A Child with Enlarged Extremities - A Case of Macrodystrophia Lipomatosa

K Gunasekaran, N Sundareswaran, G Gopinath

Abstract

Macrodystrophia lipomatosa (ML) is a rare, non-hereditary, developmental anomaly that occurs because of the progressive proliferation of all mesenchymal elements of single or multiple digits or entire extremity, with a disproportionate increase in fibroadipose tissue. Commonly one or few digits of an extremity will be enlarged and present as macrodactyly or as enlarged limb. Lower limb involvement is more common and frequently unilateral. The diagnosis of ML is made by accurate clinical assessment and imaging modalities, such as plain X-ray, computed tomography scan, magnetic resonance imaging, and confirmed by histopathological study. In this case, we described a 10-year-old child who was brought to us with enlarged upper and lower extremities and was diagnosed as a case of ML with the help of clinico-radiological studies and presented here because of focal gigantism involving all four limbs, which is very rare.

Key Words: Enlarged extremities, focal gigantism, macrodactyly, macrodystrophia lipomatosa

Introduction

There are good number of causes for enlarged extremities in both pediatric and adult population. One among them is macrodystrophia lipomatosa (ML). It is congenital in origin but not hereditary. It is a rare developmental anomaly, presents with localized gigantism involving one or more extremities, commonly one or few digits will be enlarged and present as macrodactyly or as enlarged limb. Lower limb involvement is common and is frequently unilateral. Rarely, bilateral presentation is noted either in both lower or both upper limbs. To the best of our knowledge, localized gigantism due to ML involving all four extremities had not been reported. In this case study, we describe a 10-year-old child who had enlarged upper and lower extremities and was diagnosed to be a case of ML with the help of clinico-radiological studies.

Case Report

A 10-year-old male child was brought for enlarged feet and hands since birth. He was delivered normally at term with birth weight of 3.8 kg. There were no neonatal or maternal complications. At birth, parents noticed mildly enlarged feet and hands, and at 3 months, striking enlargement of feet and hands was observed. The enlarged extremities had always been disproportionately large and were progressive and fast growing in relation to the remaining body. His physical and mental developmental milestones were normal, and there was no difficulty in playing or carrying out day-to-day activities. There was no visual impairment or any comorbidity. There was no family history of similar symptoms, and he was born to non-consanguineous parents. On general examination, his weight was 22 kg and height was 127 cm. Feet were hypertrophied, especially second and third toes were more enlarged in both feet [Figure 1], and there was no tenderness or warmth. Similarly, both hands were enlarged [Figure 1], and there were no functional restriction in both lower and upper limbs. Arterial pulsations were normally felt. Sensation was normal in all the extremities. There were no nodules or pitting edema or café-au-lait spots seen. No audible bruits or thrills were noticed over extremities. There were no other musculoskeletal abnormalities found. Although there was gigantism, no discrepancy in limb length was noticed. Cognition and higher mental function were normal for his age and education. Nervous system and other systems were clinically normal.

Address for correspondence:
Dr. K Gunasekaran,
Department of Neurology,
Government Mohan Kumaramangalam Medical College Hospital, Salem, Tamil Nadu, India.
E-mail: drgunasekaranneuro@gmail.com

How to cite this article: Gunasekaran K, Sundareswaran N, Gopinath G. A child with enlarged extremities – A case of macrodystrophia lipomatosa. Indian J Dermatol 2020;65:409-13.

Received: October, 2018. Accepted: November, 2018.
His investigation results were as follows: basic blood and urine investigations were normal. Plain radiographs showed enlarged metatarsal and phalanges in the second and third toes of both feet with soft tissue hypertrophy [Figure 2]. Similar to that, plain radiograph of hands demonstrated enlarged metacarpal and phalanges in the index finger as well as enlarged phalanges in the middle and ring fingers of both hands with soft tissue hypertrophy [Figure 3]. Non-contrast-enhanced computed tomography (CT) scan of lower limbs showed marked increase in subcutaneous fat seen as abnormal hypodense areas of fat attenuation in both legs and feet [Figure 4]. Similarly, CT scan of hands and forearms showed abundant accumulation of adipose tissue [Figure 5]. Magnetic resonance imaging (MRI) showed prominent subcutaneous fat in the feet and legs [Figure 6], which was completely suppressed in T2 fat sat (short-tau inversion recovery-STIR) sequence. Similarly, prominent subcutaneous fat was noted in the MRI of both hands and forearms [Figure 7], which was suppressed in STIR sequence. There was no narrow signal alteration noted in the phalanges, carpal, metacarpal, or tarsal bones. Histopathological examination of the hypertrophied soft tissue could not be done, as parents were not willing. Doppler study of both lower and upper limbs showed no evidence of arteriovenous fistula or arteriovenous malformation. Ultrasonomogram of abdomen did not show any visceromegaly or tumor mass. MRI brain with screening of spine was performed that showed no significant abnormalities. Pituitary gland and sella appeared normal. With these clinical and radiological features, diagnosis of ML was made out.

