Macrodystrophia lipomatosa: Review of clinico-radio-histopathological features

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ABSTRACT

We report clinical, radiological, and pathological features of three cases of macrodystrophia lipomatosa, which is characterized by progressive proliferation of all mesenchymal elements, with a disproportionate increase in fibroadipose tissue. The peculiarity in our report is the involvement of contralateral hand and feet in our 1st case in contrast to usual presentations of this rare condition, which is often unilateral. In our 2nd case, the lateral aspect of foot was involved as against the usual finding of involvement of medial aspect in lower limbs, also another surprising finding in this case is increase in the size of lesion after puberty. Coming to our 3rd case, enlargement of almost whole of an upper extremity with profound involvement of middle, ring and little finger along with total sparing of the thumb is in itself an extremely rare occurrence as in upper limb, mainly the lateral aspect of hand and related digits bear the burnt and more over involvement of whole limb is itself an unique phenomenon.

Key words: Fibroadipose, localized gigantism, macrodystrophia lipomatosa

INTRODUCTION

Macrodystrophia lipomatosa (MDL), a rare entity which is usually identified in the neonatal period with further progress usually halts at puberty, can be thought of being analogous to static localized gigantism, described by Barsky in 1967.

CASE REPORTS

Case 1

A two-and-a-half-year-old male child presented with grossly enlarged left index finger and thumb along with the enlargement of contralateral great and second toe associated with lateral curvature of the left middle finger since birth. No gait disturbance and no difficulty in playing or other activities [Figure 1ai and aii].

Case 2

A 29-year-old female presented with localized soft tissue swelling involving lateral aspect of left foot and the corresponding lateral three digits since early childhood. According to the patient, lesion has been increasing in size over the last six months causing pain and discomfort during walking and other routine activities. The patient is giving history of once been operated at the age of six years followed by recurrence [Figure 1b].

Case 3

A 10-month-old male child presented with swelling involving the whole upper limb with enormous bulbous enlargement of middle, ring and index fingers of right hand which according to the parents is present since birth [Figure 1c]. In all the 3 cases, local examination revealed normal texture and temperature with a soft doughy feel on palpation without any abnormal pulsations. General examination was unremarkable in all the three cases.

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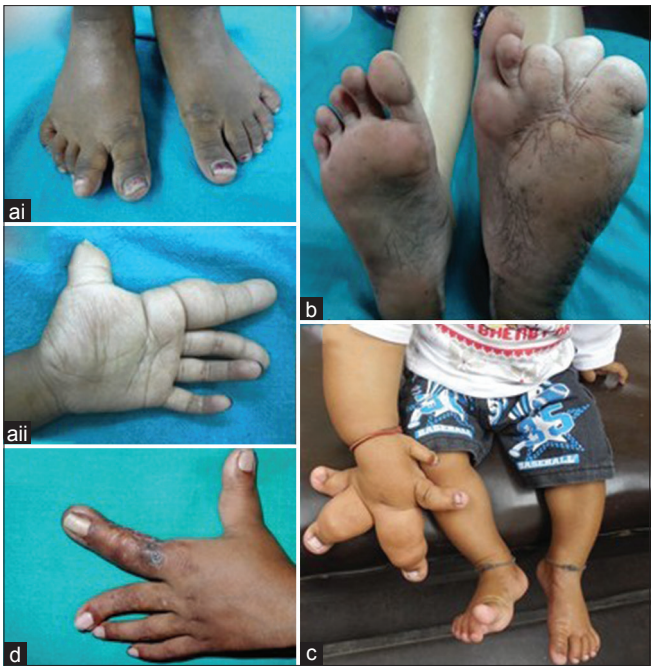

Figure 1: (ai) Macrodystrophia lipomatosa (MDL) over right great and second toe associated with lateral curvature of the left middle finger. (aii) Similar lesions over left index finger and thumb. (b) MDL involving lateral aspect of left foot and the corresponding lateral three digits. (c) MDL involving the whole right upper limb and middle, ring, and index fingers of right hand. (d) MDL over left index finger and thumb after surgical debulking

Radiography

Radiography revealed an increase in the soft tissue mass with concomitant enlargement in both length and breadth of corresponding bones with decreased bone density and trabecular prominence. No soft tissue calcification, phlebolith, expansile lytic lesion, or any evidence of pseudoarthrosis was noted [Figure 2a–c].

Ultrasonography

Ultrasonography (USG) and Colour Doppler study revealed increased echogenic soft tissue component suggestive of fatty tissue without any significant internal vascularity in all the three above-mentioned cases [Figure 3a–c].

Histopathological examination

Histopathological examination (HPE) revealed an increase in fatty tissue in the subcutaneous zone [Figure 4a–c].

A formal diagnosis of MDL was made in all the three cases followed by referral to plastic surgery department for further management. Patients are under regular follow up.

