A series of three cases of macrodystrophia lipomatosa: Rare cause of focal limb gigantism

Chiranjeev Kumar Gathwal, Kulvinder Singh, Saru Singh, Monika B. Gathwal, Santosh Munde, Gaurav Malik, Vikas Verma

ABSTRACT

Introduction: Macrodystrophia lipomatosa (MDL) is defined as rare congenital entity characterized by proliferation of mesenchymal elements predominantly the fatty component resulting in localized or regional enlargement of part or whole limb. It usually presents with cosmetic problems and variable functional deformity. Even though history and physical examination are usually diagnostic, radiological evaluation especially the cross-sectional modalities can reliably distinguish MDL from other forms of localized congenital gigantism.

Case Series: Here we are presenting a series of three cases of macrodystrophia lipomatosa (MDL) presented to radiology department with history of disproportionate focal overgrowth of limb.

Conclusion: Macrodystrophia lipomatosa is a rare form of congenital localized gigantism. As there are numerous etiologies of focal gigantism and clinical distinction may be difficult at times, radiological evaluation is very useful in confining the differential diagnosis. Imaging, with magnetic resonance imaging (MRI) in particular, provides vital clues to diagnose the condition which is confirmed by histopathology. Though surgical consultation is often offered for cosmetic reasons, it should be delayed till puberty or when functional problems exist as surgical results are not very rewarding with significant recurrence rate.
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Keywords: Fatty overgrowth, Macrodactyly, Macrodystrophia lipomatosa, Soft tissue overgrowth

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INTRODUCTION

Macrodystrophia lipomatosa (MDL) is rare congenital entity characterized by proliferation of mesenchymal elements predominantly the fatty component resulting in localized or regional limb overgrowth. This disease is
characterized by localized gigantism due to overgrowth of all mesenchymal elements predominantly the fibrofatty tissue and should be differentiated from macromelia or hemihypertrophy [1]. The term macrodystrophy lipomatosa was first coined by Feriz in 1925 for describing lower extremity gigantism cases, which were associated with massive overgrowth of the accompanying adipose tissue. Barsky later gave a more detailed description of the focal MDL causing local gigantism of digits [2]. Here we are presenting a series of three cases of macrodystrophia lipomatosa.

**CASE SERIES**

**Case 1**

An 18-year-old female was referred from orthopedic department with a history of progressive enlargement of middle and ring fingers of right hand since childhood and progressive decrease in movements since two years. Her parents initially noticed the abnormally enlarged fingers at age of 2–3 years which increased significantly over the years asymptomatically. However, later on she developed progressive pain and variable loss of motion for last two years. She was also operated for this condition for cosmetic reasons by local surgeon. However, details were not available with the patient. At presentation she had enlarged phalanges, pain and inability to completely flex the fingers. There was no evidence of pitting edema, bruit or other sensory changes. She also had kyphosis-scoliotic deformity of spine which was noticed at 7–8 years with significant progression over the years. She had taken treatment with frequent physiotherapy sessions. However, no significant improvement occurred. There is no history of past operative treatment for kyphosis-scoliotic deformity. There was no family history of similar illnesses.

