



Case Report

Macrodystrophia Lipomatosa: A rare presentation

Sushil G. Kachewar*[¶] MD DNB, Smita B. Sankaye[†] MD and Devidas S. Kulkarni* MD

*Department of Radiodiagnosis, [†]Department of Pathology, Rural Medical College, PIMS, Loni, Maharashtra, India

(Received 08 November 2010 and accepted 02 December 2010)

ABSTRACT: Macrodystrophia lipomatosa is a rare congenital nonhereditary mesenchymal hamartomatous malformation resulting in localized gigantism of parts of extremities that manifests clinically as macrodactyly or megalodactyly. Radiological and Pathological hallmark is the disproportionate fibroadipose tissue proliferation in subcutaneous tissue, nerve sheaths, and periosteum that leads to soft tissue and bony enlargement. As imaging is the key to early diagnosis, findings on various modalities like plain radiographs, CT scan and MRI are highlighted here.

KEY WORDS: *Macrodystrophia lipomatosa; CT; MRI*

INTRODUCTION

Macrodystrophia lipomatosa is a congenital local gigantism of the hand and foot characterized by proliferation of all mesenchymal components, particularly fibroadipose tissue. Macrodystrophia lipomatosa comes to clinical attention because of cosmetic reasons, mechanical problems secondary to degenerative joint disease or development of neurovascular compression. This report depicts two cases, one involving two fingers of hand with syndactyly and the other involving toes. The aim of presenting this article is to emphasize the importance and adequacy of imaging in early non-invasive diagnosis of this entity.

CASE REPORT

Case 1

A two year old, right hand dominant female child presented with disproportionate overgrowth of the ring and middle finger of right hand with syndactyly since one year of age. She had no pain or neurovascular symptoms. There was no similar family history. Physical examination showed non-tender, soft tissue mass on palmar aspect of the enlarged ring and middle finger of right hand with

dorsal angulation of fingers (**Figure 1**). No skin nodules, cafe-au-lait spots or other anomalies were identified.



Figure 1: Clinical photograph of right hand shows enlarged, fused ring and middle finger (arrow) as compared to the remaining fingers.

Plain radiograph of the right hand revealed enlarged phalanges of the ring and middle finger, together with overgrowth of soft tissue (**Figure 2a**). CT demonstrated excessive subcutaneous fat along the palmar aspect of the ring and middle finger, corresponding to the distribution of the median nerve (**Figure 2b**). Reduction of the subcutaneous fat was attempted mainly for cosmetic reasons.

[¶]Correspondence at: Department of Radiodiagnosis, Rural Medical College, PIMS, Loni, Maharashtra, India; Mobile: +9921160357, Email: sushilkachewar@hotmail.com



(a)



(b)

Figure 2: Plain radiograph (a) and coronal reformatted CT (b) show soft tissue swelling and proliferation of fat on palmar aspect of the ring and middle fingers (arrow), along with dorsal angulation and syndactyly (arrow).

Case 2

A three year old male child presented with excessive overgrowth of the second and third toes of right foot since one year. No limitation of motion or neurological deficits was demonstrated. On physical examination, non-tender, soft tissue mass was palpable on the plantar aspect of the enlarged toes with dorsal angulation of second toe. There were no overlying cutaneous changes, pitting edema or bruit. Plain radiograph showed soft tissue swelling of the second and third toe of right foot with hypertrophy of the phalanges. MRI revealed proliferation of subcutaneous fat in the plantar aspect of the second and third toes of right foot, corresponding to the distribution of the planter nerve (Figure 3a and b). Underlying bones revealed normal marrow signal intensity.



(a)



(b)

Figure 3: T1 Coronal MRI (a) reveals proliferation of fatty tissue (arrow) on plantar aspect of the second and third toes of right foot with signal intensity similar to that of subcutaneous fat as seen by fat suppressed STIR coronal image (b).

