CASE REPORT

Macrodystrophia lipomatosa of the leg and foot: A case report and a concise review

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Abstract:

Macrodystrophia lipomatosa is a rare, congenital, non-hereditary anomaly of hand or foot which may gradually involve whole of the limb. It is characterized by excessive proliferation of fibroadipose tissue. We report a case of an eighteen-month-old girl with gradual enlargement of right leg and foot. She was diagnosed as a case of macrodystrophia lipomatosa on radiograph and ultrasound of the affected part. Now the patient has been advised for debulking surgery by the plastic surgeon. Early diagnosis and surgical intervention can improve the functional disability to a greater extent and provide the child a near normal life.

Keywords: Macrodystrophia lipomatosa, localised gigantism, overgrowth syndromes

Introduction

Macrodystrophia Lipomatosa (MDL) is a rare, non-hereditary congenital anomaly of the extremities which is characterized by unusual proliferation of the mesenchymal components, more commonly the subcutaneous adipose tissue [1]. The disproportionate enlargement causes marked functional disability and cosmetic problems. In comparison to the involvement of the proximal and upper limbs, the incidence of distal extremity and lower limb involvement is described more frequently in the literature [2]. The incidence of this anomaly cannot be discussed due to its rarity. Although the etiopathogenesis is not fully understood, new research suggests that Pik3Ca may play a major role in some patients with this overgrowth syndrome [3]. We report a case of an eighteen-month-old girl with gradual enlargement of the right foot and leg, and we aim to provide a concise review about the topic.

Case Report

An eighteen-month-old girl was brought to our tertiary care hospital with the complaint of gradual enlargement of the right leg and foot predominantly the great toe and the second toe (Figs. 1A and 1B).

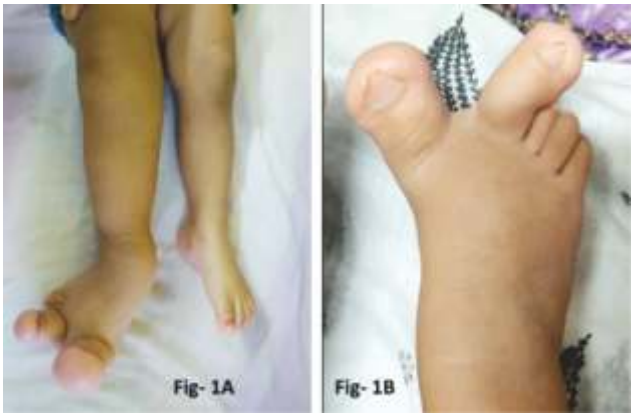

Figure 1A: Showing the hypertrophy of right leg and foot as compared to the left leg and foot

Figure 1B: Showing the hypertrophy of great toe and the second toe with sandal gap between them and suggests a volume of fatty tissue between them and under the foot

The great toe and second toe started to expand a few days after the baby was born, and throughout the course of these 18 months, the entire right foot and leg was affected. The left lower extremity and upper extremities were both healthy. There was no prior history of injury, pain, or skin lesions. No notable family history existed. All the other developmental domains were in normal range, and she could support herself while standing. The affected limb appeared flabby upon examination. Up to the knee, there was disproportionate hypertrophy that was non-tender and soft in consistency. Over the affected leg, there was no bruit or pitting edema. The affected limb's X-ray revealed hypertrophy of the soft tissues in the foot and leg as well as overgrowth of the foot's bones (Figs. 2A and 2B).

The results of the ultrasonography were compatible with the findings of MDL and showed evidence of disseminated fatty hypertrophy that appears hyperechoic down the right leg and foot with normal vascularity. The parents were advised to use magnetic resonance imaging to determine the degree of tissue involvement and hypertrophy, but due to financial restrictions, we were unable to do so and that is the limitation of the report. The child was determined to have macrodystrophia lipomatosa of the right leg and foot based on the results of the radiograph and ultrasound. Now, the patient is advised for debulking and corrective surgery by the plastic surgeon.

