Case Reports in Oncology

Case Rep Oncol 2019;12:63–68

DOI: 10.1159/000496685
Published online: January 21, 2019

Case Report

Macrodystrophia Lipomatosa of the Finger: A Case Report

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Keywords
Macrodystrophia lipomatosa · Finger

Abstract
Introduction: Macrodystrophia lipomatosa is a rare benign condition characterized by a progressive persistent proliferation of the mesenchymal components and elements, with a disproportionate increase in fibro-adipose tissue. It causes a debilitating functional and psychological effect on the patient and his family. ***Case presentation:*** A 25-years-old male patient presented complaining of left middle finger swelling which was painless and progressive in size with no restriction of range of motion. Plain X-ray films of the left hand showed a heterogeneous lobulated soft tissue mass on the volar aspect of the middle finger which was confirmed by MRI scan. Surgery was performed by doing a left middle finger soft tissue excision and diagnosis of Macrodystrophia lipomatosa was confirmed by the histopathology report. Patient was on regular follow up in the clinic with no tumour recurrence and an excellent cosmetic as well as functional result with full finger joints range of motion. ***Conclusion:*** Macrodystrophia lipomatosa should be suspected with this kind of presentations. Combination of the clinical history and examination with the aid of radiological assays aided in the diagnosis of this extremely rare case and led to the appropriate management by excising this mass and restoring the normal function of the patient's hand.

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Introduction

Macrodystrophia lipomatosa (MDL) is a rare nonhereditary benign congenital condition, presents in the form of localized focal gigantism. It is usually characterized by persistent, progressive proliferation of the mesenchymal components and elements, with a disproportionate increase in fibro-adipose tissue [1, 2]. MDL affects either the upper or lower limb, classically occurring in a median or plantar nerve territory [1, 3]. In the literature its commonly described as lipofibromatous hamartoma (LFH), another synonym that represents a broader description, although, in the association with macrodactyly where the growth involves all mesenchymal elements it is often referred to as MDL [1]. Due to the lack of demographic evaluation by cohort studies, the incidence is still unknown [4]. However, those who are affected by the condition, usually suffer from a debilitating functional and psychological effect both on them and their respective families.[5] This work reported in line with the CARE criteria [6].

Case Presentation

A 25-year-old male patient who is otherwise healthy referred to our Sarcoma Multidisciplinary Clinic with the suspicion of a primary bone lesion, as we are the regional center for treatment of benign and aggressive musculoskeletal tumors. He complains of progressive left middle finger swelling. His family first noticed the swelling after an incident of direct trauma to the finger during early childhood. Before that, there were no abnormalities seen by the parents. The swelling was progressive in size until he underwent a local excision procedure, at the age of four years. Despite surgical intervention, the swelling recurred. The swelling was painless but continued to enlarge in size with age, with no restriction of range of motion or neurological symptoms. His main concern was cosmetic as he feels it's causing him a psychological burden: no family history of any genetic disorders, nor any similar musculoskeletal deformities. Physical examination revealed a left middle finger healed scar over the dorsal aspect of the finger; a soft tissue mass can be appreciated distal to the proximal interphalangeal joint (PIPJ) with fatty looking lobules and macrodactyly with no signs of inflammation. Full proximal and distal interphalangeal joint range of motion, fingernail neurovascular examination is within normal (Fig. 1A, B). No lymphadenopathy, systematic review is unremarkable particularly signs of neurofibromatosis. All blood investigation showed no abnormalities.

Plain X-ray films of the left hand showed a heterogeneous lobulated soft tissue mass on the volar aspect of the middle finger involving mainly the middle and distal phalanges, no arthritic changes in the joints. Furthermore, left hand MRI with IV contrast media showed a distal fatty overgrowth with striation and no capsulation surrounding the middle and distal phalanges of the middle finger, more at the palmer aspect (Fig. 2).

The treatment plan was discussed with the patient in the form of left middle finger soft tissue excision with the possibility of a skin graft or local flap coverage with the assistance of the plastic surgery team. With the placement of a tourniquet under general anesthesia, an elliptical incision made including the previous surgery scar crossing the volar aspect of the middle finger (Fig. 1C, D). Dissection around the soft tissue mass, the ulnar digital artery identified and preserved, the digital nerve is visualized proximally and found encased into the mass hence was sacrificed. The lipomatous mass was found to be thick with associated fibrous component, it was completely excised, and sent for histopathology examination. At the time of surgery, the plastic surgery team was present in case more advanced soft tissue management techniques were required. Thankfully closure was successful with the aid of a Z-Plasty, and

there was no need for coverage by a skin graft. Skin closure by 5.0 Nylon suture done and wound dressing applied. The surgery performed by the author. Distal neurovascular examination within normal. Post-operatively patient was pain-free with good distal capillary refill and discharge home in a stable condition. Advised for limb elevation and keep dry dressing. Stitches removed after two weeks and patient started on range of motion therapy to avoid joint stiffness. He was compliant and tolerated the post-operative plan with no reservations. Pathology report came back with the diagnosis of lipofibromatous hamartoma of the nerve, macrodystrophia lipomatosa of the finger (Fig. 3). The patient was on regular follow up in the clinic, and last clinical visit was two years following surgery and showed no tumor recurrence an excellent cosmetic as well as functional result with full finger joints range of motion (Fig.1E, F).

