Macrodystrophia Lipomatosa of the Foot: A Case Report

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ABSTRACT

Introduction: Macrodystrophia lipomatosa is a rare congenital sporadic form of focal gigantism. It is characterized by proliferation of all messenchymal elements of the second and third digit of the lower limb with volar and distal predominance.

Case report: Here, we report a case of 8 years old male child who presented with congenital, painless, gradually progressive enlargement of the second and third toe of the left foot. Radiograph and MRI features are suggestive of Macrodystrophia lipomatosa.

Conclusion: It is to be differentiated from other causes of local gigantism since these conditions differ in progression, prognosis, complications and management.

Keywords: Macrodystrophia, Lipomatosa, Foot

INTRODUCTION

Macrodystrophia lipomatosa is a rare congenital non hereditary form of focal gigantism characterized by progressive overgrowth of all the messenchymal elements including bones, skeletal muscle, nerve sheath and fat.[1] The overgrowth appears in specific sclerotomes most commonly in the region supplied by medial plantar nerve, that is first to third toes of lower limb.[2] Exact aetiology is not known but the endosteal and periosteal deposition of the bone is thought to be the pathogenesis of bony enlargement.[3] Imaging has vital role in diagnosis.[2]

CASE REPORT

A 8 years old male child presented with history of enlargement of the second and third digits of the left foot since birth. The enlargement was painless and progressively increasing. The child was a first-born child of non-consanguineous marriage and didn't have any significant prenatal or perinatal history. There was no family history of similar complaints or congenital anomalies. On examination, the second and third toes showed focal gigantism (second> third toe) with upturned second toe due to verrucous soft tissue overgrowth on the plantar aspect (Figure 1 a). Distal neurovascular status was intact and there was no pigmentation, ulceration or palpable thrill. Rest of the left foot, vitals and general physical examinations were normal.

Imaging was performed to assess the current status of the affected phalanges and the adjacent soft tissues before planning constructive surgery.

Radiographs of the foot showed gross increase in the soft tissues around the second and the third toes. The soft tissue overgrowth was marked more in the volar and the distal portion of the toes. There were focal lucent areas within the soft tissues, suggestive of fat deposition. Splaying, lengthening and broadening of the phalanges of the second and third toes were noted. (Figure 2 a and b).

MRI of the foot revealed asymmetrical enlargement of both the bony phalanges and subcutaneous soft tissues with splaying of the second and third toes. The fibrofatty tissues at the dorsal pulp of the toes were prominent giving mushroom like appearance. It was non-capsulated and there were no features of underlying vascular malformation. There was no evidence of invasion of the fatty tissue in the bone. The involved phalanges were otherwise normal in signal characteristic and morphology (Figure 3 a, b, c).

DISCUSSION

The term Macrodystrophia lipomatosa was coined by Feriz in 1925.[4] It is a rare congenital disorder of focal gigantism characterized by overgrowth of all the messenchymal elements (Soft tissues, fat, bone and muscle) of the digits.[5,6] It occurs most frequently in the distribution of plantar nerves in the lower extremity and in the distribution of median nerve in the upper extremity.[2] There is higher male

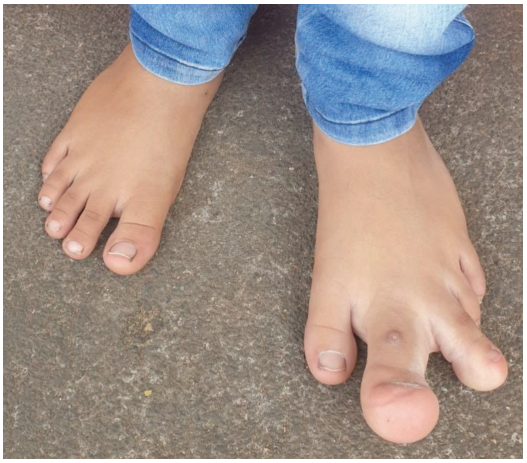

Figure-1: Photograph showing enlargement of the second and third toe of left foot