The initial physical examination revealed the presence of local gigantism of middle and ring fingers of right hand as seen in volume rendered technique (VRT) image (Figure 1A). Variable restriction of movements of involved fingers at presentation was also noticed. There were no overlying skin changes or cutaneous stigmata seen. There was no neurological deficit. The other digits and contralateral foot were normal. Kyphosis-scoliotic deformity was also seen at the time of examination. Plain radiographs (Figure 1B) showed enlargement of 3rd and 4th fingers with overgrowth of soft tissue as well as bony metacarpals and phalanges. There was increased lucency in soft tissue compartment of affected fingers suggestive of fat. Secondary degenerative changes as joint space reduction, marginal osteophytes and variable partial ankylosis were also noticed at metacarpophalangeal and interphalangeal joints. Plain radiographs (Figure 1C) reveal significant kyphosis-scoliotic deformity with right sided convexity centered at D12 vertebra. Vertebral anomalies can also be appreciated in dorsolumbar spine. Ultrasonography and color Doppler using high-resolution probe revealed abundant increase in the adipose tissue without any increase vascularity or any vascular malformation. Non contrast computed tomography (NCCT) hand (Figure 1D–E) revealed overgrowth of bony as well as soft tissue components of affected fingers. Metacarpals and phalanges showed growth in length and width with significant disproportionately overgrowth of fat in increased soft tissue compartment. Degenerative changes with partial ankylosis at metacarpophalangeal and interphalangeal joints can also be appreciated. No other imaging of spine was available at time of presentation. The patient underwent surgery of fingers for cosmetic reasons. Histopathology confirmed the presence of significantly excess adipose tissue in the excised specimen scattered within thin mesh-like fibrous tissue extending up to periosteal surface. A complete diagnosis of recurrent macrodystrophia lipomatosa affecting middle and index fingers of right hand was made.

**Case 2**

A nine-year-old female presented to radiology department for radiographic evaluation with a long history of progressive disproportionate enlargement of the medial two toes of the left foot. The overgrowth of toes was noticed in childhood which increased progressively with age. Initially it was asymptomatic but later on she developed difficulty in wearing slippers, walking with recurrent injury to the foot. On physical examination, 1st and 2nd toes of left foot were disproportionately enlarged with excessive soft tissue component on the plantar aspect. There was also widening of the web space between 1st and 2nd toes. There were no overlying cutaneous changes, pitting edema or bruit. Rest of the toes of left foot as well as right foot appeared normal. Plain radiography (Figure 2) of the left foot revealed disproportionately enlarged 1st and 2nd toes involving both bony and soft tissue components. There was increase in length and diameter of the involved phalanges with preserved cortex and trabecular pattern. There is also splaying of bony phalangeal ends with dorsal angulation; involving the 2nd toe more markedly. The overgrown soft tissue component showed increased lucency suggesting increased adipose tissue. Biopsy revealed encapsulated lobules of well differentiated fatty tissue extending up to periostium. Based on typical clinical history, characteristic radiographic findings and histopathology, diagnosis of macrodystrophia lipomatosa was made.

**Case 3**

A three-year-old girl was referred to our department for magnetic resonance imaging (MRI) evaluation of local gigantism of the right foot. Parents gave history of enlargement of right foot since birth, particularly the 2nd and 3rd toes for which the patient was operated at one and a half years of age in some remote hospital. There
was some cosmetic improvement after the operation. However, operative records were not available at time of presentation. She had been relatively asymptomatic with only mild pain over her sole from plantar keratosis. However, in last six months parents noticed further accelerated overgrowth of her right foot including the adjacent great toe. Physical examination revealed diffuse enlargement of her right foot with exceptionally large first and third toes. Also painless non-compressible soft tissue overgrowths were palpable at the lateral aspect of ankle and plantar aspect of foot. No dilated superficial vein or vascular mark was noticed. Contralateral foot was normal. No other skeletal abnormality was found. Previously, there was plain CT scan done from outside with the patient. Plain CT scan revealed significantly overgrown right foot with overgrowth of both bony and soft tissue compartments. The overgrown soft tissue compartment was predominantly lucent suggestive of fatty nature. Both CT scan and reconstructed 3D bony images revealed enlarged and thickened 1st, 2nd and 3rd metatarsals and residual phalanges with widening of inter-digital gap. Postoperative changes can be appreciated in 2nd and 3rd toes. Magnetic resonance imaging scan of the right lower leg, ankle and foot was performed. There was increased deposition of subcutaneous fat at the lateral and posterior aspects of the ankle, posterior and plantar aspects of the heel, plantar surface of foot with disproportionately enlarged first and residual third toes. The increased fatty overgrowth had the same signal intensity as rest of the normally visualized subcutaneous fat. Streaky T1-hypointense and T2-hypointense signal intensities suggestive of fibrous strands could be observed inside the overgrown subcutaneous fat. The right tibial nerve distal part was fusiform enlarged with abundant lipomatous tissue seen as T1 hyperintense component with suppression on FatSat sequences interspersed among the thickened nerve fascicles (Figure 3).

