CASE REPORT

Macrodystrophia lipomatosa with fibrolipomatous hamartoma of medial plantar nerve and lipomatosis of thigh

Taruna Yadav (1), Galib Mirza (1), Pushpinder Singh Khera (1), Pawan Garg (1)

(1) Department of Diagnostic and Interventional Radiology, All India Institute of Medical Sciences, Jodhpur-342008, Rajasthan, India

ABSTRACT

Macrodystrophia lipomatosa (MDL) is a rare congenital non-hereditary condition characterised by an abnormal overgrowth of mesenchymal tissue of a digit or an extremity. Although MDL is interchangeably described by terms such as macrodactyly and macrosomia, the entity has to be differentiated from other acquired and hereditary causes of macrodactyly. Rare association with osteochondromas and lipomas in other parts of the body has also been reported. We present a case with gradual progressive swellings of the right great toe and right thigh, which was diagnosed to be a case of MDL of the great toe with fibrolipomatous hamartoma of medial plantar nerve and lipomatosis of the thigh based on characteristic imaging findings.

KEYWORDS

Macrodystrophia lipomatosa; Fibrolipomatous hamartoma; Lipomatosis Extremity.

INTRODUCTION

Macrodystrophia lipomatosa (MDL) is an important rare cause of macrodactyly. Here,

hypertrophy of mesenchymal elements, particularly fibroadipose tissue, occurs in a nerve territory distribution. The most common site of involvement is along medial plantar nerve in the lower limbs and along median nerve distribution in the upper limbs. Several terms including partial acromegaly, macrosomia, elephantiasis, megalodactyly, macrodactyly, macroceir, club finger and others have been used synonymously to describe localized gigantism of a digit [1]. Nevertheless, it is essential to differentiate this non-hereditary condition from other conditions of macrodactyly that are either hereditary, syndromic or acquired.

CASE REPORT

A 10-year-old schoolboy presented with gradual painless progressive enlargement of the great toe of the right foot as well as a swelling around the right knee, extending from the lower thigh to the mid leg (Figure 1). Swellings were noticed by parents since infancy, but they were concerned now due to a gradual increase in size. His major complaints were cosmetic disfigurement and difficulty fitting into shoes. He had no history of trauma or pain or other clinical symptoms. The patient had consulted two paediatricians

Correspondence to:

Taruna Yadav
Department of Diagnostic and Interventional Radiology, All India Institute of Medical Sciences, Jodhpur, India
Email: drtanu.taruna@gmail.com

Received: 06 October 2019 | Accepted: 17 February 2020

How to cite this article:

Taruna Yadav T, Galib Mirza G, Pushpinder Singh Khera PS, Pawan Garg P. Macrodystrophia lipomatosa with fibrolipomatous hamartoma of medial plantar nerve and lipomatosis of thigh. Sudan J Paediatr. 2020;20(1):62–67.
<https://doi.org/10.24911/SJP.106-1570297771>

Figure 1. Clinical photograph showing enlargement of soft tissues along the right foot in the region of first metatarsal and phalanges of the great toe (black arrows). Another soft tissue swelling is seen along right knee (white arrows) with normal width of the lower leg.

before coming to the institute, but no definitive diagnosis was made. There was no family history of similar complaints or neurofibromatosis (NF). On examination, no overlying skin changes, pitting or bruit were noted. He did not have any neurocutaneous manifestations of NF.

Radiograph of the right foot demonstrated that the bones (first metatarsal, proximal and distal phalanges) of the great toe were enlarged and bulkier with surrounding soft tissue swelling, but the cortex and their normal form were preserved. The radiograph of the right knee revealed an abnormal soft tissue density around the right knee joint with no bony abnormality (Figure 2).

Figure 2. (a) Radiograph of the right foot demonstrated that the bones (first metatarsal, proximal and distal phalanges) of the great toe were enlarged in both the length and width with surrounding soft tissue swelling, but the cortex and their normal form were preserved. (b) Radiograph of the right knee revealed ill-defined abnormal soft tissue density from lower thigh to upper thigh.

MR imaging confirmed an abnormal overgrowth of predominantly fibroadipose tissue, resulting in focal gigantism of the right first metatarsal region and great toe (over dorsal and plantar side) with incurving of soft tissue. Abnormal unencapsulated soft tissue growth was predominantly fat-containing with few fibrous strands. The median plantar nerve was enlarged with the presence of fat in-between the nerve fascicles giving a 'coaxial cable appearance' suggestive of fibrolipomatous hamartoma of the nerve (Figure 3).

