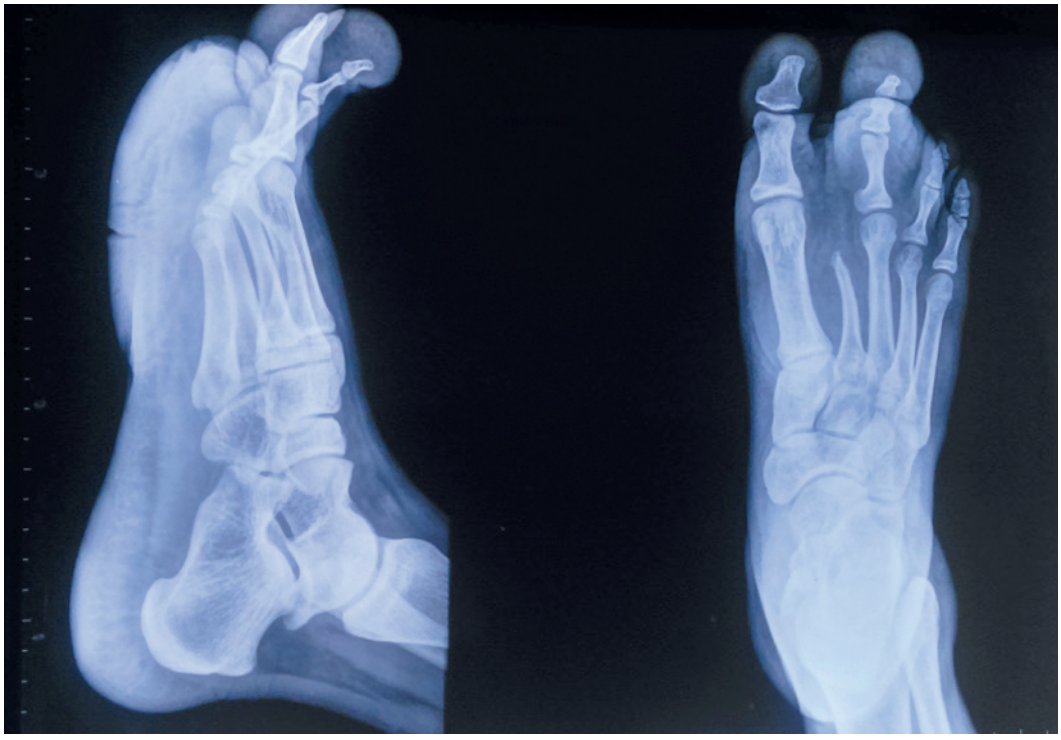

Figure 2. Preoperative radiograph. Lateral view and Anteroposterior view.

Ultrasound

Ultrasound on grayscale and Doppler images showed generalized soft tissue thickening without aberrant blood flow or calcifications.

Histopathology

Histopathology analysis was performed after the first stage debulking procedure. The results showed adipose tissue with dispersed delicate, thread-like fibrous tissue significantly increased.

Diagnosis

Based on the clinical and radiological findings, it was diagnosed as an MDL. After the histological findings, the diagnosis was confirmed.

Management

Line of treatment in this case.

1. First stage debulking procedure of the plantar aspect of the foot (Figure 3);

The surgical procedure was performed under spinal anesthesia after applying a tourniquet. In the first stage, debulking of the enlarged plantar aspect of the right foot was performed after keeping an adequate skin tag for closure. During surgery, excessive fibro-adipose tissue was noted.

2. Second stage excision osteotomy of third toe proximal phalanx (Figure 4).

The surgery was also conducted under spinal anesthesia after applying a tourniquet. As a proximal phalanx of the third toe was enlarged, an excision osteotomy was planned after comparing it with the opposite foot radiograph. The incision

