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Case Report

Macrodystrophia lipomatosa of finger—A rare case report ☆

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ABSTRACT

Macrodystrophia lipomatosa is a rare form of nonhereditary congenital localized gigantism involving upper and lower limbs and is characterized by overgrowth of all the mesenchymal elements predominantly fibro-adipose component, in the distribution of a particular nerve, usually median nerve. It usually presents with progressive painless overgrowth of the involved limb, toe, or digit and is associated with macrodactyly. It might cause limitation of the movement of the involved part. Imaging has an important role in diagnosing this condition and differentiating it from malignant mimics. Imaging findings include hypertrophy of the mesenchymal elements of the involved digits and/or limbs predominantly fibro adipose component with associated overgrowth of the phalanges. In this case report, we present a case of unilateral involvement of index finger and thumb with associated macrodactyly.

Introduction

Macrodystrophia lipomatosa (MDL) is a rare form of localized gigantism. It is a benign congenital nonhereditary condition. It is usually identified in the neonatal period [1]. It is characterized by a marked increase in all mesenchymal elements, which is dominated by fibro-adipose tissue in a fine fibrous network involving periosteum, bone marrow, nerve sheath, muscle, and subcutaneous tissue [2]. The characteristic presentation is a painless enlargement of the second or third digit of the hand or foot. It is usually unilateral. It usually involves digits in the distribution of median nerve. Occasionally it can also involve an entire limb [1]. Imaging shows marked hypertrophy of the phalanges and the overlying soft tissue predominantly the adipose tissue of the involved digits. It can be associated with syndactyly, polydactyly, or clinodactyly. The condition is treated with surgical intervention to improve the cosmetic appearance while preserving neurologic function.

Case report

A 35 year old female presented to orthopedics outpatient department (OPD) with complaints of swelling in the index fin-

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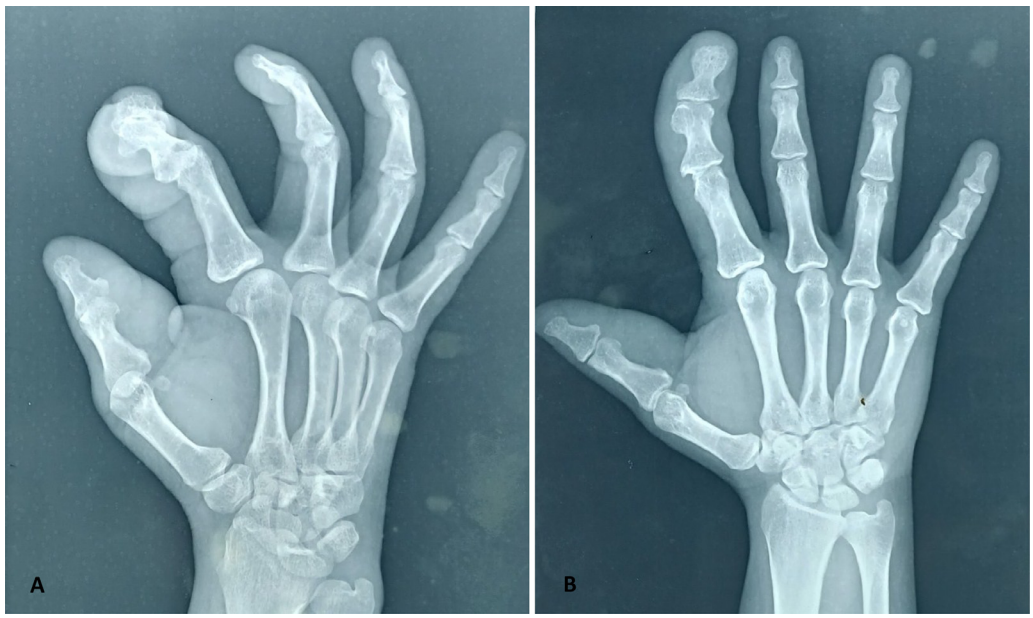

Fig. 1 – X-ray right hand oblique (A) and AP (B) views showing hypertrophy with widening of all the phalanges with few bony outgrowths involving first and second digits. Overgrowth of the overlying soft tissue is seen. There is joint space narrowing and sclerosis involving proximal interphalangeal joint of second digit due to secondary degenerative changes.

ger and thumb of her right hand, which caused mild discomfort to her with a history of on and off pain. She first noticed this swelling in her childhood but she took no treatment as it caused no significant disability in her daily routine work. The swelling gradually increased since childhood. There was no history of trauma. It was painless and caused no restriction of mobility of fingers or any neurologic symptom. The patient presented to OPD due to cosmetic reasons and mild discomfort while finger movements.

