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Case Report

Surgery Section

Macrodystrophia Lipomatosa of the Toe: A Rare Case Report

ALAGAR RAJA DURAIRAJ[1], SURYA RAO RAO VENKATA MAHIPATHY[2]

ABSTRACT

Macrodystrophia lipomatosa is a rare congenital hamartoma presenting as a localized or generalized gigantism of a limb or digit manifesting from infancy to late adulthood. It is a progressive enlargement of the soft tissue components, especially fibrofatty tissue. The patient presents to us because of cosmetic reasons or mechanical issues secondary to degenerative joint disease, or features of neurovascular compression due to large osteophytes. Here, we present a case of this anomaly of the left second toe with complaints of difficulty in walking and wearing shoes, for which toe reduction surgery with partial amputation was done.

Keywords: Congenital, Cosmetic, Digit, Hamartoma

CASE REPORT

A five-year-old presented to us with an enlarged deformed left second toe since birth. There was history of difficulty in walking and wearing footwear. On examination, there was a macrodactyly of the left second toe with a gross lateral deviation. The nail plate complex was normal. All other toes were normal [Table/Fig-1,2].

Both upper limbs were normal and there was no evidence of any other congenital abnormality investigated by chest X-ray, ECG, echocardiogram and an ultrasound of the abdomen. There were no dysmorphic facial features. X-ray of the foot revealed a left second toe with enlarged, broad and splayed phalanges with an excess of soft tissue components [Table/Fig-3].

CT revealed macrodactyly due to hypertrophy of fibrofatty soft tissues with mild bony hypertrophy of distal and middle phalanges of left second toe with lateral angulation of toe with no vascular hypertrophy in the region, suggestive of macrodystrophia lipomatosa. After explaining the condition and the consequences of treatment, we proceeded for surgery. Under general anaesthesia, incision was made on the neutral line of medial aspect of left second toe and deepened in layers [Table/Fig-4].

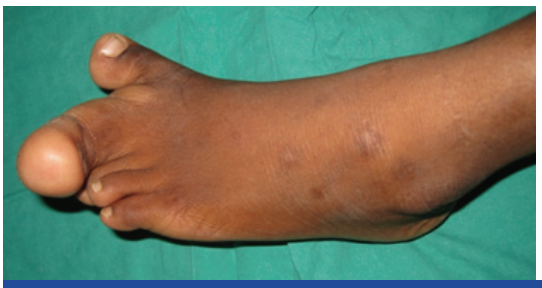
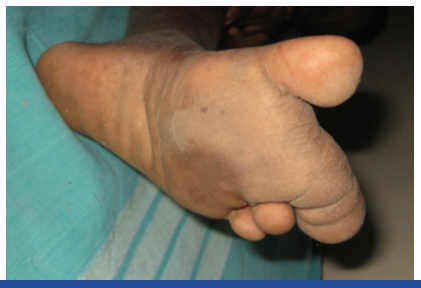
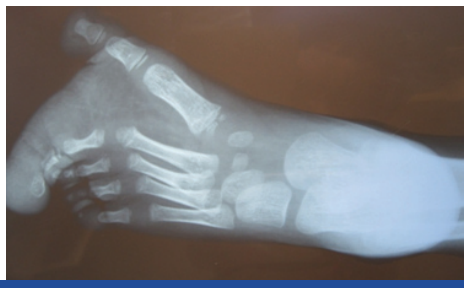
Toe reduction surgery was done by excising large amount of lipomatous tissue and partial amputated at proximal interphalangeal joint. The excess skin was trimmed and closure was achieved using a fillet flap [Table/Fig-5].

Postoperative period was uneventful and patient was discharged on the 10th post-op day [Table/Fig-6].

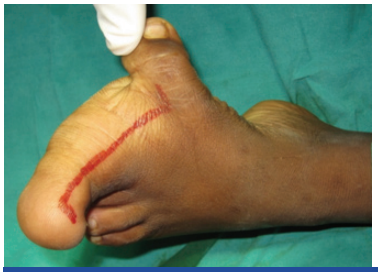
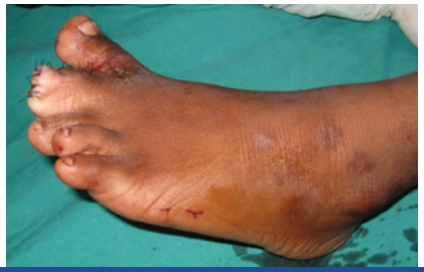
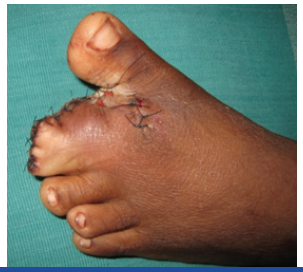
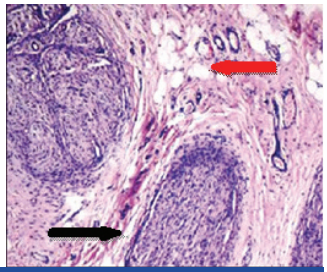
Histopathogy showed lobules of mature adipocytes separated by dense fibrocollagenous stroma with few congested capillaries and nerve bundles suggestive of a neurolipomatous hamartoma, clinically correlating with macrodystrophia lipomatosa [Table/Fig-7].