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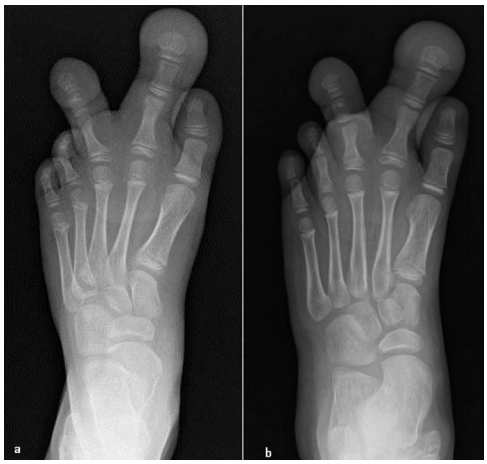
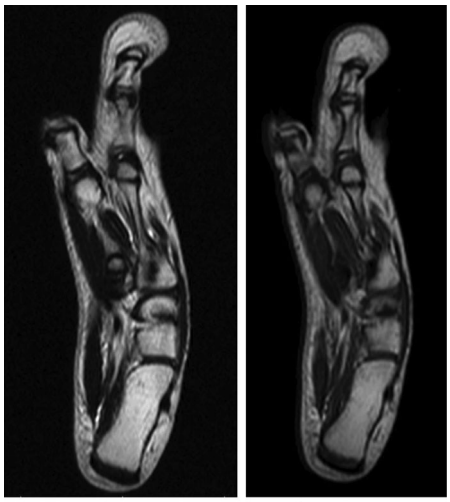
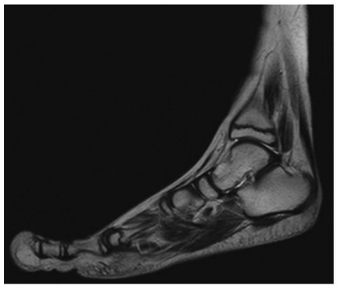


Figure-2: a, b Radiograph showing gross soft tissue swelling, lengthening and broadening of the phalanges of the second and third toes



a T1 Coronal b T 2 Coronal



c T 2 Sagittal

Figure-3: (a) T1 Coronal, (b) T 2 Coronal, (c) T 2 Sagittal MR images showing asymmetrical enlargement of both the bony phalanges and subcutaneous soft tissues

predominance.[2,7] Volar and the distal portion of the digits are more commonly affected.[5] The second and third digits of the lower limb are the most commonly affected sites.[6–8] The involvement of lower limb is 15 times more common than the upper limb.[8] Barsky and his colleagues defined two types of this condition. These included the more common static form kind wherein the increase in size of the enlarged digits is proportional to that of the rest of the body and the less common progressive firm type wherein the enlargement of the digit is disproportionate to the growth of rest of the body.[2,7,8]

The exact aetiology is not known. However, the pathogenesis of bone increment is attributable to the endosteal and periosteal deposition of the bone.[2] Various hypothesis proposed for etiopathegenesis include lipomatous degeneration, disturbed fetal circulation in the utero, error in segmentation and damage of extremity buds.[2,6,7]

On the basis of radiographic findings, fibrolipomatous hemartoma is a close differential diagnosis but the presence of bony hypertrophy in macrodystrophia lipomatosa helps in differentiation.[5,7] Also, fibrolipomatous hemartoma usually presents as isolated nerve lesion and associated with intramuscular fat deposition.[7] Radiolucencies present in the soft tissue representing fatty tissues favour the diagnosis of macrodystrophia lipomatosa.[2,5]

The other differential diagnosis includes neurofibromatosis type I, hemangiomatosis, lymphangiomatosis, proteus syndrome and Klippel-Trenaunay-Weber syndrome.[9] These are unlikely in absence of family history.[2] NF has systemic abnormalities in addition to family history. Hemangiomatosis is clinically diagnosed with a palpable bruit and have worm like area of high signal intensity in T2 weighted spin echo imaging. Lymphangiomatosis present with diffuse swelling and pitting edema. Klippel-Trenaunay-Weber syndrome presents with limb hypertrophy, cutaneous hemangioma, varicose vein and AV fistula that can be confirmed by duplex scan and MRI.[2,5–8]

Proteus syndrome typically present with lung cysts, skull abnormalities , lung cysts and dermatological problems like palmar and plantar thickening, pigmented navei etc.[7]

Imaging has vital role in diagnosis of the Macrodystrophia lipomatosa which is confirmed by histopathology.

Medical help is seeked for cosmetic reason more than the mechanical region. Treatment consists of surgical correction by debulking procedures, epiphysiodesis and various osteotomies which are aimed to cosmetic restoration along with preservation of neurological function.[2,7,8]

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