Discussion

MDL is a form of macrodactyly that affects mainly the hands and feet, commonly involving the middle and index fingers along the area that is supplied by the median nerve [7, 9]. Ulnar nerve distribution involvement is sporadic [1, 2]. The presentation is usually unilateral, with bilateral limb involvement being infrequent, with an equal distribution among both genders [2, 10]. Although it described in the literature as lipofibromatous hamartoma (LFH) the term macrodystophia lipomatosa used with the association of macrodactyly [1]. Other names used as lipofibroma, fibrolipoma, intraneural lipoma [11, 12].

In this case, we report a unilateral progressive swelling on the right middle finger associated with macrodactyly which mimics the presentation of MDL in the previous literature [1, 2, 7–10]. Approximately 108 cases were reported and identified as MDL since 1950, with the first one presented by Feriz in 1925 [5, 13]. Tahiri et al., did a comprehensive review of LFH in the literature and looked into 180 cases between 1946 to 2012. They reported 32% of cases associated with macrodactyly, and 8% were due to trauma which is related to our case [14].

Clinical presentation varies due to the etiology of the condition and nerve involvement. A progressive mass enlargement of the forearm, wrist, and digits compromises the majority of presentations [14]. Neurological symptoms with varying degrees have been described depending on which nerve is involved and degree of involvement, where the median nerve was the most common [14]. Carpal tunnel syndrome observed in association with MDL and LFH which warranted an additional surgical intervention in some cases [14].

MDL categorized into two types, the first type is static which is characterized by proportional growth of the diseased site, in the other hand, a disproportional progressive growth distinguishes the second type; hence it's called progressive [5, 8]. The static type is less common than the progressive [4]. Hypotheses about the pathogenesis of MDL is controversial but mainly attributed to either lipomatous degeneration that could arise from trauma, abnormal fetal vasculature and circulation, extremity bud damage or intrauterine segmentation errors and affected nerve hypertrophy [4, 15].

Radiological modalities have a significant role in diagnosing and evaluating MDL including Plain radiograph (X-ray), Computed tomography (CT), Ultrasonography (US) and Magnetic Resonance Imaging (MRI). X-ray findings include overgrowth of the soft tissue and bony structures with translucency detected in the soft tissue as a result of increased infiltration of adipose tissue. Increased bone growth and proliferation of fat along the area innervated by concerned nerve observed in MRI scan. A typical finding of MRI is excessive unencapsulated

fibro-fatty infiltration [2, 15, 16]. MRI and X-ray highly contributed to diagnosing our patient with MDL which was confirmed by histopathology.

Differential diagnosis of macrodactyly that must be in consideration while evaluating patients with hand or foot hypertrophy and swelling include hemangiomatosis, Proteus syndrome, neurofibromatosis, lymphangiomatosis and Klippel-Trénaunay-Weber syndrome [15].

Progressive soft tissue mass enlargement raises the suspicion of benign and malignant tumors. Benign conditions such as lipoma, ganglion cyst, congenital syndromes as Dejerine Sottas and Weber syndrome, as well as a traumatic neuroma [17]. Caution must be in consideration for isolated soft tissue masses with worrisome MRI findings that suggest potential malignant entity such as schwannoma, neurofibroma, and soft tissue sarcoma [15].

In conclusion, Macrodystrophia lipomatosa should be in consideration with this kind of presentations and can be misdiagnosed easily. Combination of the clinical history and examination with the help of radiological assays and histopathology aided in the diagnosis. Recognition of the presence of macrodactyly and investigating its causes along with the identification of nerve involvement can and led to the appropriate management. Restoring the normal function of the patient hand and relief of the psychological burden from the patient should be the primary goal.

Statement of Ethics

The authors have no ethical conflicts to disclose.

Disclosure Statement

The authors declare no conflict of interest.

