

Macrodystrophia Lipomatosa: Clinico-patho-radiological Correlation

Abstract

Macrodystrophia lipomatosa is a rare congenital nonhereditary developmental anomaly. It is characterized by hamartomatous proliferation of the soft tissue leading to disproportionate enlargement of the limbs and digits. Since it leads to diagnostic dilemma, it has to be differentiated from various other conditions as they differ in course, prognosis, complications, and treatment. Herein, we present two cases with localized gigantism and discuss the various differential diagnoses and need for clinico-patho-radiological correlation for diagnosis of this rare entity.

Keywords: *Benign lipomatous lesion, hamartoma, macrodactyly*

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Introduction

Macrodystrophia lipomatosa (ML) is a rare form of nonhereditary congenital macrodactyly characterized by progressive overgrowth of mesenchymal elements with a disproportionate increase in the amount of fibroadipose tissue. This abnormality occurs most frequently along the median nerve distribution in the upper extremities and the plantar nerve in the lower extremities.^[1-3] Usually, one or more digits of the unilateral limb are affected. There is no gender predilection; however, males are more affected than females.^[1] Due to rarity, the incidence is unknown. Earlier, a variety of descriptive terms were used for this condition, especially in the pediatric age group such as macrodactyly, megalodactyly, digital gigantism, macromelia, partial acromegaly, macrosomy, and limited gigantism. ML leads to diagnostic dilemma and has to be differentiated from various other conditions as they differ in course, prognosis, complications, and treatment. The patient outcome is dependent on the severity of the disease. In the present cases, cytology in conjunction with radio imaging aided in reaching the diagnosis, followed by subsequent corrective surgical management.

Case Reports

Case 1

A 3-year-old female child presented with a progressive disproportionate enlargement of the second and third toes of the right foot

since birth [Figure 1]. There were no history of any pain or neurovascular symptoms and also no family history of extremity gigantism. The developmental milestones were according to the age. On physical examination, there was nontender enlargement of the second and third toes of the right foot. No overlying cutaneous changes, pitting edema, or bruit was seen. All the other extremities were found to be normal on examination. A plain X-ray of the right foot was performed that revealed soft-tissue swelling involving the second and third toes with hypertrophy of the phalanges [Figure 2]. Gray-scale ultrasound and color flow Doppler revealed diffuse soft-tissue thickening, without any abnormal calcifications or abnormal blood flow. Fine-needle aspiration from the second digit yielded blood-tinged fatty aspirate. Smears examined revealed fragments of mature adipose tissue [Figure 3]. No malignant cells were identified. The background revealed fat droplets admixed with red blood cells (RBCs) and few inflammatory cells. Correlating the lesion's clinical presentation, imaging and cytological finding possibility of benign mesenchymal lesion of lipomatous origin was considered with a differential diagnosis of ML and fibrolipomatous hamartoma. Debulking surgery was performed. Histopathological examination of the sent material showed mature fibroadipose tissue involving the subcutaneous plane. The nerve sheath and the surrounding muscle fibers were free of fatty infiltration. Thus, a definitive diagnosis of ML was rendered which confirmed the initial cytological possibilities.

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How to cite this article: Mehra R, Bhartiya R, Agrawal P, Kumari N. Macrodystrophia lipomatosa: Clinico-patho-radiological correlation. *Indian J Med Paediatr Oncol* 2017;38:559-62.

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Access this article online

Website: www.ijmpo.org

DOI: 10.4103/ijmpo.ijmpo_65_16

Quick Response Code:



Case 2

A 21-year-old right hand dominated female presented with a progressive disproportionate enlargement of distal

arm and digits of the right hand since birth. The patient denied having any pain or neurovascular symptoms, and there was no family history of extremity gigantism. Twelve years back, she had a history of amputation of



Figure 1: Clinical image showing enlargement of the second and third toes of the right foot



Figure 2: X-ray image showing hypertrophy of soft tissues of the 2nd and 3rd digit of the right foot

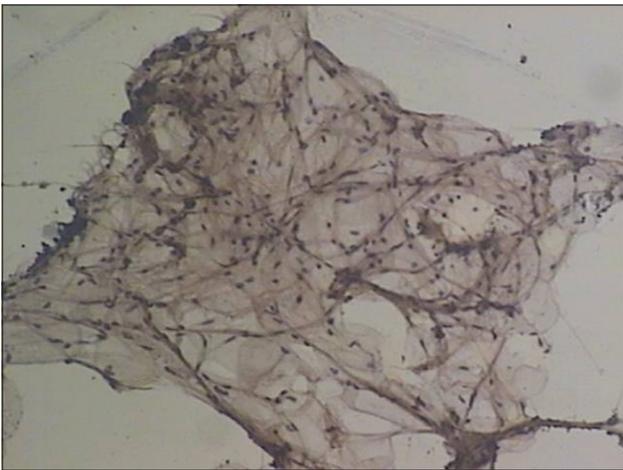


Figure 3: Cytosmears reveal mature adipocytes (Pap, x100)



