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Case Report

Radiology Section

Macrodactyly Lipomatosa in the Left Index Finger and Thumb: A Radiological and Clinical Perspective

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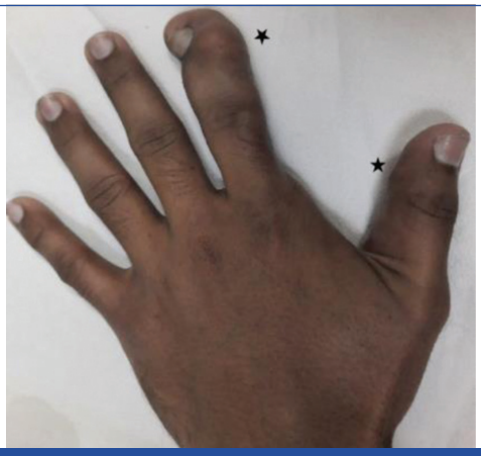
ABSTRACT

Macrodystrophia Lipomatosis (MDL) is a rare congenital, non-hereditary, and benign form of gigantism characterised by the overgrowth of adipose tissue in a specific area of the body, usually affecting the hand or foot in the distribution of the median and plantar nerves, respectively. Here, the authors present a case of a 21-year-old male patient, who presented to the Surgical Outpatient Department (OPD) with the gradual, progressive enlargement of the left index finger and thumb over the last six months, experiencing difficulty in holding objects and maintaining grip. This case highlights the importance of radiological imaging in diagnosis and treatment planning. Macrodystrophia lipomatosis should be considered as a differential diagnosis for hypertrophic digits, even in adult patients.

Keywords: Adipose tissue, Congenital, Gigantism, Plantar nerve

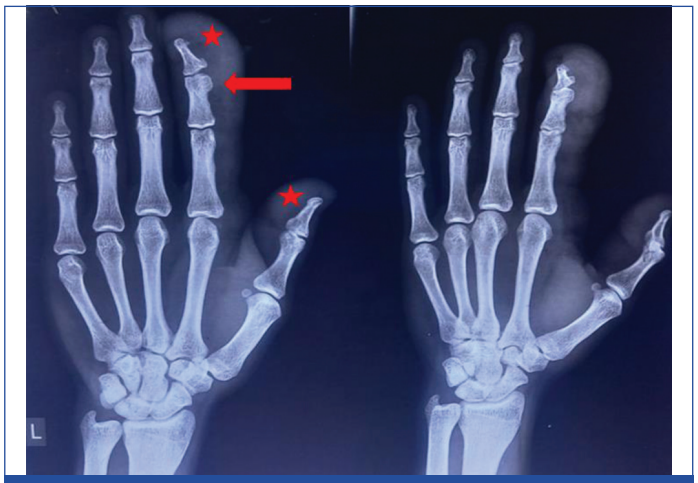
CASE REPORT

A 21-year-old male patient presented to the surgical OPD with gradual, progressive enlargement of the left index finger and thumb over the last six months, leading to difficulty in holding objects and maintaining grip. The left index finger and thumb had been enlarged since birth, but a significant increase in size occurred in the last six months [Table/Fig-1]. There was no previous history of trauma or significant medication intake. Upon examination, the left thumb and left index finger were enlarged, but no tenderness or discoloration was noted, and there was no neurological deficit or restriction of range of movement.

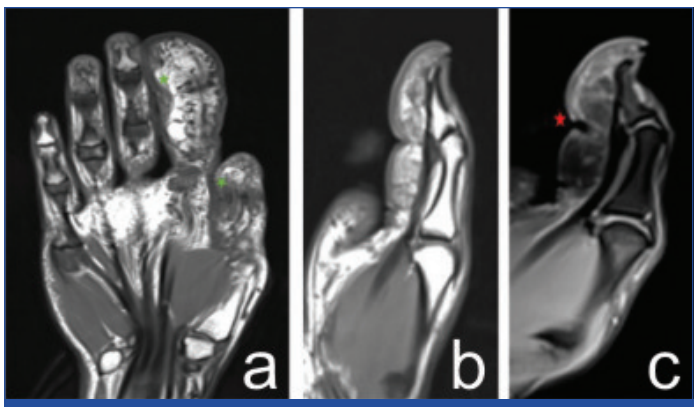


[Table/Fig-1]: Clinical view of left-hand shows increased circumference of left thumb and index finger (black star).

A conventional radiograph demonstrated an increased soft-tissue component in the index finger and thumb of the left hand. Osseous hypertrophy with bony outgrowth was noted at the head of the middle phalanx of the index finger [Table/Fig-2]. Magnetic Resonance Imaging (MRI) of the left hand revealed gross subcutaneous thickening along the radial aspect of the left index finger and the medial aspect of the left thumb, with suppressed fat signal intensity on Fat Suppressed Proton Density (FS-PD) sequences suggestive of fatty proliferation [Table/Fig-3]. The patient underwent debulking surgery with excision of the excessive lipomatous component. Histopathological examination of the excised mass confirmed the diagnosis of MDL, showing adipocyte cells, with no evidence of malignancy [Table/Fig-4]. The patient has been symptom-free and has not experienced a recurrence after the surgery during the three-year follow-up.