DISCUSSION

Macrodystrophia lipomatosa is a rare type of non-hereditary congenital localized gigantism characterized by progressive hypertrophy of mesenchymal elements with a disproportionate increase in the amount of fibrofatty tissue [1]. It is usually present during infancy, but causes problem as the child grows. The term was coined by Feriz in 1925 [2]. It typically presents as a painless enlargement of the 2nd or 3rd finger or toe. It is usually unilateral but may involve adjacent digits in the distribution of the median or plantar nerves and at times can involve the entire limb [2]. The disorder is almost always unilateral, with an equal sex ratio [3]. Macrodystrophia lipomatosa is associated with other congenital

[Table/Fig-1]: Pre-operative photograph (dorsal view). **[Table/Fig-2]:** Pre-operative photograph (plantar view). **[Table/Fig-3]:** Plain X-ray left foot showing lateral deviation and soft tissue hypertrophy of second toe with elongated and splayed phalanges.

[Table/Fig-4]: Photograph showing the skin incision. **[Table/Fig-5]:** Immediate postoperative photograph. **[Table/Fig-6]:** 10th postoperative day. **[Table/Fig-7]:** Microscopic view showing thickened nerve bundles (black arrow) with intervening adipose tissue (red arrow). H & E, 40 X.

limb anomalies like syndactyly, brachydactyly, clinodactyly and polydactyly [1]. A distinguishing characteristic in macrodactyly of the hand, which is absent in cases affecting the foot, are hypertrophy and tortuosity of the digital nerves [4]. The soft tissue hypertrophy is most marked at the volar aspect of the distal ends of the digits resulting in a dorsal angulation of the involved digit. Progressive macrodactyly is more common than the static type where growth of the affected parts is faster than the rest of the body. Even though macrodystrophia lipomatosa is considered a progressive form of macrodactyly, growth stops at puberty [5]. The disease may present with functional problems, such as difficulty in grasping or gait, but surgery is usually sought for cosmetic rather than mechanical reasons [6]. Our patient had difficulty in walking mainly due to a severely deformed second toe, but the main concern of the patient and his parents was cosmesis.

A number of imaging modalities, such as plain radiography, USG, CT scan, and MRI, have a definite role in the evaluation of macrodystrophia lipomatosa. Plain radiography reveals skeletal and soft tissue hypertrophy, with fat translucencies in the soft tissue. The phalanges are enlarged and splayed, appearing as a mushroom, with endosteal and periosteal bone deposition, more commonly of the distal phalanx. The overlying soft tissues are markedly overgrown, especially in the volar and distal aspects. Within the soft tissues focal lucent areas representing fat may be seen, which is characteristic and are indicative of the fatty nature of the soft tissue. Decreased joint space, subchondral bone cysts, and osteophytes are the secondary osteoarthritic changes that occur during the second decade of life [6]. Ultrasonogram and CT show the proliferation of fat along the nerve territory [7,8]. MRI demonstrates the excess fibrofatty tissue, which has signal characteristics similar to subcutaneous fat, i.e., high signal on T1W, intermediate signal on T2W, and low signal on fat-suppressed sequences. MRI demonstrates accumulation of fat in the subcutaneous tissues without an obvious capsule [9]. Soler et al., proposed that MRI should be used to detect fibroadipose tissue masses with proportional enlargement of other mesenchymal tissues as the diagnostic method of choice for macrodystrophia lipomatosa [10].

Macrodystrophia lipomatosa is characterised by a marked increase all mesenchymal elements [9]. Microscopy shows a large amount of adipocytes interspersed in flimsy sieve-like fibrous tissue [11]. Other structures involved include subcutaneous tissues, nerve sheaths, muscles, periosteum, and bone marrow. The aetiopathogenesis is not clearly defined, but the proposals include fatty degeneration, altered fetal circulation, changes in growth inhibiting factors, and segmentation problems [12].

There are many conditions mimicking macrodystrophia lipomatosa which include neurofibromatosis type I, vascular malformations & their syndromes, neurofibrolipomatosis, and Proteus syndrome. The most difficult to differentiate from macrodystrophia lipomatosa is neurofibromatosis, which is diagnosed by a positive family history and characteristic cutaneous features like café-au-lait spots on the skin and soft tissue nodules. On the other hand, macrodystrophia lipomatosa has features of bony overgrowth and deposition of adipose tissue in subcutaneous tissues, tendons, muscles, and nerves.

The mainstay of treatment for macrodystrophia lipomatosa is surgery. Surgical excision improves the cosmetic appearance while maintaining the neurologic function. Superior results are obtained by meticulous planning, function preserving debulking surgeries and partial amputations. A high recurrence rate of about 33% to 60% makes the management of macrodystrophia lipomatosa difficult warranting regular follow-up [7].

CONCLUSION

Macrodystrophia lipomatosa is a progressive hamartoma with hypertrophy of fibrofatty tissue of all the layers of soft tissue causing local gigantism. A thorough clinical examination, imaging and histopathology are crucial in clinching the diagnosis. The treatment is surgical but the results are variable.

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PARTICULARS OF CONTRIBUTORS:
1. Associate Professor, Department of Plastic Surgery, Saveetha Medical College Hospital, Thandalam, Kanchipuram, Tamil Nadu, India.
2. Associate Professor, Department of Plastic Surgery, Saveetha Medical College Hospital, Thandalam, Kanchipuram, Tamil Nadu, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Surya Rao Rao Venkata Mahipathy,
Associate Professor, Department of Plastic & Reconstructive Surgery, Saveetha Medical College Hospital, Thandalam, Kanchipuram Dist. 602105, Tamil Nadu, India.
E-mail: surya_3@hotmail.com

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