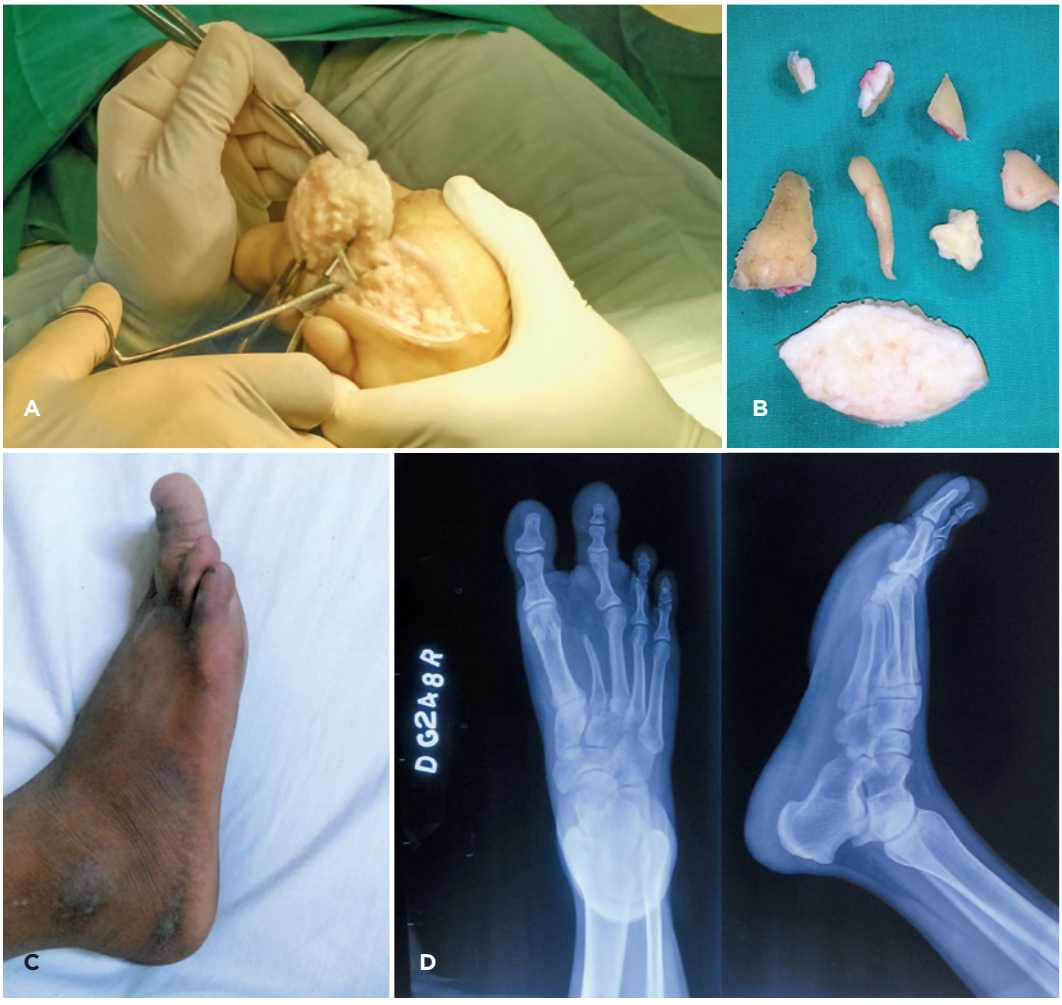

Figure 3. (A) Debulking procedure of the right foot sole; (B) Debulked soft tissue sample; (C) Clinical photo after first stage debulking procedure; (D) Postoperative radiograph showing decreased soft tissue translucency compared to preoperative.

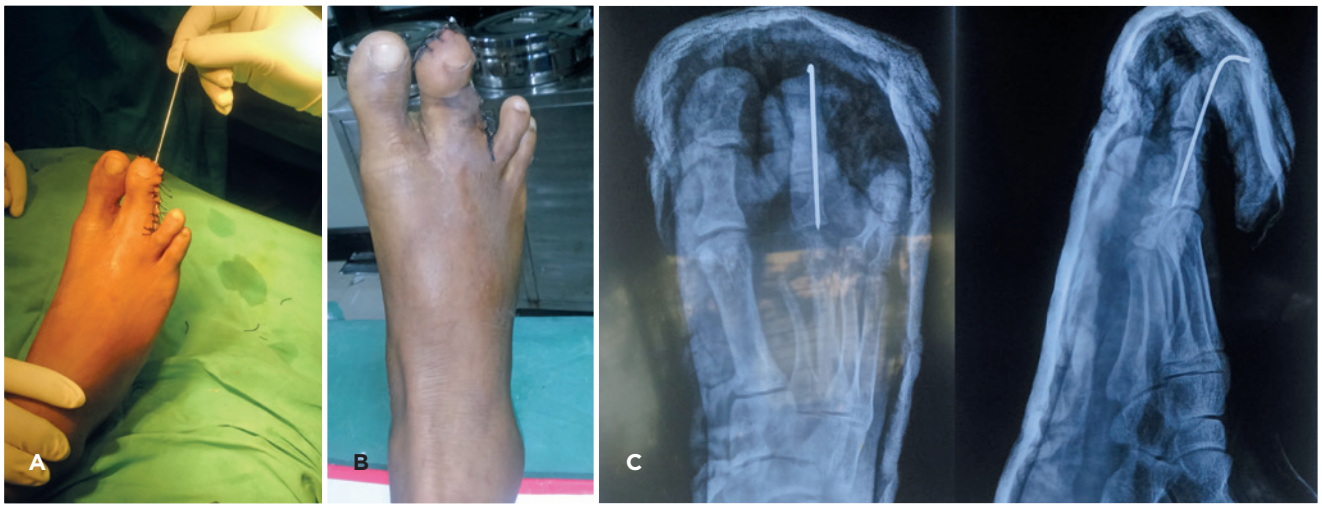

Figure 4. (A) Kirschner wire fixation after second toe proximal phalanx osteotomy; (B) Postoperative clinical photo after second stage surgery; (C) Postoperative radiograph after proximal phalanx osteotomy with Kirschner wire in situ.

was made on the lateral side of the third toe, and a bony fragment was removed from the proximal phalanx shaft. After the osteotomy, the proximal and distal fragments were fixed with a 2 mm Kirschner wire. The wound was closed in layers. A plaster of Paris slab was applied below the knee for six weeks.