Our first case recently underwent debulking operation with resultant cosmetic improvement without any complications [Figure 1d].

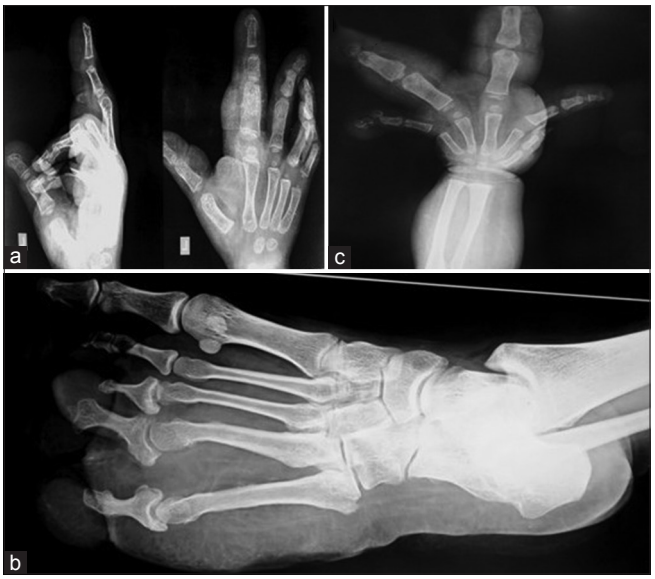

Figure 2: (a) Radiography of left hand revealed increase in soft tissue mass with concomitant enlargement in both length and breadth of corresponding bones with decreased bone density and trabecular prominence. (b) Similar details found in X-ray of left foot. (c) Similar details found in X-ray of right forearm and hand

DISCUSSION

MDL is a rare congenital, nonhereditary condition characterized by localized gigantism involving any part of body but in majority of cases affecting digits and extremities and can be associated with fibrolipomatous hamartoma (FLH) of nerves.[1,2] Rarely it leads to mechanical issues and the main cause of seeking management is cosmetic disfigurement. An interesting finding is because of some yet unknown reason the condition prefers distribution of median and plantar nerves.

The term macrodystrophia lipomatosa was first used by Feriz in 1925 to describe unilateral overgrowth of the lower limb and may be associated with syndactyly, symphalangism, polydactyly, brachydactyly, or clinodactyly.[3] Rarely association can be found with lipomatous growths in intestines, pulmonary cysts, calvarial abnormalities, pigmented nevus, and so on.[4] Various hypotheses such as lipomatous degeneration, errors in the segmentation during limb bud development and disturbed fetal circulation has been given but the exact etiology remains unknown.[5] Other conditions that can be considered in the differential diagnosis are hemangiomatosis, lymphangiomatosis, FLH, Klippel–Trenaunay–Weber syndrome, Mafucci syndrome, Ollier disease, and Proteus syndrome [Table 1].[6-8]

Different imaging modalities, such as plain radiography, USG, computed tomography scan, magnetic resonance imaging, and HPE, all have roles in the evaluation of MDL.

Table 1: Differentiating features between macrodystrophia lipomatosa and other clinical mimickers

Conditions	Macrodystrophia lipomatosa	Proteus syndrome	Neurofibromatosis type 1 (plexiform neurofibroma)	Klippel–Trenaunay–Weber syndrome	Lymphangiomatosis and hemangiomatosis
Family History	Negative	Positive	Positive	Positive	Positive
Caf'e-au-lait spots	Absent	Absent	Present	Absent	Absent
Presence of cutaneous hemangiomas and varicose veins	Absent	Absent	Absent	Cutaneous hemangiomas and varicose veins present	Hemangiomas present, Varicose veins absent
Affection-unilateral or bilateral	Usually unilateral overgrowth	Usually unilateral overgrowth	Usually bilateral affection	Usually unilateral overgrowth	No such preponderance
Presence of palmar and plantar cerebroid thickening	Absent	present	Absent	Absent	Absent
Increased risk of developing certain tumors	Absent	Present	Present	Absent	Absent
Opthalmologic changes	Absent	Present	Present	Absent	Usually Absent