Discussion

ML occurs because of the proliferation of all mesenchymal elements of an extremity, including the phalanges, nerves, muscles, and vessels, with a disproportionate increase in fibroadipose tissue\(^1\) and presents as focal or regionalized gigantism without an abnormal tall stature.\(^4\) The degree of overgrowth is faster than the normal growth pattern and this abnormal growth ceases at puberty.\(^3\) This abnormal growth develops in a specific sclerotome region of the body,\(^5\) commonly the lateral aspect of the hand, along the median nerve distribution and the medial aspect of the foot along the medial plantar nerve distribution with the predilection for the second and third digits.\(^6\) Less commonly, an entire upper or lower limb may be involved.\(^2\) The lower extremities are more often involved than the upper limb. However, the report of ML involving all four extremities is not seen in literature.

Although it commonly presents in childhood, the clinical findings can be recognized as early as the

\(\text{References}\)
neonatal period to late adulthood and slight male preponderance is noticed.[2] Feriz, in 1925 coined the term ML.[3] The etiology of ML is not known. Many hypotheses suggesting the cause have been put forth. These include lipomatous degeneration, fetal circulation abnormality, and damage of extremity bud and errors in the segmentation in intrauterine life and hypertrophy of the concerned nerve.[3]

Typical x-ray findings of ML include soft tissue and osseous hypertrophy, presence of radiolucent areas due to the abnormally proliferated adipose tissue, and degenerative joint disease. In addition, there may be elongated, thickened phalanges with splayed distal ends, resembling a “mushroom” shape.[3] Doppler flow studies of the involved limbs show no increased vascularity. In CT scan, characteristic findings noted are osseous hypertrophy as well as excessive growth of soft-tissue containing abnormal hypodense areas of fat attenuation.[1] MRI is the investigation of choice for establishing the

Figure 3: Plain radiograph of hands in anteroposterior plane demonstrates enlarged metacarpal and phalanges of the index finger as well as enlarged phalanges of the middle and ring fingers of both hands with soft tissue hypertrophy

Figure 4: Non-contrast-enhanced CT scan, in the coronal and axial plane, showing abnormal hypodense areas of fat attenuation in both legs

Figure 5: Non-contrast-enhanced CT scan of both hands and forearm, in the coronal plane, showing abnormal hypodense areas of fat attenuation

Figure 6: MRI in coronal view showing prominent subcutaneous fat in the feet and legs. No marrow signal alteration noted in the phalanges, carpal, and metacarpal bones.
diagnosis. MRI easily demonstrates the excess fibrofatty tissue, which has signal characteristics as hyperintensity on T1-weighted and T2-weighted images and complete suppression (hypointensity) on STIR sequence. The fat is not encapsulated, the fibrous strands within the fatty tissue are seen as low-signal-intensity linear strands on T1-weighted images. Neural thickening may also be visualized.

The most prominent histopathological finding noted in ML is the increase in adipose tissue scattered in a fine lattice of fibrous tissue, which involves the bone marrow, periosteum, muscles, nerve sheaths, and subcutaneous tissues. However, histopathologic examination is not routinely needed in the diagnostic process of ML, as MRI shows characteristic findings sufficient to make the diagnosis without histology, though there may be room for the latter in research work.

The differential diagnosis for ML are neurofibromatosis type 1 (plexiform neurofibroma), fibrolipomatous hamartoma, lymphangiomatosis, hemangiomatosis, Klippel-Trenaunay-Weber syndrome, Maffucci syndrome, Ollier disease, and Proteus syndrome. However, most of these conditions generally present as focal gigantism involving single limb. But, our patient showed enlargement of all extremities. Hence, other differentials which result in “pseudoacromegaly” or “acromegaloidism” are excluded. This includes Sotos syndrome, a congenital, genetic overgrowth syndrome in which advanced bone age, acromegalic features, macrocephaly, characteristic facial features, seizures, neonatal hypotonia, neonatal jaundice, scoliosis, congenital heart defects, hypothyroidism, and intellectual impairment are seen. Similar to Sotos syndrome, other rare genetic overgrowth syndromes such as Beckwith-Wiedemann syndrome, Weaver syndrome, and Fragile X syndrome also have to be ruled out. In our patient, the diagnosis was readily established by history and clinical examination, supplemented by radiographic and detailed CT and MRI assessment.