Discussion

Although this disease is typically diagnosed in newborns, problems usually arise as the child ages. There is no sex predilection of this anomaly, but the association of the PIK3CA gene may be seen[2]. Unilateral foot involvement affecting the second and third toe is more commonly seen in this condition.

Despite functional issues, such as trouble in grasping or walking, surgical consultation is typically taken for aesthetic rather than functional reasons. In our case, the patient had difficulty walking and could not wear the same size shoes on both feet, yet cosmesis was the patient's priority.

According to Southard et al.[3], some patients experience affected limb pain due to the rising size, shape, and number of Pacinian corpuscles.

Imaging studies are essential to diagnose this anomaly because they can identify the kind of hypertrophied tissue present. Simple radiographs might show soft tissue enlargement and skeletal abnormalities. The characteristic radiolucencies reveal the lipid composition of the soft tissue. According to Soler et al.[4], magnetic resonance imaging is the preferred diagnosis technique for MDL, which demonstrates excessive fibro-adipose tissue with fibrosis and associated abnormal growth of other mesenchymal tissues.

A large amount of adipose tissue is evident in histopathological results, dispersed throughout a delicate, mesh-like fibrous structure[5]. Involved tissues include bones, muscles, nerves, and subcutaneous tissues. Even though there are several histological alterations, the exact pathophysiology is unknown.

However, a few potential mechanisms have been proposed, including segmentation defects, abnormal fetal circulation, growth-inhibiting hormone abnormality, and fat cell degeneration[6].

Differential diagnosis frequently includes proteus syndrome, fibrolipomatous hamartoma of nerve, Klippel-Trenaunay-Weber syndrome, neurofibromatosis type I, lymphangiomatosis, hemangiomatosis, and lymphangiomatosis.

Neurofibromatosis is the most challenging condition to differentiate from MDL among them. However, hereditary history and specific skin lesions like café-au-lait spots and nodules can help to diagnose neurofibromatosis.

Furthermore, MDL can be differentiated from diseases of similar features by negative family history, bony enlargement, and adipose tissue deposition in nerves, muscles, and tendons with macrodactyly, which are characteristic features of this disease. According to Tatu et al.[7], hypertrophy or atrophy of bony and cartilaginous components with interphalangeal joint ankylosis and exostosis may be seen. Basophilic degradation, hyalinosis of collagen tissue, and subcutaneous nerve enlargement with medullary cavity invasion by fat cells are also characteristic findings.

The preferred treatment course for MDL is surgical. The main surgical goal is to make them seem better cosmetically while retaining as much neurologic function as possible. Good outcomes can be achieved with selective and well-planned partial amputations and numerous debulking surgeries. However, if the deformity is not severe and there are no signs of neurological involvement, surgery should be postponed until the patient grows.

Nerve injury is a potential side effect of excessive debulking operations; the published incidence is between 30% to 50%. Treating MDL is difficult due to the local recurrence rate between 33% and 60%[8]. Using debulking and proximal phalanx osteotomy, in our case, we achieved a satisfying result regarding aesthetic and functional outcomes. We operated on the patient with the first stage debulking procedure and second stage osteotomy four weeks apart for proper planning of the bony procedure after the soft tissue procedure, better wound healing, and reduced extensive surgery-related pain and complications. However, we advised the patient for regular follow-ups to determine whether the condition is static or progressive.

Macrodystrophia lipomatosa is the progressive enlargement of soft and bony tissues, causing mechanical and cosmetic problems. A rare disorder should be identified early for prompt intervention and functional improvement. The basic surgical strategy to treat this condition is enhancing the aesthetic look while maintaining neurological function. Good outcomes can be obtained with prudent, well-planned debulking procedures and partial amputation. Surgery should be planned following the patient's maturity, severity, and neurological involvement.

Author' contributions: The author contributed individually and significantly to the development of this article: JD *(https://orcid.org/0009-0008-4691-2087) Conceived and planned the activities that led to the study, interpreted the results of the study, participated in the review process, performed the surgeries, bibliographic review, survey of the medical records. The author read and approved the final manuscript.*ORCID (Open Researcher and Contributor ID) .

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