Case Study

MACRODYSTROPHIA LIPOMATOSA OF THE HAND

Rai Santosh¹*, Chakraborti Shrijeet²

1. Department of Radiodiagnosis, KMC Mangalore (Manipal Univeristy) KMC Hospital, Attavar, Mangalore 575001, Karnataka, India

2. Department of Pathology, KMC Mangalore (Manipal Univeristy)

ABSTRACT

Macrodystrophy Lipomatosa is an unusual form of localized gigantism. Lower limb is more frequently involved than upper limbs. Here we report a case of Macrodystrophy Lipomatosa of the hand. We describe the characteristic radiographic and MRI findings that distinguish the abnormality from other causes of localized gigantism. MR images confirmed excess fatty proliferation and fatty infiltration of the involved muscles. MR angiography did not show any significant vascular supply to the lesions. These findings were sufficed for giving a radiological diagnosis of Macrodystrophy Lipomatosa. After cosmetic surgery, histopathology of specimen showed massive increase in adipose tissue, interspersed in a fine mesh of fibrous tissue associated with fibrolipomatous hamartoma of a plantar nerve. Radiological findings can be used to confidently diagnose and differentiate this condition from other conditions causing localized gigantism. This differentiation is significant since the various conditions causing localized gigantism differ in their course, prognosis, complications and treatment.

Keywords: gigantism, macrodystrophia lipomatosa, MR imaging.

INTRODUCTION:

Macrodystrophy Lipomatosa is an unusual form of localized gigantism ¹. The etiopathology is obscure. It is classified as a development anomaly. There is marked increase in all mesenchymal elements particularly fibroadipose tissue. Sometimes a nerve can be entrapped within the adipose tissue ². Its involvement is usually unilateral but bilateralism is not rare. Lower limb is more frequently involved than upper limbs. Soft tissue overgrowth is usually more marked over the volar / palmar aspects of the digit. The growth of the digits usually cease at puberty ³. Patient seeks surgical correction for cosmetic reasons. Secondary degenerative changes occur later in adolescence or middle age. Here we report a case of Macrodystrophy Lipomatosa of the hand which came for Magnetic Resonance Imaging (MRI) and was worked up academically and confirmed histopathologically and per-operatively. We describe the characteristic radiographic and MRI findings that distinguish the abnormality from other causes of localized gigantism. The patient was informed that details of the case would be used for academic publication.

CASE HISTORY:

A young lady aged 29 years presented with painless swelling the thumb and index finger of her right hand (Fig 1). The fingers were slightly enlarged since birth and had continued to grow since then. There was no prior history of trauma. On examination, the
fingers were found to be enlarged. Skin over the fingers was thickened and non tender. There was no bruit felt. Consistency was non-uniform being firm and soft in places. There was no family history of similar lesions.

X-ray of the hand showed increase in size of the fingers described with soft tissue overgrowth (Fig 2). There were also degenerative changes noted in the interphalangeal joints of the fingers involved. There were no phleboliths noted on the radiograph. Color Doppler did not show any vascular lakes in this area. MRI and MR angiography of the hand was performed. MRI (0.2 T GE Signa Profile) showed increase in the amount of soft tissue including fat and few vascular elements around the distal phalanx of thumb finger and proximal, middle and distal phalanx of the index finger on the palmar and lateral aspects (Fig 3). MR images confirmed excess fatty proliferation and fatty infiltration of the involved muscles (Fig 4). MR angiography did not show any significant vascular supply to the lesions. These findings were suffice for giving a radiological diagnosis of Macrodystrophia Lipomatosa. The per-operative specimen showed glistening fat elements surrounding a neural structure (Fig 5). After cosmetic surgery, histopathology of specimen showed massive increase in adipose tissue, interspersed in a fine mesh of fibrous tissue associated with fibrolipomatous hamartoma of a plantar nerve (Fig 6). These findings were consistent with Macrodystrophia Lipomatosa.

DISCUSSION:

Macrodystrophia lipomatosa is a rare disorder characterized by three-dimensional enlargement of one or more fingers or toes with predominantly fibroadipose tissue. Radiographically, it appears as hypertrophy of soft tissues and bones. MR images show hypertrophy of fatty tissue, cortical bone thickening and fatty infiltration of muscle. The pathologic findings are infiltration and hypertrophy of adipose tissue in subcutaneous tissue, nerve sheaths, and periosteum. Radiological differential diagnosis of macrodystrophia includes both congenital and acquired disorders. Congential anomalies include conditions like haemangioma, hyperostotic macrodactyly, lymphangioma, Olliers disease, Klippel-weber-Trenaunay syndrome, plexiform neurofibromatosis, melorrheostosis. Radiologists play a key role in the final diagnosis using the various modalities of Radiography, Ultrasound, Computed Tomography and MRI. Radiological findings can be used to confidently diagnose and differentiate this condition from other conditions causing localized gigantism. This differentiation is significant since the various conditions causing localized gigantism differ in their course, prognosis, complications and treatment.

**Key Messages:** Macrodystrophia Lipomatosa is an unusual form of localized gigantism. Lower limb is more frequently involved than upper limbs. Here we report a case of Macrodystrophia Lipomatosa of the hand. MR images confirmed excess fatty proliferation and fatty infiltration of the involved muscles. After cosmetic surgery, histopathology of specimen showed massive increase in adipose tissue, interspersed in a fine mesh of fibrous tissue associated with fibrolipomatous hamartoma of a plantar nerve.
Figure 1

(a) Swelling the thumb and index finger of her right hand and insert (Radiograph of the hand showed increase in size of the fingers described with soft tissue overgrowth. Degenerative changes noted in the interphalangeal joints of the fingers involved)

(b) Coronal T1 weighted MR image reveals increased fatty tissue in the involved fingers.

(c) Coronal Fat suppressed image confirms the predominant fatty tissue in the enlarged first finger.

(d) Histopathology of specimen showed massive increase in adipose tissue, mature adipocytes in lobules with interspersed blood vessels, collagen fibres, nerve bundle (arrowhead) and Pacinian corpuscle (asterix).
REFERENCES:


