Case Report

Macrodystrophia Lipomatosis: A Case Report

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Received: 10-02-2013
Revised: 20-04-2013
Accepted: 30-04-2013

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ABSTRACT

Macrodystrophia Lipomatosa is a rare cause of congenital macrodactyly characterized by progressive proliferation of all mesenchymal elements, with disproportionate increase in fibroadipose tissue. This anomaly is reportedly more common in the foot than in the hand with predilection for the plantar and median nerve distribution. This rare cause of gigantism of limb can be confused with other common causes like congenital lymphedema. We report a case of 2 years old female child presenting in outdoor as a painless progressive enlargement of index finger, middle finger and thumb of right hand since 6 months of age causing mechanical difficulty.

Key words: *local gigantism, macrodactly*

Introduction

Macrodystrophia lipomatosis is a rare form of localised gigantism characterised by painless enlargement of 2nd or 3rd digit of hand or foot. It is usually unilateral but may involve adjacent digits in the distribution of median nerve in the upper extremity and in the distribution of plantar nerve in the lower extremity. It is usually recognised at birth or in the neonatal period. As the patient grows the deformity begins to interfere with joint function. Plain x-ray suffices and demonstrates the splayed, lengthened and broadened phalanges with endosteal and periosteal bone deposits but MRI is helpful in distinguishing macrodystrophia lipomatosis from other causes of moacrodactly. [1] It demonstrates the accumulation of fat in the subcutaneous tissues that is not capsulated.

Case Report

A 2 year old female girl born of a non consanguineous marriage to a primi mother by full term normal delivery at hospital with normal antenatal course and normal routine antenatal ultrasounds. She was brought by her parents for cosmetic reasons in the outdoor for correction of painless progressive enlargement of index finger, middle finger and thumb of right hand. (Fig. 1)

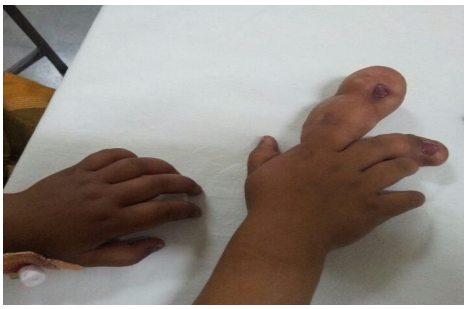

Fig.1 Shows enlargement of index finger, middle finger and thumb of right hand

There was no history of any trauma. No abnormality was noticed at birth by her parents or health workers. Parents noticed a bulbous swelling progressive since 6 months of age. On examination there was no other congenital anomaly or neurocutaneous marker, no visible pulsations, no redness or swelling of any other part of hand. Patient did not have any pain in the fingers or hand, but there was difficulty in mechanical functions like holding a spoon or writing. All the developmental milestones were normal of her age group. X-ray of both hands were done, and right hand x-ray revealed lengthened, broadened phalanges of index and middle finger corresponding to the localised gigantism /macrodactly while the left hand x-ray was within normal limits.

Fig.2 X- ray showing lengthened, broadened phalanges of index and middle finger

MRI of right hand and upper limb was done to rule out different causes of macrodactly and to show its relation to median nerve. MRI revealed proliferation of subcutaneous fat that was not capsulated with bony enlargement of index and middle finger thus confirming the diagnosis of macrodystrophia lipomatosis. (Fig. 2, 3)

There was involvement of digital nerve of one finger. Child was taken up for surgery and debulking of fingers was done. Intra operative period and post operative stay was uneventful. Histopathology of specimen was suggestive of marked proliferation of subcutaneous fat that was not capsulated with diffuse bony enlargement as well as widening of phalanges of index and middle finger.

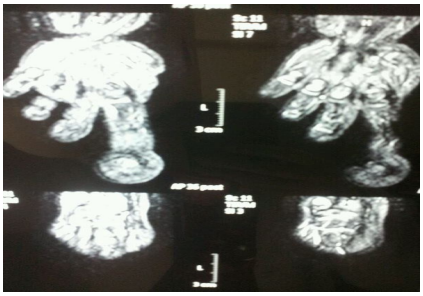

Fig.3 MRI showing proliferation of subcutaneous fat with bony enlargement of index and middle finger