On physical examination, there was diffuse nontender swelling of the index finger and thumb involving proximal, mid and distal phalynx. There was no associated erythema, increased surface temperature or any scar over the overlying skin. There was no pulsatility or audible bruit over the swelling. Range of motion of all the fingers was normal. There was no associated sensory loss. There was no lymphadenopathy. There were no rashes over any part of the body. All routine blood investigations were within normal range.

Plain x-ray showed diffuse hypertrophy of the first and second digit of right hand causing widening of all the phalanges with few bony outgrowths. Overgrowth of the overlying soft tissue is seen (Fig. 1). There is joint space narrowing and sclerosis involving proximal interphalangeal joint of second digit due to secondary degenerative changes [8,9].

Magnetic resonance imaging (MRI) showed diffuse hypertrophy of fatty tissue in index finger and thumb predominantly on the volar aspect. There was broadening of the phalanges with tiny bony outgrowths from middle and distal phalynx with mild cortical irregularity. There was also associated deformed alignment of the distal and middle phalynx of index finger with medially angulated distal phalynx (Fig. 2).

The patient was advised for debulking surgery coupled with osteotomy, however, she refused the surgery considering the complications of nerve injury that might limit the movements of the fingers. Therefore, the patient is being managed conservatively and kept on follow-up on OPD basis.

Discussion

MDL is a rare congenital nonhereditary condition characterized by localized gigantism involving the upper and lower limbs. It commonly involves the index and middle fingers of the hand in the distribution of the median nerve in the majority of cases reported [3,4]. Distribution in the area of the ulnar nerve can also be involved but is sporadic and less commonly seen [5,6]. The condition is usually unilateral and bilateral involvement is even rarer. No sex predilection is seen [6,7]. In literature, this condition has been described by other names such as Lipofibroma, Fibrolipoma, and Intraneural lipoma [8,9].

In this case report, we present a case of MDL with unilateral involvement of the index and the thumb of right hand. This entity was first described by Feriz in 1925 [3]. Tahiri et al. [10] comprehensively studied 180 cases reported in the literature between 1946 and 2012 and found that 32% of the cases were associated with macrodactyly and 8% of the cases were associated with trauma. Our case was associated with macrodactyly.

Clinically it typically presents as progressive painless enlargement of the digits, wrist or forearm [1]. Neurologic symptoms depend on the involvement of the nerve like median nerve or ulnar nerve. It can also cause carpal tunnel syndrome which requires surgical intervention [10].

In literature, it had been described in 2 forms that is, static which is the proportionate growth of the involved side, and

