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DELAYED PRESENTATION OF RARE OCCURRENCE OF MACRODYSTROPHIA LIPOMATOSA OF FINGERS

General Surgery

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ABSTRACT

Macrodystrophia lipomatosa is a rare congenital non-hereditary disorder that has significant impact on patient morbidity.[1]It is characterised by progressive proliferation of all mesenchymal elements, with disproportionate increase in fibroadipose tissue.[2]Macrodystrophia lipomatosa presents as localised gigantism of the hand or foot and comes to clinical attention for cosmetic reasons, mechanical problems secondary to degenerative joint disease, or development of neurovascular compression.[3,8] It is a hamartomous enlargement leading to localized or generalized gigantism of a limb which can present anywhere from infancy to late adulthood.This disease is characterized by progressive enlargement of the fibrofatty tissue.[4,6]Imaging findings include hypertrophy of the mesenchymal elements of the involved digits and/or limbs predominantly fibro adipose component with associated overgrowth of the phalanges.[5,6] Macrodystrophia lipomatosa affects either the upper or lower limb, classically occurring in a median or plantar nerve territory.[7]In the vast majority of patients, overgrowth is unilateral and localized in the fingers or toes.[8]In this case report, we present a case of unilateral involvement of ring finger and middle finger of right hand with associated macrodactyly with a diagnosis of Macrodystrophia lipomatosa.

KEYWORDS

Macrodystrophia lipomatosa,Macrodactyly,Congenital limb deformities.

INTRODUCTION-

Macrodystrophia lipomatosa is a rare form of localized gigantism. It is a benign congenital nonhereditary condition. It is usually identified in the neonatal period [9]. It is characterized by a marked increase in all mesenchymal elements, which is dominated by fibro-adipose tissue in a fine fibrous network involving periosteum, bone marrow, nerve sheath, muscle, and subcutaneous tissue [10].It comes to clinical attention because of cosmetic reasons, mechanical problems secondary to degenerative joint disease.Radiological investigations, especially MRI, help to make a definitive diagnosis noninvasively and to differentiate it from other causes of macrodactyly.[11] A variety of terms have been used to nominate the condition like macrodactyly, megalodactyly, digital gigantism, macromelia, partial acromegaly, macrosomy, and limited gigantism.[12,13]Pathogenesis is still not fully understood, and various hypotheses have been proposed, including abnormal regulation of growth factors or genetic abnormalities such as gain-of-function mutations, which activate the protein kinase PI3K/AKT cell signaling pathway [14].The affected area grows in a normal pattern or much faster than the unaffected area, causing a gigantic disproportional appearance.[15,16]Both surgical and non-surgical approaches can be used in the management of Macrodystrophia lipomatosa, with varying complications and outcomes in different cases.[17,18]Due to the lack of demographic evaluation by cohort studies, the incidence is still unknown [19]. However, those who are affected by the condition, usually suffer from a debilitating functional and psychological effect both on them and their respective families.[20]In the vast majority of patients, overgrowth is unilateral and localized in the fingers or toes.The rate of accelerated growth varies from patient to patient and even digit to digit.The etiology of Macrodystrophia lipomatosa is still not elucidated because of its rarity.[21,22]Imaging studies play an important role in the diagnosis of this disease as they allow characterization of the nature of the hypertrophied tissue.[23]