The imaging findings were consistent with macrodystrophia lipomatosa and which was further confirmed on biopsy. Based on clinical history, radiographic evaluation and histopathological examination diagnosis of recurrent macrodystrophia lipomatosa was made.

DISCUSSION

Macrodystrophia lipomatosa (MDL) is rare congenital entity characterized by proliferation of mesenchymal elements predominantly the fatty component resulting in localized or regional enlargement of part or whole limb. This abnormality occurs most frequently in the distribution of median and plantar nerves in upper and lower extremities respectively. Usually, one or more digits of the unilateral limb are affected; there have been few reports of involvement of entire limb, bilateral limbs and abdominal wall as well [3, 4]. In most reported cases to date, the lesions are present at birth or develop after the operation. However, operative records were not available at time of presentation. She had been relatively asymptomatic with only mild pain over her sole from plantar keratosis. However, in last six months parents noticed further accelerated overgrowth of her right foot including the adjacent great toe. Physical examination revealed diffuse enlargement of her right foot with exceptionally large first and third toes. Also painless non-compressible soft tissue overgrowths were palpable at the lateral aspect of ankle and plantar aspect of foot. No dilated superficial vein or vascular mark was noticed. Contralateral foot was normal. No other skeletal abnormality was found. Previously, there was plain CT scan done from outside with the patient. Plain CT scan revealed significantly overgrown right foot with overgrowth of both bony and soft tissue compartments. The overgrown soft tissue compartment was predominantly lucent suggestive of fatty nature. Both CT scan and reconstructed 3D bony images revealed enlarged and thickened 1st, 2nd and 3rd metatarsals and residual phalanges with widening of inter-digital gap. Postoperative changes can be appreciated in 2nd and 3rd toes. Magnetic resonance imaging scan of the right lower leg, ankle and foot was performed. There was increased deposition of subcutaneous fat at the lateral and posterior aspects of the ankle, posterior and plantar aspects of the heel, plantar surface of foot with disproportionately enlarged first and residual third toes. The increased fatty overgrowth had the same signal intensity as rest of the normally visualized subcutaneous fat. Streaky T1-hypointense and T2-hypointense signal intensities suggestive of fibrous strands could be observed inside the overgrown subcutaneous fat. The right tibial nerve distal part was fusiform enlarged with abundant lipomatous tissue seen as T1 hyperintense component with suppression on FatSat sequences interspersed among the thickened nerve fascicles (Figure 3).

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within the first weeks of life, but generally only starts to cause problems as the child grows. Though Feriz in 1925 coined the term macrodystrophia lipomatosa, Barsky in 1967 described two forms of macrodactyly, static as enlargement of digits or limb in proportion of rest of the body and progressive form as disproportionate overgrowth of digits or limb and is usually associated with fatty overgrowth [5].

Exact etiopathogenesis of MDL being not well understood, the possible mechanisms given in literature include lipomatous degeneration, disturbed fetal circulation, disturbances of growth inhibiting factors, errors in segmentation, and trophic influence of tumefied nerve. The pathogenesis of bony enlargement is because of endosteal and periosteal deposition of bone [6].

The symptomatic problems associated with macrodystrophia lipomatosa are cosmetic and mechanical. Cosmetic problem is the usual presenting complaint in all ages but mechanical problems are encountered in adolescence due to secondary degenerative joint changes causing restricted movements and functions. Since overgrowth mainly involves the volar and plantar aspects, it can produce dorsal deviation of affected parts. This may lead to interference in normal day to day activities or make patient prone to repeated trauma. Osteophyte overgrowth can sometimes cause compression of adjacent nerves and vessels, most commonly seen in carpal tunnel syndrome [7, 8]. Other associations include lipomatous growths in intestines and other tissues, calvarial abnormalities, pigmented nevus, pulmonary cysts, syndactyly, polydactyly, clinodactyly, brachydactyly and symphalangism [9].

