Case report

Macrodystrophia lipomatosis : a rare congenital anomaly.

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Abstract

Macrodystrophia Lipomatosis is rare congenital hamartomatous enlargement of soft tissue components leading to gigantism of single, multiple digits or entire limb. Clinical findings are recognised as early as in neonatal period to late adulthood. Patients usually present for either cosmetic corrections or mechanical problems. We report a case of 8 year old female with progressive increase in size of left 2nd and 3rd toes. X-ray findings show congenital focal enlargement of soft tissues and bones of 2nd and 3rd toes and adjacent metatarsals. Clinical and radiological features of our patient points out towards this uncommon condition.

Keywords :- congenital, hamartomatous, gigantism, metatarsals.

Introduction

Macrodystrophia lipomatosis [ML] is distinct clinical entity often misdiagnosed as other forms of macrodactyly. It is a rare congenital form of macrodactyly characterised by increase in mesenchymal elements especially overburdance of fibrofatty tissue on plantar aspect of foot.

Historical Insight

ML term was first used by Ferz in 1925. In 1967 Barsky gave detailed description of local gigantism and he described 2 forms of it that is 'static' and 'progressive'. It was later confirmed that Ferz was dealing with progressive form of the disease.

ML is congenital but not hereditary. Clinical findings can be recognised as early as neonatal period to late adulthood.

Case report

A 8 year old female presented with painless increase in size of left 2nd and 3rd toe since birth. There was no history or complains of any functional loss, sensory loss, any ulcers or cyanosis of enlarged digits or the whole limb. There was no history of any delayed milestones or developmental delay in the patient. There was no family history of gigantism localised or general in the patient.

Patient was school going girl in 4th standard with normal sleep pattern and normal bladder bowel functions.

On Examination; patient was conscious oriented with no signs of pallor, icterus, clubbing, peripheral cyanosis or lymphadenopathy. We could not find any king of skin nodules, hemangiomas or cafe-au-lait spots. Local examination of the affected foot revealed there was both increase in length and width of 2nd and 3rd toes with increase in subcutaneous tissue mass which was non tender to touch (as shown in figure 1 and 2). There was no evidence of edema, any venous ulcers, any varicose veins in the patient. All peripheral pulses were intact.

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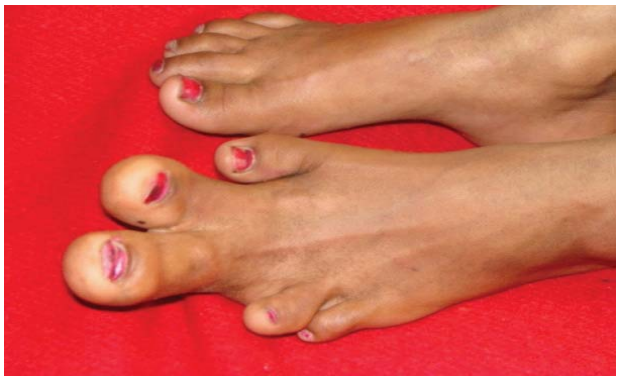

Figure 1: showing lateral view of hypertrophied 2nd and 3rd toes of left foot

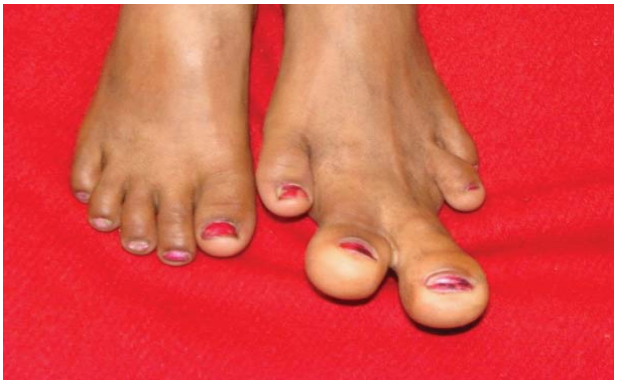

Figure 1: showing front view of hypertrophied 2nd and 3rd toes of left foot

Temperature and colour of toes are comparable to rest of limbs. There was no restriction of movement at involved joints. On investigation; no remarkable abnormality was seen in complete blood counts, urine routine and microscopy and biochemistry profile.