Case Report-

A 42 year old male from Maharashtra, presented to our opd with complaints of overgrowth of the ring finger and middle finger in right hand since birth.Left hand was normal.Visual examination was suggestive of macrodactyly of middle and ring finger of right hand,which caused mild discomfort to him with a history of no tenderness. He first noticed it's overgrowth in his childhood but he took no treatment. The overgrowth gradually increased since childhood. There was no history of trauma. It did not cause any neurologic symptom. The patient presented to OPD due to cosmetic reasons and mild discomfort.There was no family history of this disease and patient decided to undergo surgery due to inability to work and cosmetic reasons. Radiological examination of the right wrist with AP/LAT view revealed bony hypertrophy/overgrowth of middle finger and ring finger with deformity of underlying bones and diffuse soft tissue swelling. The rest of the visualised bones were normal along with normal radiocarpal, carpometacarpal, intercarpal and interphalangeal joints in configuration and alignment. The periarticular soft tissue is normal.Based on the clinical presentation, radiographic, and MRI findings, a diagnosis of macrodystrophia lipomatosa was confirmed. The patient was informed about the available treatment options, including surgical intervention and he agreed to the amputation of the affected fingers. The pre-operative procedure included testing the patient for Anti HCV, HBSAG(Plain), HIV I and II all of which were non-reactive (negative). The CBC report revealed mild Leucopenia and the PBS showed normocytic normochromic RBCs. The coagulation studies, LFTs, RFTs, TFTs, Lipid profile, Random blood sugar and Urine reports were all under normal limits. An ECG was done and electrocadiography revealed Grade 1 diastolic dysfunction and right ventricular dysfunction. The patient was pre operatively given NBM, Inj TT (0.5ml IM), Inj XST and Inj Cefuroxine.A pre anaesthetic check up was suggested to the patient.Racket incision was taken at the level of metacarpal level on the middle finger and this incision was deepened and then the digital arteries were ligated and cut was taken in between metatarsal bone and it was amputated with gigli

saw.On the ring finger,the tendons were incised and cut was taken on the proximal interphalangeal joint and the amputation was succesfully done by securing the digital vessels and digital nerve,then flaps were sutured with ethion(3-0).

DISCUSSION-

Image-Pre operative clinical image of overgrowth/hypertrophy of middle and ring finger of right hand.

Image-Radiological Image of AP/LAT view of right hand suggestive of Macrodystrophia lipomatosa.

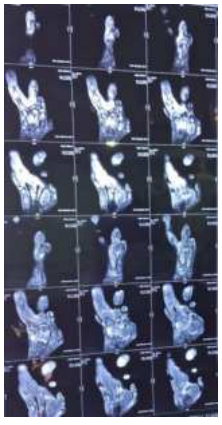

Image-radiological Image[mri] Showing Diffuse Hypertrophy Of Fatty Tissue In The Middle Finger And The Ring Finger Of The Right Hand.

Macrodystrophia lipomatosa is a rare congenital non-hereditary anomaly characterized by an abnormal overgrowth of the mesenchymal elements resulting in gigantism of single or multiple digits or the entire limb.[23,24]Macrodactyly, an unusual congenital anomaly in which there is hamartomatous proliferation of soft tissue of the affected digit, which is also called local gigantism of which macrodystrophia lipomatosa is a rare cause.[24] It is characterised by excessive growth of fibro-fatty tissue with unusually large fatty lobules, apparently fixed by a mesh of dense fibrous tissue.

Hypertrophy and tortuosity of the digital nerve, a striking feature in macrodactyly of the hand, is notably absent in cases affecting the foot[24,25]Feriz in 1925 coined theterm Macrodystrophia lipomatosa. Barsky in 1967 gave a detailed description of local gigantism and he differentiated two forms of it, namely static and progressive .[13,25]The etiology of it remains obscure but theories include lipomatous degeneration, disturbed fetal circulation, an error in segmentation, the trophic influence of a tumefied nerve and the in utero disturbance of a growth-limiting factor.[26,27,28] Macrodystrophia lipomatosa usually presents at birth and may be associated with anomalies like syndactyly,polydactyly, brachydactyly or clinodactyly. Association with small osseous protuberances, which resemble osteochondromas and lipomas, in other parts of the body has also been reported.[19,28]The second and third digits of the hands and feet are most frequently involved,corresponding to the median nerve and medial plantar nerve supply in the upper and lower limbs,respectively. Involvement of the ulnar nerve distribution, is extremely unusual. [20,29]