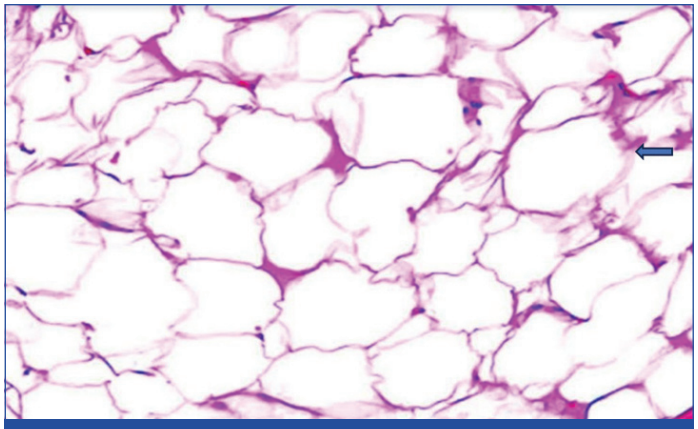
[Table/Fig-2]: Plain radiograph showing increased soft tissue growth in left thumb and left index finger (red star). Osseous hypertrophy with bony outgrowth is seen in the head of middle phalanx of the left index finger (red arrow).



[Table/Fig-3]: Magnetic Resonance Imaging (MRI) coronal T1W image showing a) subcutaneous thickening (green star) along the lateral aspect of the left index finger and b) the medial aspect of the left thumb, c) showing suppression on PDFS images suggestive of fatty proliferation (red star).

DISCUSSION

Macrodystrophia lipomatosa is a rare congenital disorder characterised by the excessive growth of mesenchymal tissue, predominantly adipose tissue, in the affected limb, resulting in the enlargement of one or more digits or extremities in a sclerotomal distribution [1,2]. The age of presentation ranges from as early as the neonatal period to late adulthood.



[Table/Fig-4]: The Hematoxylin and Eosin stain (40x) of the excised specimen showed adipocyte cells (arrow).

The exact pathogenesis of macrodystrophia lipomatosa is not well understood. Fat deposition is observed in the subcutaneous tissue, muscular compartments, along nerve sheaths, and may involve the periosteum with invasion of the medullary cavity.

It is classified into three types [3]:

Nerve territory-oriented type, with or without nerve enlargement, affecting the hand or foot in sclerotome distribution, as depicted in this case where the digits of the hand (thumb and index finger) are involved without nerve enlargement.

Diffuse lipomatous type involving the entire extremity with all digits without affecting the nerve.

Mixed pattern involving the enlargement of the digit in a sclerotome pattern with the enlarged limb.

Macrodystrophia lipomatosa typically presents as a unilateral enlargement of one or more digits or extremities. The affected digit or extremity is disproportionately larger than the opposite side, especially on the volar aspect. The enlargement may be progressive or static. Patients may also present with functional impairment, such as difficulty in fine motor movements, grasping objects, or performing daily activities. The longstanding pressure effect results in neurovascular compression, bony outgrowths, and arthritic changes. The osteophytes and early degenerative changes exacerbate the nerve compression further, causing entrapment syndromes [4]. Syndactyly, polydactyly, clinodactyly, symphalangism, fibro-lipomatous haematoma of the median nerve, pigmented nevus, and lipomatous growths in other parts can be associated with it [5].

Radiographic evaluation of macrodystrophia lipomatosa reveals soft tissue swelling due to increased adipose tissue density. Bony osteophytes, cortical thickening, and reduced joint space can also be seen. Ultrasonography may also demonstrate increased thickness of subcutaneous fat with or without enlarged nerve trunks [6].

The MRI is considered the imaging modality of choice for the diagnosis and evaluation of macrodystrophia lipomatosa. It shows high signal intensity on T1WI, intermediate signal on T2WI, and low signal on fat-suppressed sequences. MRI can reveal the extent and distribution of this unencapsulated adipose tissue involvement, as well as the presence of other associated abnormalities, such as nerve entrapment [7].

Imaging modalities and clinical findings help differentiate it from other causes of localised gigantism like Klippel-Trenaunay syndrome, Proteus syndrome, Maffucci syndrome, Ollier syndrome, Lymphangiomatosis, Beckwith-Wiedemann syndrome, Vascular malformation, lymphangiomatosis, and neurofibromatosis [8].

The outcome of macrodystrophia lipomatosa is variable, with some patients experiencing minimal functional impairment, while others may have significant disability. Treatment options for macrodystrophia lipomatosa are limited and may include conservative measures, such as physical therapy or occupational therapy, to improve hand function and reduce pain. Surgical intervention may be considered in cases of severe functional impairment or cosmetic disfigurement [9]. The surgical options include debulking procedures, with or without additional osteotomies, and amputation. Osteotomies and amputation are performed in cases with severe secondary bone and joint changes [10]. Complications post-surgery include nerve injury, a high recurrence rate, and an inapparent difference in size post-debulking. The recurrence rate and incidence of nerve injury post-debulking can range from 33% to 60% and 30% to 50%, respectively [4].

CONCLUSION(S)

Macrodystrophia lipomatosa is a congenital mass; however, it can present in adulthood. Thus, MDL, though a rare condition, should be considered in the differential diagnosis of any slow-growing, painless mass in the subcutaneous tissue of the hand or foot, irrespective of the age of presentation.

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