The complications associated with ML are cosmetic and mechanical. Although cosmetic problem is the usual presentation at all ages, mechanical problems are encountered in adolescents and adults because of secondary degenerative joint changes and compression of neurovascular structures causing impairment of function. Especially, osteophyte overgrowth may cause entrapment neuropathies, such as carpal tunnel syndrome, cubital and tarsal tunnel syndromes.

For both mechanical and cosmetic problems, surgical intervention is the treatment of choice, depending upon the symptoms and age of the patient, as well as on the extent and severity of the disease. Surgery in the form of localized amputations or debulking is being done. In focal gigantism with a macrodactyly, removal of a ray is the most appropriate procedure that can lead to a cosmetic improvement and does not cause any functional or neurologic problem. Multiple debulking procedures, epiphysiodesis, and various osteotomies are indicated for more severe forms of the disease. These procedures should be planned and used judiciously. Because, the incidence of nerve injury following extensive debulking or lesion removal is high. In addition, increased recurrence rate is also noted. Moreover, it is to be remembered that progressive hypertrophy associated with ML usually stops at puberty.

We decided to manage our patient conservatively after discussion with plastic surgeon because, in spite of the cosmetic issue, there were no functional impairment. Moreover, he was in prepubertal stage. In addition, we planned to monitor the child periodically to ensure mobility and functionality. Should these be compromised or the disfigurement become excessive, the plastic surgeon would intervene.

**Conclusion**

Gross disfigurement and functional disability may complicate ML. The surgical management consists of improving cosmetic appearance and preserving mobility. The outcome of surgery may not be very gratifying in extensive and severe form of the disease. This case is presented here because of the rare presentation (involvement of all four extremities) of this rare condition.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient’s parents have given his/her/them consent for his/her/their images and other clinical information to be reported in the journal. The patient’s parents...
understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

**Financial support and sponsorship**
Nil.

**Conflicts of interest**
There are no conflicts of interest.

**References**

1. Sharma R, Gupta P, Mahajan M, Arora M, Gupta A. X-ray and computed tomography findings in macrodystrophia lipomatosa of the foot with secondary osteoarthritic changes diagnosed in an elderly female: A case report. Radiol Bras 2017;50:132-4.

2. Abdulhady H, El-Sobky TA, Elsayed NS, Sakr HM. Clinical and imaging features of pedal macrodystrophia lipomatosa in two children with differential diagnosis review. J Musculoskelet Surg Res 2018;2:130-4.

3. Khan RA, Wahab S, Ahmed J, Chana RS. Macrodystrophia lipomatosa: Four case reports. J Pediatr 2010;36:69.

4. Kwon JH, Lim SY, Lim HS. Macrodystrophia lipomatosa. Arch Plast Surg 2013;40:270-2.

5. Jain R, Sawhney S, Berry M. CT diagnosis of macrodystrophia lipomatosa. A case report. Acta Radiol 1992;33:554-5.

6. 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;69:502-7.

7. Dhanasekaran J, Reddy AK, Sarawagi R, Lakshmanan PM. Imaging features of macrodystrophia lipomatosa: An unusual cause of a brawny arm. BMJ Case Rep 2014. 2014 pii: Bcr2014204899. doi: 10.1136/bcr-2014-204899.

8. Soler R, Rodriguez E, Bargiela A, Martijnez C. MR findings of macrodystrophia lipomatosa. Clin Imaging 1997;21:135-7.

9. Dahlqvist P, Spencer R, Marques P, Dang MN, Glad CAM, Johannsson G, et al. Pseudoacromegaly: A differential diagnostic problem for acromegaly with a genetic solution. J Endocr Soc 2017;1:1104-9.

10. Baujat G, Cormier-Daire V. Sotos syndrome. Orphanet J Rare Dis 2007;2:36.

11. Kozanoglu E, Koc F, Goncu K. Macrodystrophia Lipomatosa with Multiple Entrapment Neuropathies: A Case Report. Int J Neurosci 2008;118:545-53.