Different radiographic tools are utilized in Macrodystrophia lipomatosa. Plain radiography (X-ray) can detect any abnormalities in bone, soft tissues, and joints.The phalanges, especially the distal phalanx, are elongated, broad, and splayed, sometimes giving rise to a mushroom-like appearance.Secondary osteoarthritic changes like joint space narrowing, sub chondral cysts, and osteophytes often develop in adolescence or early adulthood.[15,29,30] Magnetic resonance imaging (MRI) can show fat tissue predominance and the condition of nerves and their sheaths.The fat in Macrodystrophia lipomatosa is not encapsulated. The fibrous strands within the fatty tissue are seen as low-signal-intensity linear strands on T1W images.The fibrofatty tissue is detected by the MRI same as that of the subcutaneous fat such as high signal on T1W , intermediate signal on T2W and low signal on fat supressed sequences.[17,30]Computed tomography (CT) scans are used to detect proliferation of fat with bone overgrowth. Ultrasonography can be performed to detect any calcification and abnormal blood flow.Both USG and CT scan can be used to demonstrate the proliferation of fat along the nerve territory[30,31]

The differential diagnoses of Macrodystrophia lipomatosa and macrodactyly include neurofibromatosis, hemangiomatosis, lymphangiomatosis, Proteus syndrome, and fibrolipomatous hamartomas, Klippel-Trenaunay-Weber syndrome.[32]

Neurofibromas show marked hyperintensity on T2W images and are seen in close relation to nerves.In neurofibromatosis, the enlarged digits are usually bilateral, there may not be contiguous involvement of an extremity, and the distal phalanges are not the most seriously affected. The soft tissue lucency associated with neurocutaneous manifestations in neurofibromatosis have not been reported with macrodactyly.[32,33]In hemangiomatosis,on MRI, long TR/TE sequences show a septate configuration of high signal-intensity channels, corresponding to the vascular channels and fibrous strands found in hemangiomas.[34]Lymphangiomas are hyperintense to muscle on T1W and hyperintense to fat on T2W images. Clinically, diffuse swelling and pitting edema are found.[35]Proteus syndrome presenting with hemihypertrophy may simulate Macrodystrophia lipomatosa, but associated abnormalities like calvarial changes, pulmonary cysts and intra-abdominal lipomas help to arrive at the correct diagnosis.[36]In Fibrolipomatous hamartoma the deposition of fat is within the nerve sheath, while in macrodystrophia lipomatosa it occurs throughout the involved part of the digits/extremity.

Fibrolipomatous hamartoma may show a speckled appearance on MR, correlating with its histologically known architecture.[34,37]Surgical intervention is the treatment of choice for macrodystrophia lipomatosa. The main surgical principle in treating these lesions is to improve the cosmetic appearance while preserving the neurologic function as much as possible. Through judicious and planned multiple debulking operations and partial amputations, good results can be achieved.[36,37,38]

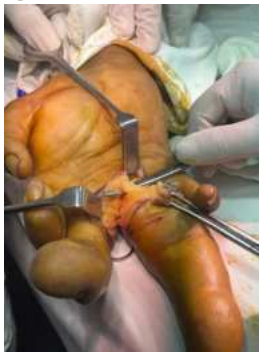

Image-Intraoperative photo with photo of clinical specimen of Macrodystrophia lipomatosa taken during the surgical procedure of amputation of the finger.

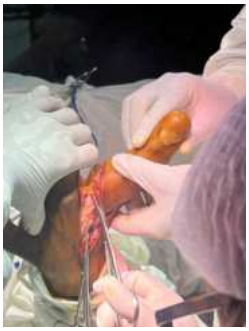

Image-Intraoperative photo with photo of clinical specimen of Macrodystrophia lipomatosa taken during the surgical procedure of amputation of the finger.

Image-Post operative image taken after the completion of the surgical procedure.

Image-Post operative image of the right hand which was diagnosed with Macrodystrophia lipomatosa.

CONCLUSION-

Macrodystrophia lipomatosa is progressive hamartomatous enlargement of the fibrofatty tissue involving all the layers of soft tissue and even bone more commonly leading to localized gigantism.Determination of the cause of macrodactyly is clinically difficult due to many probable etiologies. Radiological findings help distinguish this condition from other causes of localised gigantism.[35,37]Surgery might be a better choice of management than observation, taking into account future complications in the absence of surgery and the beneficial outcomes of surgical procedures for patients. Patients should be followed up regularly to determine the incidence of reoccurrence.A detailed understanding of the anatomy of the hand and fingers, especially the digital nerve plexus, is crucial to facilitate the detection of common abnormalities associated with macrodactyly. [23,38]

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