In addition, a large unencapsulated asymmetric lipomatous lesion was evident in the right lower thigh and upper leg, measuring approximately 20 cm. This lipomatous lesion was extending from anterior subcutaneous tissue into the knee joint posterior to patellar tendon (Figure 4). Rest of the knee joint structures and ligaments

Figure 3. (a,b) Sagittal T1- and T2-weighted images show asymmetric hypertrophy of fibroadipose tissue along the plantar surface of great toe (white arrows), resulting in dorsal angulation of distal phalanx of great toe, respectively. (c) Axial T1 fat-suppressed (FS) image shows suppression of signal in the great toe soft tissue similar to rest of the subcutaneous fat in other phalanges. Axial (d) T2 and (e) T1 images show enlargement of the medial plantar nerve along with the presence of fat in-between the nerve fascicles giving 'coaxial cable appearance' suggestive of fibrolipomatous hamartoma of the nerve (red arrows) with fat signal suppression on T1FS image (f) with no bony abnormality.

were normal. The underlying bones in the leg and foot showed normal signal intensity with no evidence of periosteal thickening, marrow oedema or erosion.

Hence, a diagnosis of MDL of the great toe with fibrolipomatous hamartoma of medial plantar nerve and thigh lipomatosis was made. Counselling was done regarding the conservative and surgical treatment options. As the progression of MDL might cease at puberty, the patient chose conservative treatment and follow-up until puberty.

DISCUSSION

As coined by the physician Feriz in 1925 to describe localized gigantism of the lower extremity, the term 'MDL' is used to denote a rare congenital non-hereditary condition characterised by the overgrowth of all mesenchymal elements of a digit or an extremity [1,2].

There is an overgrowth of all mesenchymal-derived elements including fibroadipose tissue, bones, tendons, nerves and vessels with disproportionate growth of fibroadipose tissue element in a sclerotome distribution. This

Figure 4. (a) Sagittal T2WI shows unencapsulated hypertrophy of subcutaneous fat in the lower thigh to leg region (white arrows), extending into knee joint (green arrow). (b) Axial T1WI shows fat anterior to patella (red arrows) which is suppressed on T1FS (c) images.

excessive fibroadipose tissue is not encapsulated [3,4]. The cause is unclear although various hypotheses relating it to abnormal foetal circulation, nerve hypertrophy and so on have been postulated. PIK3CA gene mutation is associated in few cases. The condition is apparent since infancy and usually produces no concern except related to cosmesis. Growth ceases at puberty and occasional clinical manifestations include secondary degenerative joint disease and neurovascular compression symptoms due to tissue overgrowth [4].

Although the second and third digits of the lower extremity are most commonly involved, it is seen to involve other digits and the upper extremity too along the distribution of the medial plantar and the median nerves, respectively, predominantly affecting the distal and volar aspect. Syndactyly, clinodactyly, polydactyly or symphalangism can be associated with it [1–3].

Based on the pattern of involvement, MDL can be divided into three types: (a) ‘nerve territory-

oriented type’ which is the most common pattern. Here, hypertrophy of soft tissues particularly that of fat occurs in a nerve territory/sclerotomal distribution with or without nerve enlargement. (b) ‘Diffuse or pure lipomatous type’ – least common pattern. Only adipose tissue hypertrophy occurs involving the whole extremity with all the digits involved in the hand/foot without the involvement of the nerves. (c) ‘Mixed pattern type’ can occur in some cases which show adipose tissue hypertrophy in a nerve territory in the hand/foot with diffuse lipomatous hypertrophy in the remaining limb [3,4,8]. This case had a mixed pattern type of distribution.

Nerve territorial distribution can also be divided into (a) along a single nerve (median or median plantar nerve) and (b) along adjacent nerves in the same limb (the median and ulnar nerve in the upper limb, medial and lateral plantar nerves in the lower limb) [4,8]. Our case had single nerve distribution along medial plantar nerve. Goldman and Kaye described six cases of MDL. Lower

extremity was more commonly affected along with second digit involvement (hand or foot) in their series, which corresponds to medial plantar nerve in the foot and median nerve in the hand [1]. Our case had first digit (great toe) involvement.

Prabhu et al. [4] described the largest series (31 cases) of MDL with unilateral involvement in 30 cases. Median nerve territory was most commonly involved in the upper limb with medial plantar nerve territory in the lower limb. Phalangeal abnormalities such as syndactyly (two cases), polydactyly (one case) and symphalangism (one case) were also present in few cases.

Based on the growth pattern, MDL can be as static or progressive [1,5]. Static means the affected digit is growing at the same rate as the rest of the limb. This case was progressive type in which the affected digit is growing more rapidly than the rest of the limb.

Different imaging modalities can be employed for the diagnosis. On plain radiographs, typically soft tissue and bony hypertrophy with lucencies in the soft tissue mass due to adipose tissue may be seen with elongated, broad and splayed phalanges. Secondary osteoarthritic changes including joint space narrowing, subchondral cysts and osteophytes may be seen later in the course of the disease [4].