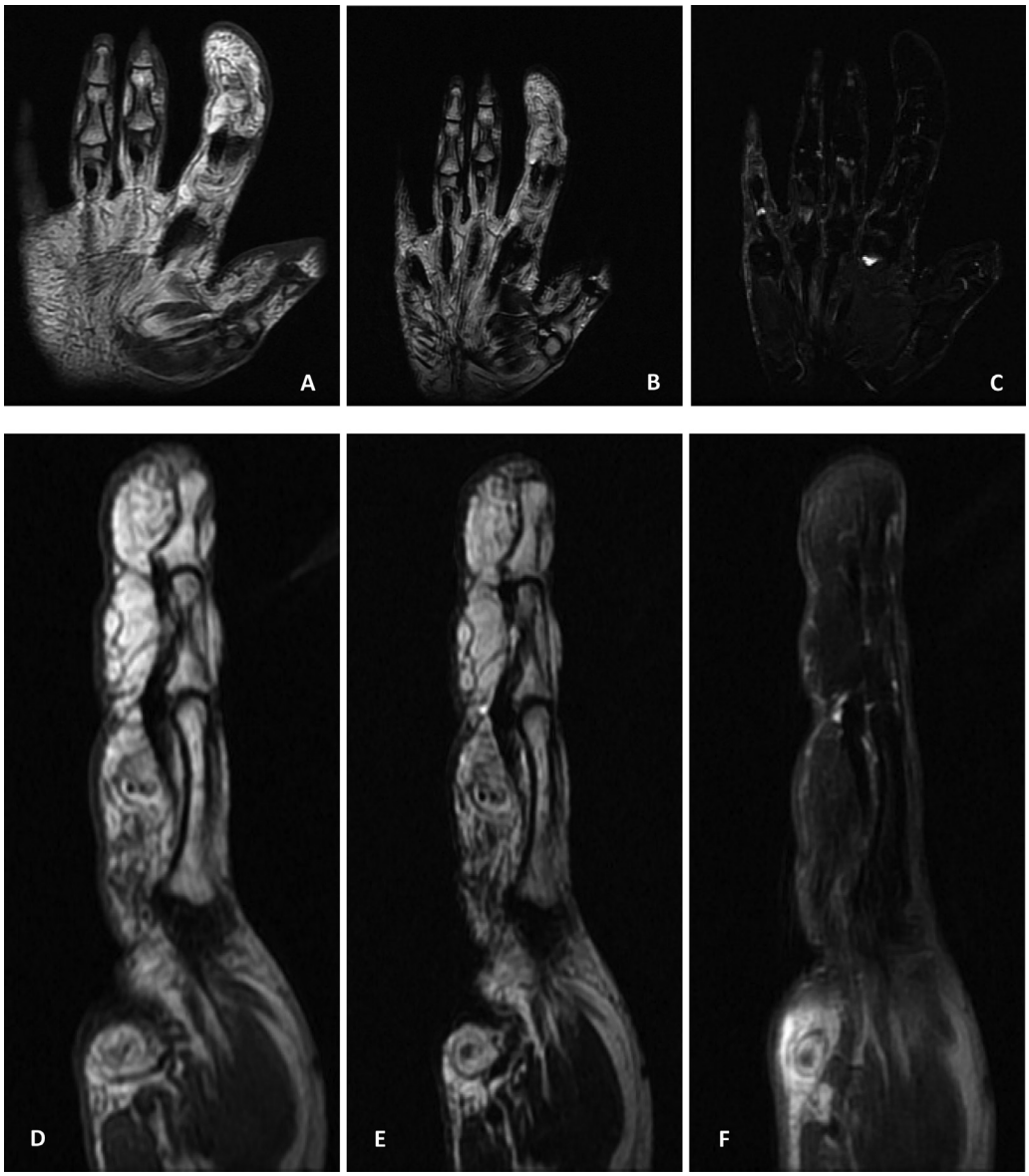

Fig. 2 – Coronal T1 (A), coronal T2 (B), coronal STIR (C), sagittal T1 (A), sagittal T2 (B) and sagittal PDFS (F) sequences show diffuse hypertrophy of fatty tissue in index finger and thumb predominantly on the volar aspect. There is broadening of the phalanges with tiny bony outgrowths from middle and distal phalynx with cortical irregularity.

progressive which is the disproportionate continuous growth of the involved site, the latter being more common [11,12].

Hypotheses about the pathogenesis of MDL is controversial but is characterized by a marked hypertrophy of all the mesenchymal elements that predominantly consist of adipose tissue in a fine fibrous network involving periosteum, bone marrow, nerve sheath, muscle, and subcutaneous tissue [2].

Imaging has an important role in diagnosing MDL including x-ray, ultrasonography, computed tomography and MRI. X-ray findings include overgrowth of the phalanges of the involved digits with bony outgrowths and associated overgrowth of the overlying soft tissue due to the proliferation of the mesenchymal elements. MRI demonstrates accumulation of fat in the subcutaneous tissues without any appreciable capsule. There might be fibrous strands seen within this accumulated fat with cortical thickening, osseous hypertrophy and bony outgrowths from the underlying bone. Neural thickening may also be visualized.

MRI is helpful in distinguishing MDL from other causes of macrodactyly. Differential diagnosis of MDL includes neurofibromatosis, fibrolipomatous hamartoma, hemangioma, arteriovenous malformation, Klippel-Trenaunay-Weber syndrome. In fibrolipomatous hamartoma of the median or ulnar nerve, there is fatty tissue accumulation within the nerve sheath rather than within the region [13].

The condition is surgically treated with debulking operation and/or partial amputation to improve the cosmetic

appearance while preserving the neurologic function of the involved part.

Progressive soft tissue enlargement can be benign or malignant and must be assessed meticulously to rule out the possibility of maliganant entities like schwannoma, neurofibroma and soft tissue sarcoma. Characteristic history and imaging findings may help narrow down the diagnosis.

Conclusion

MDL should be included in the differentials of progressive congenital swelling of the fingers with characteristic imaging findings. Macrodactyly and nerve involvement can be accurately defined by MRI study to plan appropriate management to restore the function of the involved hand and remove the psychological burden of the patient for it being malignant.

Patient consent

A written informed consent was obtained from the patient for publication of this case, images and the clinical details.

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