Imaging studies play an important role in characterization of nature of the hypertrophied tissue. Typical radiographic findings of macrodystrophia lipomatosa include excessive growth of soft tissue as well as osseous tissue. The presence of predominant radiolucent areas in overgrown soft tissue compartment suggests fatty nature of the soft tissue. Usually the volar aspect of the fingers is disproportionately involved [10, 11]. There is osseous hypertrophy and cortical thickening in the affected part of the body and this may lead to exostoses like bony outgrowths from the involved bone. Widening at the distal end of the bones gives the characteristic mushroom-like appearance [10, 11]. Ultrasonography and Doppler show large amounts of subcutaneous tissue, infiltration of the muscle and thickening of the affected nerves with absence of any increased vascularity. Excessive growth of the bone and fatty tissue proliferation are well appreciated findings detected on CT scan. The volar and plantar aspects of the fingers are disproportionately involved [10, 11].

The excessive fat seen in macrodystrophia lipomatosa is not encapsulated and MRI scan can easily demonstrate the fatty infiltration of the muscles. There may be linear T1/ T2 hypointense fibrous bands noted within this abnormal fat. The fibro-adipose tissue appears hyperintense on both T1-weighted and T2-weighted MRI images, and is
identical to that of normal subcutaneous fat tissue. Soler et al. [10] proposed that MRI scan should be used as the diagnostic method of imaging for macrodystrophia lipomatosa to detect excess fibro-adipose tissue and enlargement of other mesenchymal tissues.

Histopathological findings show an abundant increase in adipose tissue scattered fibrous tissue [11]. Underlying subcutaneous tissues, nerve sheaths, muscles, periosteum, and even bone marrow involvement can also be seen. Differentials include neurofibromatosis type I, fibrolipomatosis hamartoma of nerve with macrodactyly, hemangiomaticosis lymphangiomatosis, Klippel–Trenaunay–Weber syndrome and Proteus syndrome. Neurofibromatosis often has positive family history and certain characteristic cutaneous manifestations. Fibrolipomatosis of the nerve can also occur in isolation or with associated localized gigantism. Associated macrodactyly are seen in two-thirds cases and are usually difficult to distinguish from MDL.

It presents with typical nerve lesion associated with intramuscular fat deposition. In MDL, abnormal fat deposits are not limited within the nerve sheaths and muscles but also involve the subcutaneous tissues and tendons. Also there is periosteal involvement leading to the bony changes such as hypertrophy, exostoses, ankylosis of interphalangeal joints and fatty invasion of the medullary cavity which is quite specific for macrodystrophia lipomatosa [12].

Surgical intervention is the treatment of choice for macrodystrophia lipomatosa mainly to improve the cosmetic appearance while preserving the neurologic function as much as possible. Judicious and planned use of multiple debulking procedures, epiphysiodesis and various osteotomies are advisable to achieve the best results [13]. Surgery should be delayed till completion of growth if the deformity is not very serious and if no nervous system symptoms are present as there is localized recurrence rate of 33–60% after surgery [14].

**CONCLUSION**

To conclude, macrodystrophia lipomatosa is a rare form of congenital localized gigantism. As there are numerous etiologies of focal gigantism and clinical distinction may be difficult at times, radiological evaluation is very useful in confining the differential diagnosis. Imaging, with magnetic resonance imaging in particular, provides vital clues to diagnose the condition which is confirmed by histopathology. Though surgical consultation is often offered for cosmetic reasons, it should be delayed till puberty or when functional problems exist as surgical results are not very rewarding with significant recurrence rate.

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