References

1 Goldman AB, Kaye JJ. Macrodystrophia lipomatosa: radiographic diagnosis. AJR Am J Roentgenol. 1977 Jan;128(1):101–5.
2 Singla V, Virmani V, Tuli P, Singh P, Khandelwal N. Case Report: macrodystrophia lipomatosa - Illustration of two cases. Indian J Radiol Imaging. 2008 Nov;18(4):298–301.
3 Fritz TR, Swischuk LE. Macrodystrophia lipomatosa extending into the upper abdomen. Pediatr Radiol. 2007 Dec;37(12):1275–7.
4 Mehra R, Bhartiya R, Agrawal P, Kumari N. Macrodystrophia Lipomatosa: clinico-patho-radiological Correlation. Indian J Med Paediatr Oncol. 2017 Oct-Dec;38(4):559–62.
5 Prasetyono TO, Hanafi E, Astriana W. A Review of Macrodystrophia Lipomatosa: revisitation. Arch Plast Surg. 2015 Jul;42(4):391–406.
6 Gagnier JJ, Kienle G, Altman DG, Moher D, Sox H, Riley D; CARE Group*. The CARE Guidelines: Consensus-based Clinical Case Reporting Guideline Development. Glob Adv Health Med. 2013 Sep;2(5):38–43.
7 Kelikian H. Congenital deformities of the hand and forearm. Philadelphia: Saunders; 1974. pp. 610–35.
8 Barsky AJ. Macrodactyly. J Bone Joint Surg [Am]. 1967;49-A: 1255-1 265
9 Blacksin M, Barnes FJ, Lyons MM. MR diagnosis of macrodystrophia lipomatosa. AJR Am J Roentgenol. 1992 Jun;158(6):1295–7.
10 D'Costa H, Hunter JD, O'Sullivan G, O'Keefe D, Jenkins JP, Hughes PM. Magnetic resonance imaging in macromelia and macrodactyly. Br J Radiol. 1996 Jun;69(822):502–7.
11 Morley GH. Intraneural lipoma of the median nerve in the carpal tunnel—report of a case. J Bone Joint Surg Br. 1964 Nov;46(4):734–5.
12 Rusko RA, Larsen RD. Intraneural lipoma of the median nerve—case report and literature review. J Hand Surg Am. 1981 Jul;6(4):388–91.

13 Feriz H. Makrodystrophia Lipomatosa progressiva. Virchows Arch. 1925;260(2):308–68.
14 Tahiri Y, Xu L, Kanevsky J, Luc M. Lipofibromatous hamartoma of the median nerve: a comprehensive review and systematic approach to evaluation, diagnosis, and treatment. J Hand Surg Am. 2013 Oct;38(10):2055–67.
15 Colonna MR, Guarneri C, do Stagno d'Alcontres F, Risitano G, Cavallari V, Ribuffo D. Invasive macrodystrophia lipomatosa of the hand. Eur Rev Med Pharmacol Sci. 2014;18(18):2621–3.
16 Brodwater BK, Major NM, Goldner RD, Layfield LJ. Macrodystrophia lipomatosa with associated fibrolipomatous hamartoma of the median nerve. Pediatr Surg Int. 2000;16(3):216–8.
17 Terzis JK, Daniel RK, Williams HB, Spencer PS. Benign fatty tumors of the peripheral nerves. Ann Plast Surg. 1978 Mar;1(2):193–216.