Figure 4: Clinical image showing soft-tissue mass of the enlarged limb with disfigurement of digits and distal arm of the right upper limb

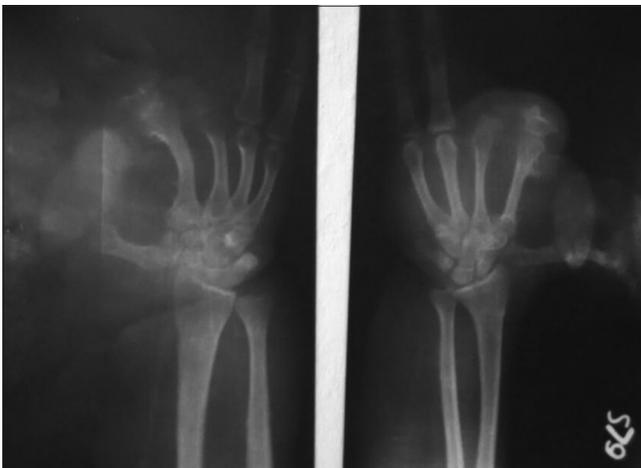


Figure 5: X-ray image showing bulbous enlargement of the thumb of the right hand

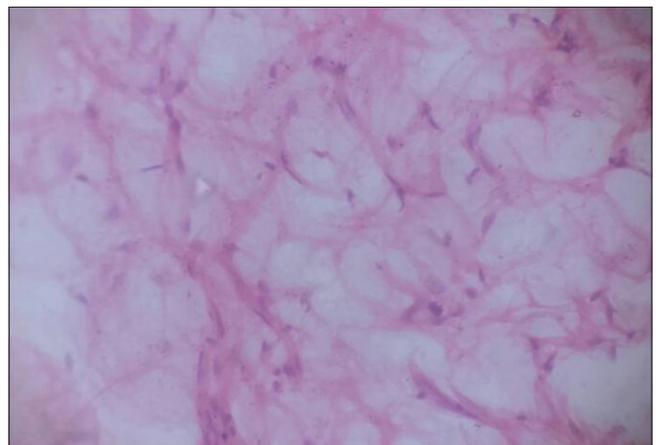


Figure 6: Section shows abundant adipose tissue (H and E, x400)

index and middle fingers at some periphery hospital. However, no records of the same were available. On physical examination, a nontender, soft-tissue mass was palpable on the volar aspect of the enlarged thumb with disfigurement of digits and distal arm [Figure 4]. There were no overlying cutaneous changes, pitting edema, or bruit. A plain radiograph demonstrated soft-tissue swelling along the volar aspect of the thumb, enlarged phalanges of the digits [Figure 5]. Gray-scale ultrasound and color flow Doppler revealed diffuse soft-tissue thickening, without any abnormal calcifications or abnormal blood flow. Fine-needle aspiration from the disproportionate areas yielded blood-tinged fatty aspirate. Smears examined revealed fragments of mature adipose tissue. No malignant cells were identified. No other spindle cell component was identified. The background revealed fat droplets admixed with RBCs and few inflammatory cells. Correlating the lesion's clinical presentation, imaging and cytological finding possibility ML was considered. The multidisciplinary team decided to perform a debulking surgery. On gross examination a yellowish soft-tissue mass was received which on cut section was fibrofatty. No firm areas or hemorrhage or necrosis was identified. Microscopic examination revealed mature fibroadipose tissue involving the subcutaneous plane [Figure 6]. The nerve sheath and the surrounding muscle fibers were free of fatty infiltration. Thus, a definitive diagnosis of ML was rendered which confirmed the initial possibility.

Discussion

ML is an uncommon congenital, nonhereditary developmental anomaly generally presenting hamartomatous proliferation of the soft tissue of the affected area also called as localized gigantism.^[1] This disease is characterized by the proliferation of all the mesenchymal elements of a digit or digits and a disproportionate increase of fibroadipose tissue, involving the nerve sheath, muscle, periosteum, and bone marrow.^[2,3]

Feriz^[4] in 1925 first used the term ML. Barsky^[5] divided ML into two types, including the static and progressive forms. In the static form, the growth rate of affected tissues is normal. In the progressive form, the growth rate of mesenchymal tissues is faster when compared with normal tissues and ceases its abnormal growth at puberty. Progressive macrodactyly is more common than static type. Exact etiology is unknown, but various hypotheses have been proposed regarding the etiopathogenesis of ML. These include lipomatous degeneration, fetal circulation abnormality, and damage of extremity bud and alteration of somatic cells in intrauterine life and hypertrophy of the concerned nerve.^[6] It may be associated with various abnormalities such as syndactyly, polydactyly, brachydactyly, or clinodactyly. The various differential diagnoses are discussed in Table 1.^[1,2,7-10]

