An unusual case of macrodystrophia lipomatosa: Imaging and pathological correlation

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Abstract

Macrodystrophia lipomatosa is a rare cause of non hereditary congenital gigantism of limb characterised by disproportionate growth of fibro-adipose tissue with progressive proliferation of all mesenchymal elements. It comes to clinical attention for cosmetic reasons. However it may also present with changes associated with degenerative joint disease, or features of neurovascular compression. It usually presents as localised gigantism, most commonly involving the 2nd or 3rd digit of the hand or foot due to an increase in subcutaneous adipose tissue. Rare imaging features, in a case of Macrodystrophia Lipomatosa that predominantly involved the fifth digit is presented.

Keywords: *Macrodystrophia Lipomatosa, fibro-adipose tissue, Localised Gigantism, Macrodactyly.*

Introduction

Macrodystrophia lipomatosa (MDL) is a non hereditary congenital developmental anomaly characterised by local gigantism due to overgrowth of all mesenchymal elements such as bone, tendon, nerve, vessels and particularly fibro adipose tissue leading to local hypertrophy[1]. These patients are rarely symptomatic. What brings them to a physician is the cosmetic disfigurement which interferes with their normal social life. Imaging plays an important role in confirming the diagnosis and demonstrating whether it is suitable for surgical operative procedure or not.

Case Report

A 22 years old female was referred for imaging analysis as she had disproportionate enlargement of the little finger of right hand [Fig. 1]. She had no pain or tingling numbness. Clinical examination showed no signs of local inflammation or increased vascularity. None of her family members had history of similar disorder. Patient felt embarrassed because of the swelling. This insidious onset, painless, progressive enlargement of her right little finger started when she was 6 years of age and progressed to the present size of 8.0 x 2.6 cm which was significantly larger compared to the normal left little finger which measured 6.0 x 1.1 cm.

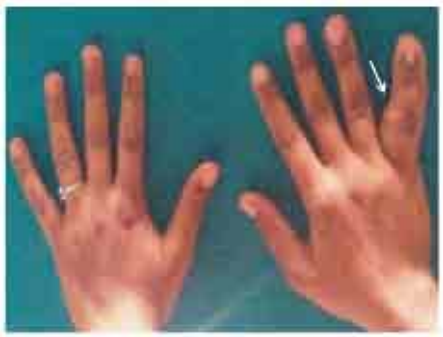

Figure 1: Photograph of both hands showing enlarged little finger of right hand.

The enlargement/swelling was non tender, non pulsatile diffuse soft tissue swelling over the volar aspect of the right little finger and extending up to the medial aspect of the wrist and palm. The overlying skin was pale and thick. Pitting oedema and bruit were absent. The initial physical examination indicated the presence of macrodactyly associated with presence of soft,

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fibrolipomatous tissue involving the little finger of the right palm.

Plain radiograph showed soft tissue thickening of right little finger without any bony deformity [Fig. 2]. High resolution ultrasound (HR-USG) demonstrated prominent diffuse subcutaneous adipose tissue involving the little finger [Fig. 3]. CT scan confirmed the fact that

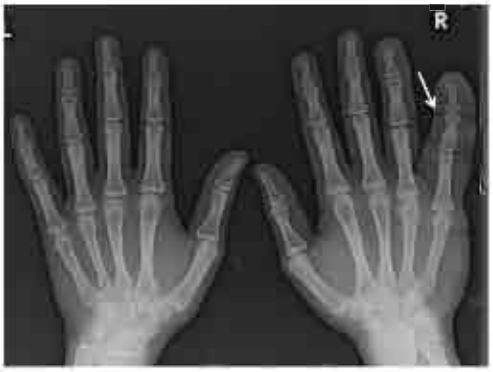

Fig. 2: Plain Radiograph of both hands (PA view) Non osseous soft tissue enlargement of little finger of right hand.

the diffusely enlarged soft tissue was adipose in nature as the density of this tissue was (-80) to (–112) H.U. which is typical for fat [Fig. 3]. T1W axial MRI images reiterated CT findings by demonstrating diffuse thickening of the soft tissues over palmar aspect of entire

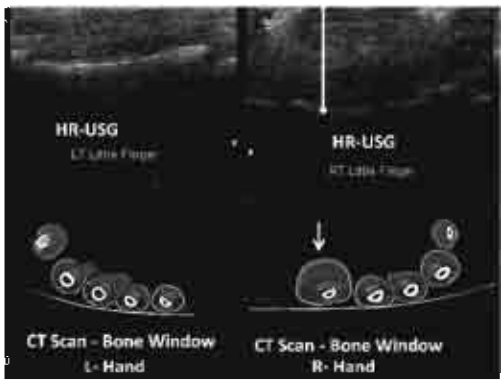

Fig 3: HR-USG and Bone window images of CT scan of both hands. There is soft tissue enlargement of little finger of right hand due to adipose tissue.

little finger as well as over the medial aspect of the proximal phalanx of ring finger and hypothenar eminence of the right hand which appeared hyperintense on T1W and hypointense on fat suppressed sequences. MRI also showed linear hypointense fibrous strands within the lesion on T1W and T2W images [Fig. 4]. The underlying

Fig. 4: MRI-T1W Axial images of Right hand confirming the presence of fat in the soft tissue enlargement. No bony abnormality seen.

flexor tendons of right little finger showed normal signal intensity. The underlying bones and their marrow appeared normal. The ulnar nerve and its branches in the affected region did not show any fibrolipomatous hamartoma. The diagnosis of MLD was made by imaging studies alone and was subsequently confirmed by surgical excision of the enlarged soft tissues.