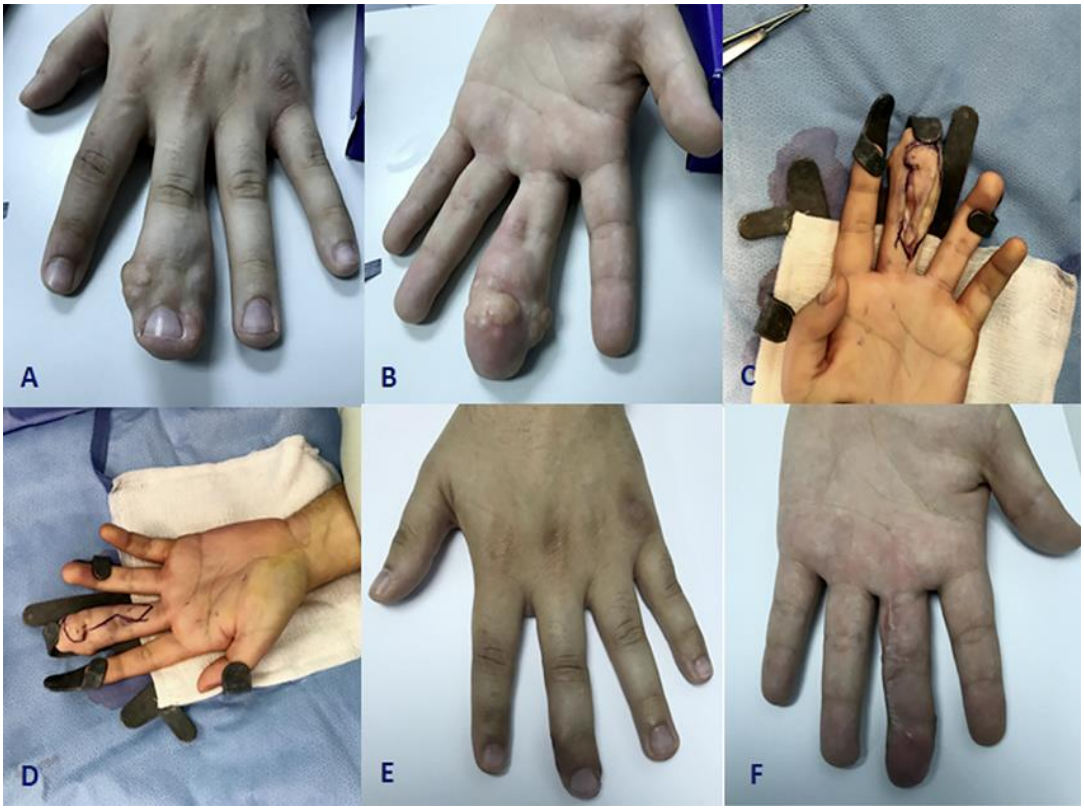

Fig. 1. **A**, **B** showed the clinical picture of the marked hypertrophy and enlargement of the middle finger. **C**, **D** intraoperative dissection of the digital nerve with the associated hypertrophied lipomatous tissue. **E**, **F** postoperative images showed complete removal of the mass and marked reduction in the size of the digit.

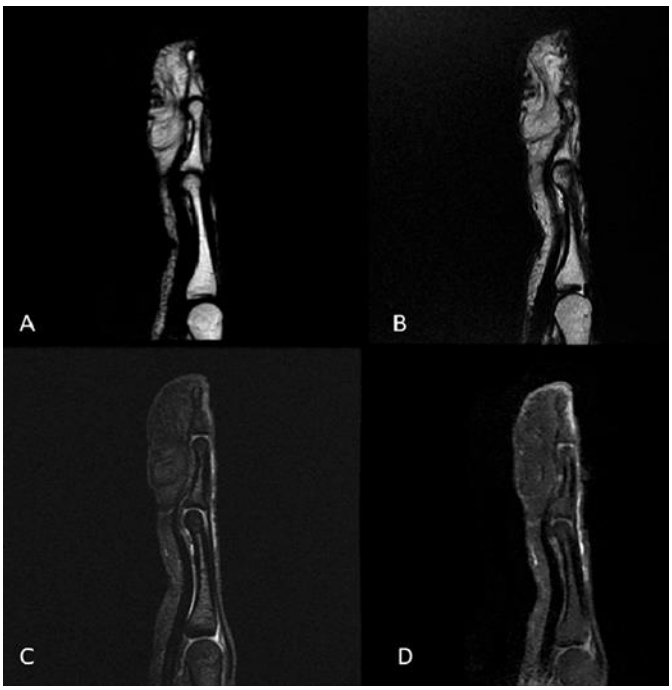

Fig. 2. **A**, **B** showed MRI of the middle finger with ill-defined soft tissue mass inspirable from the surrounding fat, mainly at the palmar aspect of the middle and distal phalanx. This mass has fat signal intensity in the form of bright T1 and T2 weighted images. **C**, **D** images showed a drop of signal in the fat sat sequences and no solid enhancing nodule on contrast-enhanced images. It shows central striation with multiple blood vessels coursing through it rather than being displaced by it. The overall picture is suggestive of distal phalangeal lipomatosis (localized Macrodystrophica lipomatosa of the distal median nerve).

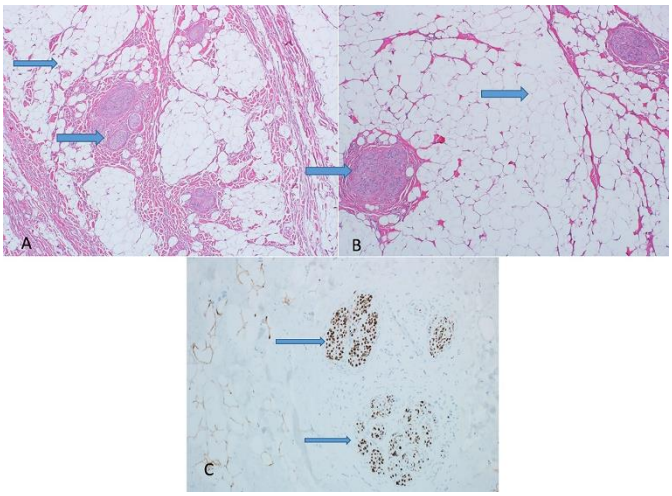

Fig. 3. In histopathology, A Gross specimen consists of two fragments of fibrofatty tissue with skin ellipse measuring 7 × 3 × 2 cm and 2 × 0.5 × 0.5 cm respectively. The resected specimen submitted for microscopic examination. **A**, **B** H/E slides showing nerve bundles surrounded by dense collagen and mature adipose tissue. **C** S100 IHC staining the nerve bundles in trapped within mature adipose tissue.