Discussion

Macrodystrophia lipomatosis is a rare, non-hereditary development anomaly appears to be more common in the hand than in the foot resulting in the overgrowth of all the mesenchymal elements of digits including phalanges, tendons, nerves, and vessels, but there is disproportionate increase in the fibroadipose tissue. [2] This congenital anomaly occurs most frequently in the distribution of the median nerve in the upper-extremity. [3] Our patient did not have any family history of similar deformity and was consistent with the literature. In 1967, Barsky defined true macrodactly as a rare congenital malformation characterized by an increase in size of all elements or structures of digit or digits. [4] According to literature only 56 cases has been documented. Feriz in 1925 1st coined the term macrodystrophia lipomatosis due to the interaction of two or more intrinsic and extrinsic agents either genetic or environmental referring to localized

gigantism in the lower extremity only. [5] In 1968, Ranawat et al accepted the term as also applicable to gigantism in the upper extremity. [6] It is usually recognized at birth or in the neonatal period. As the patient grows, the deformity begins to mechanically interfere with joint function, vascular supply and innervation. The etiopathogenesis of this condition is still not clear. Different imaging modalities, such as plain radiography, USG, CT scan, and MRI, have a role in the evaluation of MDL. Plain radiography reveals hypertrophy of soft tissue and bone, with translucencies in the soft tissue due to increased adipose tissue. The phalanges, especially the distal phalanx, are elongated, broad, and splayed, sometimes giving rise to a mushroom-like appearance. Secondary osteoarthritic changes like joint space narrowing, subchondral cysts and osteophytes often develop in adolescence or early adulthood. [7] Both USG and CT scan can be used to demonstrate the proliferation of fat along the nerve territory. [8 ,9] MRI easily demonstrates the excess fibrofatty tissue, which has signal characteristics similar to subcutaneous fat, i.e., high signal on T1W, intermediate signal on T2W, and low signal on fat-suppressed sequences. The fat in MDL is not encapsulated. The fibrous strands within the fatty tissue are seen as low-signal-intensity linear strands on T1W images.

The differential diagnoses of MDL and macrodactyly include neurofibromatosis, hemangiomatosis, lymphangiomatosis, Proteus syndrome, and fibrolipomatous hamartomas. Neurofibromas show marked hyperintensity on T2W images and are seen in close relation to nerves. A positive family history, presence of cutaneous lesions, and bilaterality favor neurofibromatosis, while hypertrophy along a nerve territory, unilaterality, and demonstration of fat within the nerve sheath on MRI favor MDL. [10] Lymphangiomas are hyperintense to muscle on T1W and hyperintense to fat on T2W images. Clinically, diffuse swelling and pitting edema are found. [8] In hemangiomatosis, a bruit may be palpable clinically and, on MRI, long TR/TE sequences show a septate configuration of high-signal-intensity channels, corresponding to the vascular channels and fibrous strands found in hemangiomas. Osseous growth is not seen in both lymphangiomatosis and hemangiomatosis. [8] Proteus syndrome presenting with hemihypertrophy may simulate MDL, but associated abnormalities like calvarial changes, pulmonary cysts, pigmented nevi, and intra-abdominal lipomas help to arrive at the correct diagnosis. Some consider MDL to be a localized form of Proteus syndrome. Fibrolipomatous hamartoma (FLH) of nerve is a rare tumor-like condition in which mature fat infiltrates the neural sheath, with the majority of the lesions occurring in the median nerve. Pathologically, in FLH, the deposition of fat is within the nerve sheath, while in MDL it occurs throughout the involved part of the digits/extremity. However, MDL may be an associated feature of FLH in as much as 30–66% of cases. [11] FLH may show a speckled appearance on MR, correlating with its histologically known architecture, i.e., neural fascicles separated by fat and connective tissue.

In conclusion, determination of the cause of macrodactyly is clinically difficult due to the many possible etiologies. However, appropriate imaging, particularly with MRI, can make the determination of the underlying process easier and can be of great help in arriving at a correct diagnosis. Imaging helps in

differentiating MDL from other causes of localized hemihypertrophy, which have different prognoses, complications, and treatment.

The rate of accelerated growth varies among patients and even among affected digits. Involvement is almost unilateral although there may be enlargement of one or more adjacent digits in the same extremity usually the anomaly occurs in the lower extremity but few cases of upper extremity has been noted like in our case 2 fingers index, and middle finger and a part of thumb was involved in the right hand. Patients frequently seek surgical correction for cosmetic purposes. The affected part is increased both in length and width and skin is thickened, pale and glossy. The most striking finding is the increase in adipose tissue interspersed in a fine mesh of fibrous tissue which involves the bone marrow, periosteum, muscles, nerve sheaths and subcutaneous tissues. The most difficult and closest differential would be neurofibromatosis. Macrodystrophia lipomatosis is unilateral but neurofibromatosis has bilateral involvement with hemangiomatous/ lymphangiomatous elements combined with mesodermal dysplasia Neurofibromatosis was ruled out in this case on the basis of clinical examination, radiology and by histopathology.

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Cite this article as: Malhotra P. Macrodystrophia Lipomatosis: A Case Report. Int J Med and Dent Sci 2013; 2(2): 220-223.
Source of Support: Nil
Conflict of Interest: No