The role of ultrasound (USG) is in primarily differentiating it from other causes of limb swellings including haemangiomas, lymphangiomas and so on. USG can demonstrate unencapsulated fatty tissue comprising the swelling. MRI stands out in demonstrating the abnormal unencapsulated fibrofatty tissue which has similar signal characteristics to subcutaneous fat (i.e., high signal intensity on T1W and T2W and low on fat-suppressed sequences) with fibrous strands which are seen as thin linear T1 hypointense strands. Fibrolipomatous hamartoma (FLH) of the median nerve or medial plantar nerve may be associated with MDL. In FLH, nerves are enlarged with fatty tissue proliferation in-between the nerve fascicles giving them a 'coaxial cable appearance'. In MDL, fat deposition occurs throughout the involved part of the digits/

extremity. However, MDL may be associated with FLH in 30–66% of cases [6].

The main differential diagnoses are NF, haemangiomas, lymphangiomas, Proteus syndrome and Klippel–Trenaunay–Weber syndrome. Most important differential is NF. Diagnostic dilemmas can occur in the cases of plexiform neurofibromas with macrodactyly or hemihypertrophy. Features favouring NF include a positive family history, bilaterality as well as cutaneous stigmata of NF. Periosteal abnormalities in NF may lead to a wavy appearance of the cortex of the enlarged bones. Thinning of bones may also occur in NF. The extent of involvement of an extremity may not be contiguous. In MDL, the extent of involvement of distal phalanges is most severe, which is not always the case in NF. Premature fusion of growth plates can occur in NF due to hemangiomas element in the plexiform neurofibroma. NF is excluded by confirming the presence of fat within the nerve sheath on MRI [1].

Lymphangiomas presents with diffuse swelling of an extremity along with pitting oedema. It is hyperintense to muscle on T1W images and hyperintense to subcutaneous fat on T2W images. Haemangiomas are characterised by septated appearing high signal intensity channels on long TR/TE sequences due to vascular channels and fibrous strands comprising the lesion. A bruit may be audible on clinical examination. However, no osseous overgrowth is seen in either of haemangiomas or lymphangiomas [6].

Multisystemic manifestations are seen in Proteus syndrome. Macrodactyly along with connective tissue nevus, calvarial thickening, vascular malformations and intra-abdominal lipomas may help to make the correct diagnosis. Klippel–Trenaunay–Weber syndrome is diagnosed by a constellation of unilateral hemi hypertrophy with port wine stains, venous malformations or varicose veins [3,7].

Depending on the severity of the disease and associated complications in MDL, conservative management or surgical treatment may be offered to the patient [4,8].

CONCLUSION

Clinical differential diagnosis of macrodactyly is diverse. Awareness of the unique presentation and imaging features of MDL among the paediatricians, radiologists and paediatric surgeons is of paramount importance in making an early diagnosis. Appropriate imaging, including radiography and ultrasound, can make the determination of the underlying aetiology non-invasively. MRI is the investigation of choice in differentiating MDL from other causes of localised hemihypertrophy and determines the exact extent of pathology.

CONFLICTS OF INTEREST

The authors declare that there are no conflicts of interest.

FUNDING

None.

ETHICS

Signed informed consent for participation and publication of medical details was obtained from the parents of the patient. Confidentiality was ensured at all stages. The authors declare that ethics committee approval was not required for this case report.

REFERENCES

1. Goldman AB, Kaye JJ. Macrodystrophia lipomatosa: radiographic diagnosis. *Am J Roentgenol.* 1977;128: 101–105. <https://doi.org/10.2214/ajr.128.1.101>
2. Singla V, Virmani V, Tuli P, Singh P, Khandelwal N. Case report: macrodystrophia lipomatosa - illustration of two cases. *Indian J Radio Imaging.* 2008;18:298–301. <https://doi.org/10.4103/0971-3026.43844>
3. Mehra R, Bhartiya R, Agrawal P, Kumari N. Macrodystrophia lipomatosa: clinico-patho-radiological correlation. *Indian J Med Paediatr Oncol.* 2017;38:559–62. https://doi.org/10.4103/ijmpo.ijmpo_65_16
4. Prabhu CS, Madhavi K, Amogh VN, Panwala HK, Sathyakumar K. Macrodystrophia lipoma-tosa: a single large radiological study of a rare entity. *J Clin Imaging Sci.* 2019;9:4. <https://doi.org/10.25259/JCIS-9-4>
5. Ozturk A, Baktiroglu L, Ozturk E, Yazgan P. Macrodystrophia lipomatosa: a case report. *Acta Orthop Traumatol Turc.* 2004;38:220–3.
6. Blacksin M, Barnes FJ, Lyons MM. MR diagnosis of macrodystrophia lipomatosa. *AJR Am J Roentgenol.* 1992;158:1295–7. <https://doi.org/10.2214/ajr.158.6.1590127>
7. Upadhyay D, Parashari UC, Khanduri S, Bhadury S. Macrodystrophia lipomatosa: radiologic-pathologic correlation. *J Clin Imaging Sci.* 2011;1:18. <https://doi.org/10.4103/2156-7514.78264>
8. Prasetyono TO, Hanafi E, Astriana W. A review of macrodystrophia lipomatosa: revisita-tion. *Arch Plast Surg.* 2015;42:391–406. <https://doi.org/10.5999/aps.2015.42.4.391>