Microscopic examination revealed fibro-fatty tissue and abundant mature adipocytes [Fig. 5].There was no evidence of neurofibromatosis.

Discussion

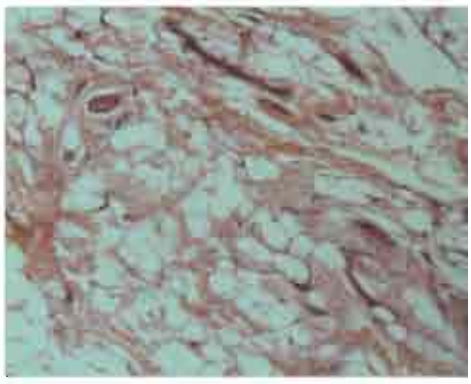

Fig. 5: Microscopic examination revealed fibro-fatty tissue.

Macrodactyly is an unusual congenital anomaly characterised by hamartomatous proliferation of soft tissues of the affected digit. It is also called local gigantism. The disorder is congenital but not hereditary. It is characterised by excessive growth of fibro-fatty tissue with unusually large fatty lobules, apparently fixed by a mesh of dense fibrous tissue. In MDL one entire extremity may be affected. The abnormal fat deposits are predominantly limited to the subcutaneous tissue,

periosteum and muscle, but may occasionally involve the nerve sheath.

Exact etiology of the disorder is not known[2]. However three probable causes have been described: Abnormal nerve supply, abnormal blood supply, or humoral mechanisms[3]. The rate of accelerated growth varies among different patients and also among affected digits. Progressive macrodactyly is more common than the static type. It may be present at birth along with syndactyly, digital deviation, thenar eminence hypertrophy, palmar and forearm hyperplasia[4].

Association with small osseous protuberances resembling osteochondromas and lipomas in other parts of the body have also been reported[5]. The involvement of the little finger is a rare and is almost always unilateral as seen in the present case.The lower limb is more frequently involved than the upper limb, which is typically along a specific sclerotome; with the most common sites being second and third digits corresponding to the median nerve and medial plantar nerve in upper and lower limbs, respectively[6].

Different imaging modalities play different roles in the evaluation of MDL. Conventional radiographs in such cases shows macrodactyly and soft tissue overgrowth, most marked along the volar aspect of the digit at its distal end[7]. Ultrasound shows enlarged soft tissue and lack of profuse vascularity. CT scan and MRI, both can charcterise the lesion, as these modalities have the potential to demonstrate the fatty nature of the enlarged soft tissue. The CT density / Hounsfield value of the lesion is (–100) to (-180) HU which indicates fatty composition. An MRI feature of the same is T1 hyperintense lesion which get suppressed on fat saturated sequences.

The differential diagnosis includes neurofibromatosis, hemangiomatosis, lymphangiomatosis, Proteus syndrome, and fibrolipomatous hamartoma (FLH).

Neurofibromas are visible in neurofibromatosis, which demonstrate marked hyperintensity on magnetic resonance imaging (MRI) in T2- weighted (T2W) images. These are seen in close proximity to nerves. Presence of positive family history, cutaneous lesions, and bilaterality favour the diagnosis of neurofibromatosis, while in Macrodystrophia lipomatosa, hypertrophy occurs along a nerve territory. Presence of unilaterality and fat within the nerve sheath favour a diagnosis of MDL.

Lymphangioma presents as a focal or diffuse swelling with pitting edema. On CT scan, these appear as multiseptated hypodense masses. On MRI, these appear hyperintense to muscle on T1W and hyperintense to fat onT2W images. A bruit may be palpable in hemangiomatosis, and on MRI, long repetition time/echo time (TR/TE) sequences may show a septated configuration of high-signal intensity channels, corresponding to the vascular channels and fibrous strands.

Klippel Trenaunay-Weber syndrome is a rare condition, usually present from birth. It involves port wine stains, excess growth of bones and soft tissue and varicose veins. Osseous growth is not seen in both lymphangiomatosis and hemangiomatosis.

In Proteus syndrome, hemi-hypertrophy occurs that may simulate MDL, but other associated abnormalities like calvarial changes, pulmonary cysts, pigmented nevi, and intra-abdominal lipomas favour the diagnosis of Proteus syndrome.

FLH of the nerve is a rare tumour like condition. In this condition, mature fat infiltrates the neural sheath, with most lesions occurring in the median nerve. Pathologically, in FLH, the deposition of fat occurs within the nerve sheath, while in MDL it occurs throughout the involved part of the digits/extremity. In FLH, neural fascicles separated by fat and connective tissue give a speckled appearance on MRI.

MDL comes to attention usually because of cosmetic complications. Amputation is the ultimate therapeutic modality and it is indicated in most cases involving the large digits[8]. Other surgical procedures performed in macrodactyly include stripping of the nerves from their branches or removal of a part of the nerve and end-to-end anastomoses to relieve pain[9,10].

Conclusion

Determination of the cause of macrodactyly is an enigma for a clinician as there are many probable etiologies. Differentiating between these conditions is important as they differ in their course, prognosis, complications, and treatment. Imaging helps in distinguishing these conditions from other causes of localised gigantism. CT scan and MRI confirm the diagnosis. MRI can demonstrate associated complications like hamartomas and bone involvement. Thus imaging modalities have the potential to solve this diagnostic enigma.

Consent

Written informed consent was taken in vernacular from the patient for publication of details, reports and images of this case

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