Macrodystrophia lipomatosa: A case report
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ABSTRACT

Macrodystrophia lipomatosa (MDL) is a rare case of congenital macrodactyly with progressive proliferation of all mesenchymal elements of a digit or extremity with disproportionate increase in fibro-adipose tissue. Cases are reported but not very massively. We report few cases of this anomaly in upper as well as lower extremity, highlighting the clinical features, differential diagnoses, and the treatment protocol that we followed for these patients.

Key words: macrodystrophia lipomatosa, macrodactyly, hypertrophy, gigantism, fibro-fatty tissues

INTRODUCTION

MDL is a rare non hereditary congenital form of localized gigantism which is characterized by progressive overgrowth of mesenchymal elements with a disproportionate increase in the amount of fibroadipose tissue. It is very frequently noticed in neonates however, problem ensues and progress as the child grows. It most frequently occurs in the region of median nerve distribution in the upper extremities and the plantar nerve distribution in lower extremities. Usually, one or more digits of the unilateral limb are affected.

CASE REPORT

There are three cases of macrodactyly: (1) involving left foot; (2) involving right foot and (3) macrodystrophy of left hand with no evidence of any deformity elsewhere.

In case 1, the patient had a lesion at the unilateral left lower extremity along the plantar nerve that had an increase in size disproportionate to the rest of his body. In case 2, the clinical picture was of macrodactyly of right foot with soft tissue swelling.

Case 1: Clinical picture was of macrodactyly involving left foot

Fig.1. Clinical photograph of left foot demonstrates the macrodactyly involving 2nd, 3rd & 4th toe

Fig.2. Plain radiograph of left foot shows soft tissue swelling on the plantar aspect of the affected toes, the distal end is predominantly affected, with splaying of the phalanges and macrodactyly

Fig.3. USG picture of left foot shows echogenic, soft tissue swelling on the plantar aspect of affected toe. On Doppler, no obvious abnormal vascularity was seen
**Case 2:** X-ray showing macrodactyly of right foot with soft tissue swelling

**Fig. 4 & 5**

**Case 3:** Patient had macrodystrophy of left hand there was no evidence of any deformity elsewhere

**Fig. 6.** Plain radiograph of left hand shows soft tissue swelling on the palmar aspect of the index finger, the distal end is predominantly affected, with splaying of the phalanges and macrodactyly

**DISCUSSION**

MDL is a rare, unilateral, non-hereditary condition involving one or more of the digits of the extremities in the distribution of median and plantar nerve in upper and lower extremities respectively. It was first of all described in 1925 by Feriz regarding a case of unilateral growth of both fibro adipose tissue and bone of the lower limb. Overgrowth of soft tissue is more at volar and distal aspect. Reported lesions hints that it usually present since birth and usually is associated with high incidence of syndactyly, polydactyly, brachydactyly or clinodactyly.¹ The disease has almost equal distribution in both sexes. Exact aetiology of MDL is not known however during embryonic development somatic defect leads to maldevelopment in limb bud and fetal circulation.¹ The lesion mostly involves middle and index fingers corresponding to the territory supplied by the branches of median nerve.²

The different imaging modalities shows specific picture of the disease. X-rays reveals overgrowth of soft tissue, lucent area of fat deposition, bony overgrowth, secondary osteoarthritis in the form of marginal cyst formation, osteophyte formation and joint space reduction.³ The Ultrasonography and CT scan demonstrates fat deposition in soft tissues in area of distribution of a particular nerve.⁴ The MRI demonstrates fat deposition as hyperintense signal on T1W and iso on T2W images, suppressed signal on fat saturation sequence. In MDL fat deposition is not encapsulated and fibrous strands are demonstrated within fat as hypointense signal on T1W images.

The differential diagnoses of the lesions are 1. Neurofibromatosis (hereditary, b/l limb involvement, hyperintense signal on T2. Lesions are close to nerve distribution.⁵ 2. Haemangiomatosis (palpable bruit clinically, vascular channels demonstrated on MRI.) 3. Lymphangiomatosis (hypointense on T1W and hyperintense on T2W Osseous growth not seen so striking in haemangiomatosis and lymphangiomatosis.⁶ 4. Klippel-trenau-weber syndrome (bone and soft tissue hypertrophy, hemangioma, hypoplasia or atresia of major
deep vein of limb with dilated superficial veins) and 5. Beckwith widemann syndrome (macroglossia, omphalocoele, umbilical hernia, organomegaly, hemihypertrophy, increased incidence of neoplasm).

The treatment of localized gigantism poses a challenging surgical dilemma, and it is treated with debulking and s os amputation. A series of 3 surgeries, designed to reduce the length, width, height, and overall bulk of the congenitally enlarged limb. The 3 procedures debulked the foot for normal ambulation and same-size shoe wear for both feet. The resulting functional and aesthetic improvements achieved through reconstructive treatment provided as desirable alternative to amputation.

CONCLUSION

With advent of newer advanced imaging modalities it has become very convenient to pin point the etiology of the lesion leading to early diagnosis and timely intervention.

AUTHOR NOTE

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