DISCUSSION

Macro dystrophia lipomatosa is a congenital, nonhereditary, progressive, localized gigantism which results in overgrowth of all the mesenchymal elements of the digit including phalanges, tendons, nerves and vessels. The localized gigantism is almost invariably recognized at birth¹. Involvement is almost always unilateral with equal incidence in male and female, although there may be enlargement of one or more adjacent digits in the same extremity². Bilateral involvement is rare³. Secondary osteoarthritic changes often develop in adolescence or early adulthood. The most common sites are the second and third digits; the fifth finger

is a rare site¹. Patients most frequently seek surgical correction for cosmetic reasons. Mechanical problems are not encountered until adolescence². The affected skin is thickened, pale, and glossy¹. The most striking pathologic finding is the increase in adipose tissue, interspersed in a fine mesh of fibrous tissue. Neural enlargement and irregularity may be prominent involving the median nerve in the hand and the plantar nerve in the foot^{2,3}. In patients with macro dystrophia lipomatosa of either upper or lower extremity, the non affected lower or upper extremity can also show multiple entrapment neuropathies that may or may not be clinically significant⁴.

True macrodactyly is a rare congenital anomaly characterized by an increase in the size of all elements of the digits¹. Neurofibromatosis, proteus syndrome, and macro dystrophia lipomatosa are included in this category. True macrodactyly is divided into two types: static and progressive. In the static type, the growth rate of the enlarged digits is normal whereas in the progressive type, the growth of the affected digits is accelerated¹. Silverman examined macro dystrophia histologically and found an association between macrodactyly and abnormalities of the median and plantar nerves⁵.

Etiology of the macro dystrophia lipomatosa remains obscure and various proposed etiologies include lipomatous degeneration, disturbed fetal circulation, an error in segmentation, the trophic influence of a tumefied nerve and in utero disturbance of a growth-limiting factor⁶. Several investigators postulate that macro dystrophia lipomatosa is an expression of neurofibromatosis. Lack of neurocutaneous or other systemic abnormalities in patients with macro dystrophia lipomatosa supports the diagnosis¹.

Plain radiographs demonstrate abnormalities in both the soft tissues and osseous structures. The soft tissue overgrowth is most marked at the distal end of the digit and along its volar aspect. The palmar and plantar overgrowth produces dorsal deviation of the affected parts. This was appreciated in case I. Lucencies in the soft tissues may reflect the overgrowth of fat which is characteristic of this anomaly. The phalanges are long, broad and often splayed at their distal ends. There is a high incidence of associated local anomalies including syndactyly and polydactyly. In case I, syndactyly is present. Clinodactyly is almost invariably present, resulting from the side-to-side variations in the accelerated rate of growth³.

CT demonstrates proliferation of fat along the nerve territory. MRI also reveals abundance of fatty tissue and fibrous thickening of a nerve. In these cases, the increased fat had the density of normal subcutaneous fat and was not encapsulated thus distinguishing them from other disorders. MRI revealed thickened nerve in the region of soft tissue

overgrowth in case II, but sometimes fatty infiltration in the nerve sheath may cause detection difficult in the subcutaneous tissue².

Disorganized new bone formation and gross soft tissue swelling due to infiltration by excessive abnormal hypodense fat is also reported. The bony overgrowth results in fusion and changes of secondary degenerative arthritis are rampant. Bony abnormalities are more clearly seen on CT scan than on plain radiographs or MRI⁷. It is also proposed that the detection of fibroadipose tissue masses with a proportional enlargement of other mesenchymal tissues on MRI should be used as the diagnostic method of choice for macro dystrophia lipomatosa⁸.

The differential diagnosis of macrodactyly includes both acquired and congenital conditions. Acquired macrodactyly includes dactylitis secondary to infection, infarction, and Still's disease; osteoid osteoma and melorheostosis and can be distinguished from the congenital type by the appearance at birth. Congenital macrodactyly including lymphangioma, hemangioma, Klippel-Trenaunay-Weber syndrome and Ollier's disease can be differentiated based on the non-fatty nature of the mass. Neurofibromatosis may be difficult to differentiate but proliferation of the subcutaneous fat and bony overgrowth in certain nerve territories are characteristic only of macro dystrophia lipomatosa^{1,3}. It can be confused with other common causes like congenital lymphedema. Although diligent clinical examination, imaging and histopathology are crucial in clinching the diagnosis, advanced imaging modalities score better due to their inherently non-invasive nature⁹. Proteus syndrome presenting with hemihypertrophy consisting of lipomatous tumor is similar to macro dystrophia lipomatosa. Proteus syndrome can be distinguished by various other associated features such as skull anomalies, pigmented naevi, lung cysts and intra-abdominal lipomas¹⁰. Fibrolipomatous hamartoma of the nerve is a rare tumor-like condition which consists of an infiltration of connective tissue and fatty element of the nerve. More than 80% of the lesions arise in the median nerve of the hand. The relationship between fibrolipomatous hamartoma of the nerve and macro dystrophia lipomatosa may be confusing. Approximately 66% of cases of fibrolipomatous hamartoma of the nerve are associated with macrodactyly. Fibrolipomatous hamartomas of the nerve may occur without macrodactyly, but those with macrodactyly are the same as macro dystrophia lipomatosa¹¹. A combination of fibrolipomatous hamartoma and macro dystrophia lipomatosa of the median nerve is reported to exist. Skipped lesions at the median nerve of the middle forearm have also been noted¹². It is suggested that MRI should be performed in cases with nerve dysfunction without an obvious cause after a thorough clinical

