

Radiological profile of a neglected case of Macrodystrophia Lipomatosa of the foot

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Citation

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Abstract

Macrodystrophia Lipomatosa is a rare disorder characterized by three-dimensional enlargement of one or more fingers or toes with predominantly fibro adipose tissue. Macrodystrophia Lipomatosa is usually diagnosed during childhood. In this case, we will discuss the radiological findings in the neglected case of a 45 year female with macrodystrophia lipomatosis.

INTRODUCTION

Macrodystrophia Lipomatosa is a rare form of localized gigantism characterized by progressive overgrowth of all the mesenchymal elements with a disproportionate increase in the fibroadipose tissue. This congenital abnormality occurs most frequently in the distribution of the median nerve in the upper extremity and in the distribution of the planter nerves in the lower extremity. Pathologically an increase in fibroadipose tissue involving subcutaneous tissue, periosteum, muscles, nerve sheath and bone marrow is present [1].

CASE REPORT

A 45 year-old female presented with enlargement of her right foot involving the first to third toes since birth. There was history of surgery in childhood at some private clinic but no records were available and no diagnosis was made. Since then soft tissue swelling is gradually increasing and the shape of foot is grossly disorganized. Over the last eight years the joints of the foot had become more painful and had reduced movement. On examination, there was gross soft tissue swelling of right foot more so along the dorsal aspect with absent second toe and gross enlargement and dorsiflexion of the third toe. There was evidence of friction blisters in the overlying skin in the area of dorsum of foot (Figure-1). Laboratory profile was unremarkable.

{image:1}

Plain radiographs of the right foot revealed disorganization of metatarsals & phalanges mainly along second and third toes. Phalanges of second fingers were removed at earlier

surgery. There was dorsal and radial deformity at the third metatarsophalangeal joint. Soft tissue swelling with radiolucency, lobulations representing overgrowth of fatty tissue, is seen at places. There was also evidence of irregular bony outgrowths mainly around head of second metatarsal (Figure-2).

{image:2}

The patient was further investigated & noncontrast computed tomography (NCCT) was performed. On NCCT, there was gross soft tissue swelling with all foot tissues infiltrated by excessive abnormal hypodense fat with CT number varying from -70 to -90. There was disorganized new bone formation in the foot around the metatarsals & phalanges. Bases of first, second and third metatarsals were fused because of the bony overgrowth. There were secondary degenerative arthritis changes in intertarsal & tarso-metatarsal joints & visualized interphalangeal joints (Figure-3).

{image:3}

Magnetic resonance imaging (MRI) demonstrated diffusely excessive fat deposition with fat lobules formation in the foot in the subcutaneous tissues, along the tendons, and around the neurovascular bundle (Figures-4 & 5). Bony abnormalities were more clearly seen on CT and were same as seen on CT.

{image:4}

{image:5}

In view of X-ray, NCCT & MRI findings, diagnosis of Macrodystrophia Lipomatosa was made and the patient was sent for corrective cosmetic surgery and the excised tissue showed excessive adipose tissue and diagnosis was confirmed.

DISCUSSION

Macrodystrophia Lipomatosa is a non hereditary, congenital developmental anomaly which results in overall progressive overgrowth of all of the mesenchymal elements of a digit including phalanges, nerves, digits and vessels [2]. In 1925 this condition was first described by Feriz, who used the term Macrodystrophia Lipomatosa to refer to localized gigantism of the lower limb. Then Golding in 1960 extended the term to involve upper limb also [3].

True macrodactyly is divided into static and progressive type. In static type the growth rate of enlarged digit is normal and in progressive type including Macrodystrophia lipomatosa, the growth of affected digit is accelerated [4]. Clinically, findings are present at birth. An equal incidence is present in males and females [3]. Involvement of an extremity is mostly unilateral and most often adjacent digits of the upper and lower extremity can be involved. Bilateral involvement is rare though reported [5].

The lower extremity is more often involved than the upper extremity. The 2nd and 3rd digits are the usual sites in the distribution of the median and plantar nerves in hand and foot respectively [6].

Involvement causes cosmetic disfigurement and later reduced functions are encountered due to secondary degenerative joint disease and moreover osteophytes overgrowth may also cause compression of adjacent nerves and vessels [2]. There is high incidence of associated local anomalies like syndactyly, polydactyly and more commonly clinodactyly [1].

Etiology is unknown and several theories exist, including lipomatosis degeneration, disturbed fetal circulation, and disturbance of growth factor in utero, error in segmentation, trophic influence of a tumefied nerve or an expression of neurofibromatosis [1].

Pathologically there is an increase in adipose tissue interspersed in the fine mesh of fibrous tissue that involves the bone marrow, periosteum, muscles, nerve sheaths and subcutaneous tissues [1].

On conventional X-Ray images macrodactyly and soft tissue

overgrowth are visible and are most marked along the volar aspect of the digit and at its distal end. This overgrowth can produce dorsal deviation of affected parts. Soft tissue radiolucency, representing overgrowth of fatty tissue, is seen. The phalanges are elongated, broad, and the distal ends are splayed and can have a “mushroom” shape. Slanting of the articular surfaces can occur and this leads to secondary degenerative joint disease manifesting in subchondral cyst and osteophytes formation [1, 2, and 3].

CT in this condition shows proliferation of fat along the nerve territory which is accompanied with bony outgrowth as was seen in our case [7, 8].

MRI in macrodystrophia lipomatosis demonstrates an excess of unencapsulated fibro-fatty tissue proliferation around the affected digits in the muscles, subcutaneous tissue and nerve sheath. Thickening of a nerve may also be seen in some cases and sometimes as in our case thickened nerve may not be seen, probably because fatty infiltration into the nerve sheath made its detection difficult with in the proliferative fat in the subcutaneous tissue [8, 9].

The differential diagnosis of congenital macrodactyly includes Neurofibromatosis, Klippel-Trenaunay-Weber syndrome, lymphangiomatosis, haemangiomatosis and fibrolipomatosis of the nerve. MRI and NCCT imaging, by characterizing the type of soft tissue proliferation can be used to differentiate between most of these diagnoses depending upon availability and macrodystrophia lipomatosis can be diagnosed confidently as happened in our case [8, 9].

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