Macrodystrophia Lipomatosa: A rare form of localized Gigantism

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Abstract

Macrodystrophia Lipomatosa (MDL) is a rare cause of localized gigantism of obscured aetiology generally affecting hands and feet. The disease is congenital but not hereditary and present in infancy or during early childhood. It is characterized by progressive overgrowth of all the mesenchymal elements in a somatosome usually of a limb, with predominance of fibro – fatty elements.

Key Words: Macrodystrophia Lipomatosa; localized gigantism; fibro-fatty; mesenchymal; macrodactily

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Introduction

We present 3 patients with MDL, who underwent Magnetic Resonant Imaging (MRI) at the Department of Radiology, National Hospital of Sri Lanka from October 2013 to March 2014. Among them, 1st patient has unusual extensive involvement of whole left lower limb from buttock to toes, while others have more typical involvement.

Case 1

Twenty one year old female presented with grossly enlarged right lower limb for evaluation prior to de-bulking surgery (Fig.1). Patient was asymptomatic except for a mild limp but had cosmetic concerns. No trophic changes of skin, pitting oedema or vascular bruit was demonstrated in the affected limb on examination. She had undergone two surgeries earlier to excise enlarged right 2nd to 5th toes during early childhood and to debulk a large lump over right buttock during later childhood. Histology revealed normal looking fatty tissues in a fibrous mesh.

Plain radiographs of the affected limb showed soft tissue thickening over the R/hemi pelvis, thigh, leg and foot with no hypertrophy of underlying bones. But shaft of metatarsal bones show thinning of cortex with expansion of distal ends (Figs. 1.2).

On MRI, diffuse deposition of soft tissue similar to subcutaneous fat, hyperintense to muscles on TIW and intermediate intense on T2W, was seen over the posterior aspect of the gluteal region and the postero-lateral aspect of entire lower limb (Fig.1.3). Above tissue showed signal loss in Short Tau

inversion recovery (STIR) sequence (Fig.1.4) confirming its fatty nature.

Extensive involvement as in this case is extremely rare in literature (Fig.1.5).

Case 2

Thirty year old male presented with enlarged right index and middle fingers since early childhood. He complained of progressive disproportionate growth of involved digits with no weakness or numbness. On examination, index finger was involved to a greater extent than the middle finger. There were no skin changes, oedema or vascular bruits over the abnormalities. Neurology of the distal limb was intact.

He underwent MRI of right forearm and hand which revealed excessive non-encapsulated soft tissue of high TIW signal along the radial aspect of index finger and middle finger. Suppression of signal in STIR confirmed fatty nature of the tissue. Underlying cortical bone and bone marrow showed no abnormality.

Case 3

Eight year old female presented with progressive enlargement of soft tissue over the thenar eminence of right hand despite early childhood amputation of 1st to 3rd fingers for localized gigantism. Histology was reported as proliferation of normal appearing adipose tissue. Unremarkable findings were elicited on examination of the rest of the right upper limb.

MRI Scan of right forearm and hand demonstrated fibro-fatty tissue proliferation in the thenar eminence filling 1st and 2nd web spaces.

Figure 1.1: Shows enlarged right lower limb. Compare with the normal left lower limb.

Fig. 1.2: Plain radiographs of the pelvis, thigh, distal leg and foot showing soft tissue hypertrophy of R/lower limb with splaying of distal ends of metatarsals

Fig. 1.3: These labeled images show appearance of hypertrophied tissues on TIW MRI. Note identical signal intensity to subcutaneous fat and lack of definite margin between normal and pathological tissues. Labeling is done to emphasize fat tissue infiltration to muscles and nerves.

Fig.1.4: Hypertrophied soft tissue
Losses signal in STIR indicative
of fat.

Fig.1.5: Extensive involvement
of whole of R/lower limb.

Fig.2.1: Coronal and
axial T1W MRI of
right hand and
fingers showing
hypertrophy of
fibro-fatty tissue of
2nd and 3rd fingers.

Figure 2.2: Axial STIR images of right
hand showing low signal adipose
tissue of index and middle fingers

Figure 3.1: Axial T1W and STIR MRI images of R/hand showing characteristic fibro fatty proliferation.

Figure 3.2: Axial T1W image of palm with hypertrophied fat.

Discussion

MDL is a rare non-hereditary disorder characterized by non-malignant proliferation of all mesenchymal elements with abundance of adipose tissue. This results in localized gigantism of the affected part of the body as described in above three cases. The aetiology is yet unknown but numerous hypotheses exist^{1,2,3}. The occurrence is sporadic with a slight predilection for males.⁴ Although the usual presentation is at birth, infancy or early childhood, late presentations are reported in adulthood. The disease is usually unilateral and localized but bilateral involvement has been reported.⁴

MDL usually involves a somatome supplied by a nerve, usually the territory of median nerve of upper limb and medial plantar nerve of lower limb. Lower limb is affected more often than the upper limb, with 2nd and 3rd digits being the most commonly affected as in our 2nd case⁵. Involvement of a whole limb as in the first case has rarely been reported⁶. MDL of abdominal wall is also reported^{7,8}.

Excess of non-encapsulated adipose tissue dispersed in a fibrous matrix's seen in subcutaneous tissue, bone marrow, muscle and nerve sheaths at histology⁹. Therefore histology of biopsied/ resected specimens is

described as fibro fatty tissue proliferation or diffuse lipoma. Neuronal enlargement especially of median and lateral planter nerves may also be seen.

Although most patients present for cosmetic reasons symptoms due to entrapment neuropathy, degenerative arthropathy could be troublesome from childhood or early adulthood¹⁰. Degenerative joint disease could present early in the life, mainly due to heaviness of limb/part and mechanical misalignment.

Plain radiographs of MDL show soft tissue overgrowth of affected limb or digit often with underlying bone changes such as increased length, width and cortical thickening. Thinning of shaft with splaying of distal ends of phalanges and metatarsals/metacarpals giving rise to “mushroom” shape has been described^{11,12} as seen in our 1st case. Other abnormalities, especially clinodactyly, may be associated findings in involved extremities¹³.

CT appearance in MDL has sparsely been reported but undoubtedly be helpful due to pathognomonic negative attenuation values of fat. On ultrasound, hypertrophied soft tissue is seen with identical echogenicity to subcutaneous fat. MRI clearly demonstrates non-capsulated fibro-fatty tissue with similar signal characteristics to subcutaneous fat on all pulse sequences. Pathological infiltration into muscles and nerves is best depicted with MRI.

The rationale of therapeutic intervention is to improve cosmesis and preserve function of the affected body part¹⁴. Surgical resection of hypertrophied parts is the standard treatment. Because of complexity of fatty tissue infiltration, there is a high recurrence rate in the range of 33%-60%¹⁵ necessitating repeated surgeries, usually by plastic surgeons to obtain satisfactory results¹⁶. Less invasive procedures such as

liposuction are sometimes used with variable success rates.

Several diseases present as macrodactily or localized gigantism¹⁷ and needs differentiation from MDL. Differential diagnoses include neurofibromatosis 1, lymphangiomatosis, haemangiomas, fibrolipomatosis of nerves, Klippel-Trenaunay-Weber syndrome, Maffucci's syndrome and Ollier's disease.

MDL could be differentiated from above by clinical and imaging features most of which usually have abnormalities elsewhere in the body other than a hypertrophied limb.

Neurofibromatosis 1 usually present with clinical stigmata and positive family history. MRI shows focal or plexiform lesions along a nerve with high signal intensity on T2 sequence and strong contrast enhancement.

Lymphangiomas are multi cystic avascular lesions with high signal in T2W.

Haemangiomas and associated Klippel-Trenaunay-Weber syndrome show characteristic clinical appearance, vascular calcifications on plain radiographs, multi cystic vascular lesions on USS and CT with flow voids on MRI. Neuronal bundles are hyper intense to muscles in fibrolipomatosis of the nerve with clear demarcation between the pathology and normal soft tissue. Beckwith-Wiedemann syndrome manifests with organomegaly and visceral tumours.

Conclusion

MDL is a rare congenital, non-hereditary cause of localized gigantism with typical imaging features. MRI in particular, shows characteristic findings which can be of much use in arriving at the diagnosis without histology. Therefore, the radiologist plays an important role in management of MDL.

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