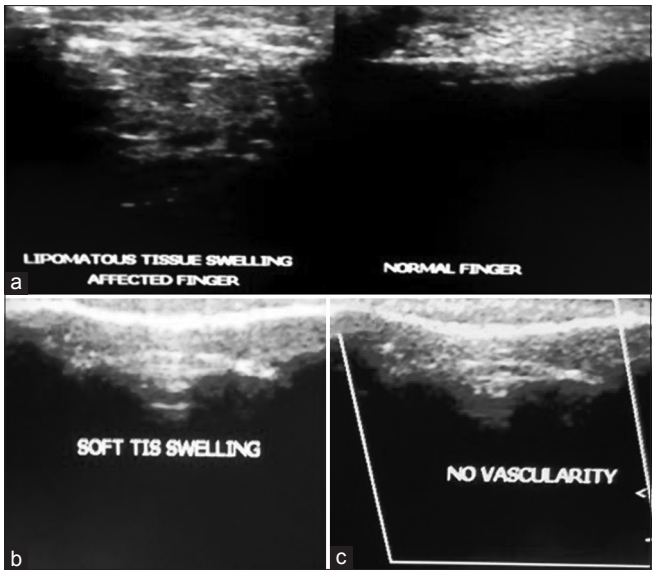

Figure 3: (a) Ultrasonography (USG) and Colour Doppler study of left hand revealed increased echogenic soft tissue component suggestive of fatty tissue without any significant internal vascularity. (b) Similar details found in USG and Colour Doppler study of left foot. (c) Similar details found in USG and Colour Doppler study of right forearm and hand

Plain radiography usually reveals lengthening, broadening, and splaying of phalanges with hypertrophy of soft tissue especially in the volar aspects. Also increased focal translucencies in the soft tissue due to increased adipose tissue can be occasionally appreciated. In Colour Doppler study, apart from an increased subcutaneous fat, the proliferation of fat along the nerve territory can be seen.[1,9] The most prominent histological finding is the increase in adipose tissue in subcutis scattered in a fine lattice of fibrous tissue, which can rarely involve bone marrow, periosteum, muscles, and nerve sheaths. Treatment of choice is surgical debulking procedures but can rarely be complicated with nerve damage. A high recurrence rate of 33%–60% has been seen following surgical treatment.[10]

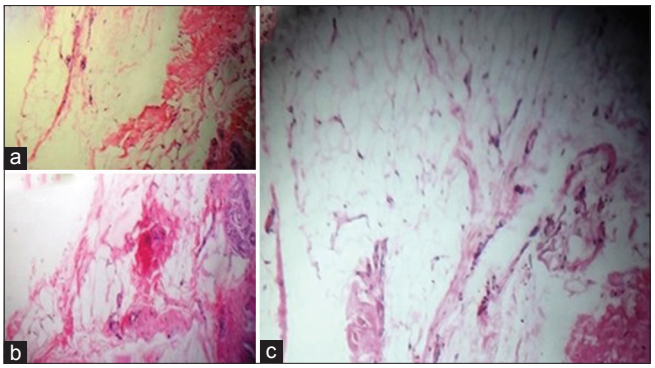

Figure 4: (a) Photomicrograph showing an increase in fatty tissue in the subcutaneous zone. In HPE, a gross increase in subcutaneous fat (H and E ×100). (b) Similar details found in photomicrograph of left foot (H and E ×100). (c) Similar details found in photomicrograph of right forearm and hand (H and E ×100)

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Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Brodwater BK, Major NM, Goldner RD, Layfield LJ. Macrodystrophia lipomatosa with associated fibrolipomatous hamartoma of the median nerve. PediatrSurgInt 2000;16:216-8.
2. Singla V, Virmani V, Tuli P, Singh P, Khandelwal N. Case report: Macrodystrophia lipomatosa – Illustration of two cases. Indian J Radiol Imaging 2008;18:298-301.
3. Gupta SK, Sharma OP, Sharma SV, Sood B, Gupta S. Macrodystrophia lipomatosa: Radiographic observations. Br J Radiol 1992;65:769-73.
4. Silverman TA, Enzinger FM. Fibrolipomatous hamartoma of nerve. A clinicopathologic analysis of 26 cases. Am J SurgPathol 1985;9:7-14.
5. Goldman AB, Kaye JJ. Macrodystrophialipomatosa: Radiographic diagnosis. AJR Am J Roentgenol 1977;128:101-5.
6. Lal NR, Bandyopadhyay D, Sarkar AK.Unilateral hypertrophic skin lesions, hemimegalencephaly, and meningioma: The many faces of

Proteus syndrome.Indian Dermatol Online J2015;6:348-51.
7. Krengel S, Fustes-Morales A, Carrasco D, Vázquez M, Durán-McKinster C, Ruiz-Maldonado R. Macrodactyly: Report of eight cases and review of the literature. PediatrDermatol 2000;17:270-6.
8. Khan RA, Wahab S, Ahmad I, Chana RS. Macrodystrophia lipomatosa: Four case reports. Ital J Pediatr 2010;36:69.
9. Curry NS, Schabel SI, Keuper JT. Computed tomography diagnosis of macrodystrophia lipomatosa. J ComputTomogr 1988;12:295-7.
10. Boren WL, Henry RE Jr, Wintch K. MR diagnosis of fibrolipomatous hamartoma nerve: Association with nerve territory-oriented macrodactyly (Macrodystrophia lipomatosa). Skeletal Radiol 1995;24:296-7.