The problems associated with a patient of ML can be cosmetic or mechanical. The overgrowth may lead to

Table 1: Differential diagnosis of macrodactyly

Differential diagnosis	Clinical and imaging	Pathological findings
Macrodystrophia lipomatosa	Affects only plantar and median nerve, localized gigantism with disproportionate increase in length and width of phalanges, metatarsus, and surrounding soft tissue	Increase in adipose tissue scattered fibrous tissue may involve the bone marrow, periosteum, muscles, nerve sheaths, and subcutaneous tissues
Fibrolipomatous hamartoma	Mostly affects median nerve, digital overgrowth, paresthesia, motor deficit, and pain along the affected nerve, speckled appearance on MR	Perineural and endoneurial fibrosis, infiltration of interfascicular connective tissue by mature fat cells
Lymphangiomatosis	Diffuse swelling, pitting edema, chylous effusion, lytic bone lesion, hyperintense to muscle on T1W and hypointense to fat on T2W, no osseous growth	Increase in both the size and number of thin walled lymphatic channels along with lymphatic spaces
Hemangiomatosis	Large and multiple bluish lesions, bruits, T2-weighted spin-echo imaging shows worm-like areas of high signal intensity, no osseous growth	Multiple vascular channels lined with a single layer of endothelium supported by fibrous tissue
Klippel-Trenaunay-Weber syndrome	Port-wine stain, osseous and soft-tissue hypertrophy, venous malformation, varicose veins	Irregular venous-type channels, lined by flat endothelium and surrounded by smooth muscle
Maffucci syndrome	Enchondroma, subcutaneous hemangioendothelioma, expansile lytic lesion in phalanges	Benign cartilaginous matrix, myxoid changes
Ollier disease	Unilateral multiple enchondromas, expansile lytic lesion in phalanges	Benign cartilaginous matrix, myxoid changes
Proteus syndrome	Hemihypertrophy with asymmetrical partial gigantism, skull abnormalities, pigmented nevi, lung cysts, intraabdominal lipomas, calvarial changes	Asymmetric overgrowth of skin, soft tissue, bone, vascular malformation, adipose tissue, hypertrophy
Neurofibromatosis	Positive family history, bilateral involvement, café au lait spots, Lisch nodules, freckling in the axilla, hyperintensity on T2W images close to nerves	Nonencapsulated, interlacing bundles of spindle cells with wavy nuclei, no adipose tissue component

MR – Magnetic resonance; T1W – T1-weighted; T2W – T2-weighted

interference in normal day-to-day activities or make patient prone to repeated injury. Mechanical problems may develop such as secondary osteoarthritis and compression of neurovascular structures causing impairment of function. Cosmetic problem is usual, presenting complaint in all ages, but mechanical problems are encountered in adolescence due to secondary degenerative joint changes causing reduced function. Electroneurography and nerve conduction velocity tests performed in these patients may reveal slowed distal motor and sensory conduction, segmental conduction block, or slowing of the peripheral nerves at the entrapment sites.

Different imaging modalities such as X-ray, ultrasonography, computed tomography (CT) scan, and magnetic resonance imaging (MRI) have a role in the evaluation of ML. Typical X-ray findings of ML include hypertrophy of soft tissue as well as osseous tissue, with translucencies in the soft tissue due to increased adipose tissue. The phalanges are elongated, broad, and splayed, sometimes giving rise to a mushroom-like appearance. Secondary osteoarthritic changes such as joint space narrowing, subchondral cysts, and osteophytes may also be seen. Excessive growth of the bone within the area innervated by nerve and fat tissue proliferation within muscle fibers are the characteristic findings detected on CT scan. MRI scans reveal the presence of abundant adipose tissue, which exhibits the same signal intensity as normal subcutaneous fat in the areas affected by the disease. There may be linear hypointense fibrous bands noted within this abnormal fat on T1-weighted imaging. There are osseous hypertrophy and cortical thickening in the affected part of the body, and this may lead to exostoses such as bony outgrowths from the involved bone.^[8-10]

The treatment is usually surgical, which is carried out due to cosmetic reasons, but since the digital enlargement stops at puberty, management should include an attempt to preserve neurological functions. For lesion involving median and plantar region, conservative approach has been advocated. Multiple debulking procedures, epiphysiodesis, and various osteotomies are indicated for more severe form

of the disease. Reconstructive surgery, which achieves a more functional result, should be preferred strategy.^[7]

ML is progressive hamartomatous enlargement of the fibrofatty tissue involving all the layers of soft tissue and even bone more commonly leading to cosmetic and mechanical problems. Diagnosis is accomplished on the basis of clinical and radiological evaluation which can be confirmed on histopathological examination. Awareness of the characteristic symptoms, imaging, pathology, and differential diagnosis of the disease is very important for the treatment of the rare entity.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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