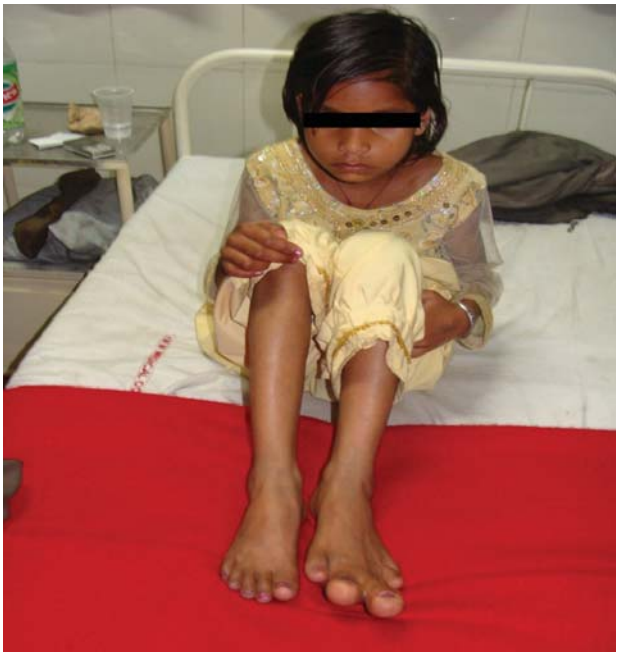

Figure 3: showing the patient with affected left foot

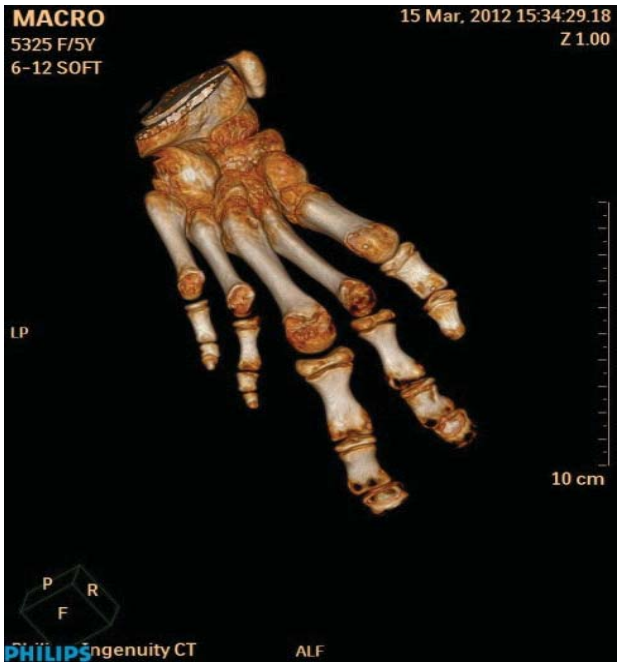

Figure 4 : CT scan left foot

On radiological examination; the size of left 2nd and 3rd metatarsal was longer and thicker than others more so of 3rd metatarsal. Phalanges of 2nd and 3rd toes also show increase in length and width as compared to other toes (as shown in figure 5). CT also depicted congenital focal enlargement of soft

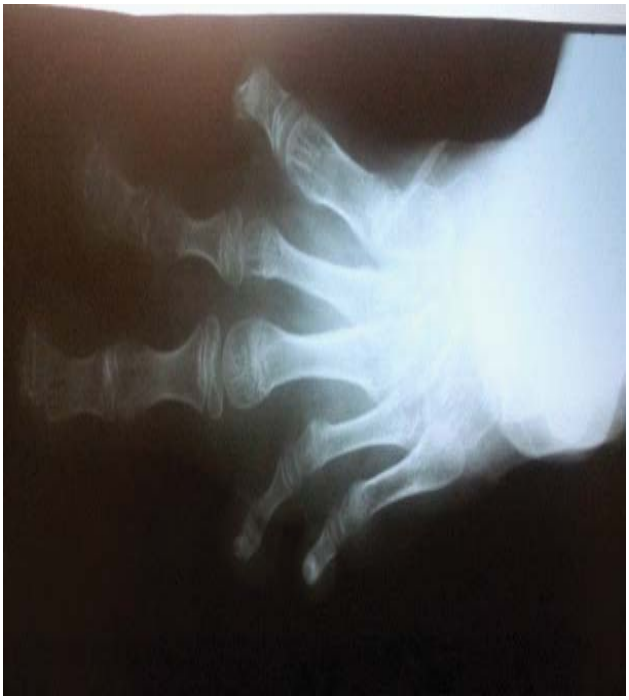

Figure 5 :shows X-ray of the left foot depicting 2nd and 3rd metatarsals longer and thicker than other digits

tissues and bonesof 2nd and 3rd toes depicting Congenital Partial Gigantism (as shown in figure 4). On basis of history, clinical examination and radiological examination the diagnosis of Macrodystrophia Lipomatosis was made.

Discussion

Macrodystrophia Lipomatosis is a rare congenital anomaly characterised by abnormal overgrowth of mesenchymal elements resulting in gigantism of single or multiple digits or entire limb. In our case there is unilateral involvement of 2nd and 3rd toe of left foot. Our patient presented at age of 8 years however the anomaly was congenital.

There is generally involvement of lateral aspect of upper limb (median nerve involvement) and/or median aspect of lower limb (plantar nerve involvement). There is predominantly distal limb involvement with lower limb predominantly involved. 2nd and 3rd digits are involved more.[1,2] Various etiologies have been postulated but still definite cause is not known [3,4,5]

1. Lipomatous denegeration
2. Fetal circulation abnormality
3. Damage to extremity bud
4. Errors in segementation in intrauterine life
5. Hypertrophy of concerned nerve

Histological features of ML are characterised by markedly increase in all mesenchymal elements dominated by adipose tissue in fine fibrous network involving periosteum, bone marrow, nerve shealth, muscle and subcutaneous tissue.[(6)]

Conclusion

ML is progressive hamartomatous enlargement of the fibrofatty tissue involving all the layers of soft tissue and even bone more commonly leading to localized gigantism Diagnosis is accomplished on the basis of clinical and radiological evaluation which can be confirmed on histopathological examination The management is mainly surgical but the outcome may not be very gratifying.

ML may be confused with many other disorders and they are as follows:-

Table 1: Listing various differential diagnosis of Macrodystrophia Lipomatosis.

DISEASE	DIFFERENTIATING FEATURE
1. Neurofibromatosis-1	Caffe-au-lait spots Soft skin nodules Positive family history
2. Klippel trenaunay weber synd.	Cutaneous hemangiomas Varicose veins
3. Proteus syndrome	Skull abnormality Pigmented naevi Lung cysts
4. Lymphangiomatosis And hemangiomatosis	These do not show osseous hypertrophy

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