examination¹³.

The management is mainly surgical but the outcome may not be very gratifying⁹, as repeated debulking and partial amputation along with the extent of possible psychological trauma necessitate that surgery be delayed, if possible until complete of growth as the disease becomes static then and limb salvage surgery can then achieve a satisfactory outcome¹⁴.

In conclusion, radiological investigations especially CT and MRI are essential to make a definitive noninvasive diagnosis of macro dystrophia lipomatosa and help to differentiate from other causes of macrodactyly.

REFERENCES

1. Barsky AJ. Macro dactyly. *J Bone Joint Surg Am.* 1967 Oct; 49(7):1255-66.
2. Blacksin M, Barnes FJ, Lyons MM. MR diagnosis of macro dystrophia lipomatosa. *AJR Am J Roentgenol.* 1992 Jan; 158(6):1295-7.
3. Goldman AB, Kaye JJ. Macro dystrophia lipomatosa: radiographic diagnosis. *AJR Am J Roentgenol.* 1977 Jan; 128(1):101-5.
4. Kozanoglu E, Koc F, Goncu K. Macro dystrophia lipomatosa with multiple entrapment neuropathies: a case report. *Int J Neurosci.* 2008 Apr; 118(4):545-53.
5. Silverman TA, Enzinger FM. Fibrolipomatous hamartoma of nerve: A clinicopathologic analysis of 26 cases. *Am J Surg Pathol.* 1985 Jan; 9(1):7-14.
6. Singla V, Virmani V, Tuli P, et al. Case Report: Macro dystrophia Lipomatosa - Illustration of two cases. *Indian J Radiol Imaging.* 2008 Nov; 18(4):298-301.
7. Mittal A, Gupta R, Pawar I, et al. Radiological profile of a neglected case of Macro dystrophia Lipomatosa of the foot. *The Internet Journal of Radiology.* 2010; 11(2): retrieved from http://www.ispub.com/journal/the_internet_journal_of_radiology/volume_11_number_2_8/article_printable/radiological-profile-of-a-neglected-case-of-macro dystrophia-lipomatosa-of-the-foot.html.
8. Soler R, Rodriguez E, Bargiela A, et al. MR findings of macro dystrophia lipomatosa. *Clin Imaging.* 1997 Mar-Apr; 21(2):135-7.
9. Khan R, Wahab S, Ahmad I, et al. Macro dystrophia lipomatosa: four case reports. *Ital J Pediatr.* 2010 Oct; 36:69.
10. Azouz DM, Costa T, Fitch N. Radiologic findings in the Proteus syndrome. *Pediatr Radiol.* 1987; 17(6):481-5.
11. Broadwater BK, Major NM, Goldner RD, et al. Macro dystrophia Lipomatosa with associated fibrolipomatous hamartoma of the median nerve. *Pediatr Surg Int.* 2000; 16(3):216-8.
12. Chiang CL, Tsai MY, Chen CK. MRI diagnosis of fibrolipomatous hamartoma of the median nerve and associated macro dystrophia lipomatosa. *J Chin Med Assoc.* 2010 Sep; 73(9):499-502.
13. Nilsson J, Sandberg K, Dahlin L, et al. Fibrolipomatous hamartoma in the median nerve in the arm - an unusual location but with MR imaging characteristics: a case report. *J Brachial Plex Peripher Nerve Inj.* 2010 Jan; 5:1.
14. Gao B, Zheng LP, Cai ZD. Limb salvage surgery in a patient with macro dystrophia lipomatosa involving an entire upper extremity. *Chin Med J (Engl).* 2010 Oct; 123(19):2744-7.