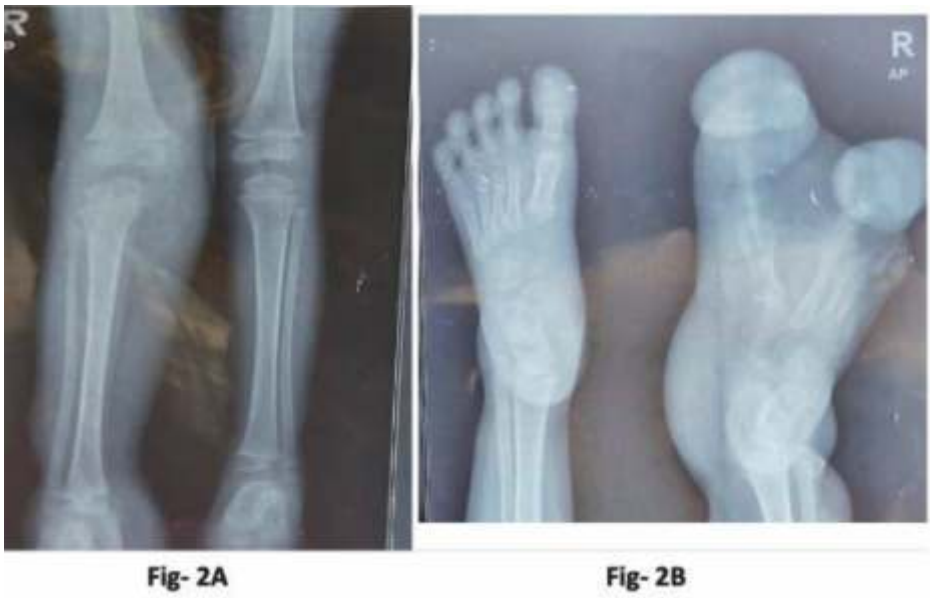

Figure 2A: Radiograph of both the legs in anteroposterior view showing the subcutaneous hypertrophy of the right leg with normal left leg with hypertrophy of the distal part of the right femur

Figure 2B: Radiograph of both the feet in anteroposterior view showing the bony overgrowth and subcutaneous hypertrophy in the left foot with normal left foot

Discussion

Macrodystrophia lipomatosa is an uncommon, non-hereditary congenital anomaly in which there is uninhibited proliferation of the mesenchymal components like muscle fiber, bony cortex, bone marrow, nerve sheath but most reported cases are associated with proliferation of the subcutaneous fibroadipose tissue [1]. Among the reported cases, most of the cases involve distal upper limb and lower limb corroborating the hypothesis that it progresses along the median nerve and plantar nerve distribution [2]. Barsky classified it into two forms- static and progressive. In static form, the affected part enlarges proportionately whereas in the progressive type, the affected part enlarges disproportionately [4]. Many reports have postulated that in MDL, the gradual progression of the affected limb ceases with attainment of puberty. This can be considered true in case of static type of MDL as growth potential of body tissue is attained at puberty therefore the inhibited growing fibroadipose tissue may halt its growth along with other body tissue [5].The etiopathogenesis is not well established for this condition but abnormality in fetal circulation, extremity bud being damaged, improper division during intrauterine life and concerned nerve hypertrophy and role of Pik3Ca are some of the hypotheses postulated by the researchers [6].The differential diagnosis of MDL is - Neurofibromatosis Type I has a strong family history and associated skin lesions like café au lait spots, axillary freckles, neurofibromas etc. and it does not involve distal digits commonly [7]. Hemangiomatosis involves blood vessels; the neovascularization is evident on doppler and an auscultatory bruit can also be present. Lymphangiomatosis can present with gross enlargement of limbs but there is associated pitting edema which is a necessary sign because of the underlying pathophysiology. Klippel-Trenanunay-Weber Syndrome is a close differential diagnosis but presence of port wine stain from birth and arteriovenous fistulae distinguishes it from MDL [8]. Beckwith Weidmann syndrome is also an overgrowth syndrome characterized by macroglossia, abdominal wall defects, neonatal hypoglycemia and visceromegaly which are not seen in MDL. Ollier disease and Maffuci syndrome are characterized by multiple enchondromas [9]. Our patient had bony overgrowth and adipose tissue proliferation causing localized gigantism but there was no skin lesion, vascular, cartilaginous, or visceral involvement. Various radiological modalities can be used to diagnose MDL, the X-ray of the affected part shows involvement of bone and soft tissue. Magnetic Resonance Imaging (MRI) shows the extent of fat proliferation and involvement of nerves. Ultrasound of the affected part can show if there is associated calcification and abnormal blood flow. Histopathology, which shows numerous adipose tissue cells infiltrating the subcutaneous connective tissue and proliferating nervous tissue in the subcutaneous region, was initially thought to be the gold standard [5]. However, as technology has advanced, genetic testing for Pik3Ca is now thought to be the most conclusive test [3]. Macrodystrophia lipomatosa can be managed by surgical and non-surgical approach depending on the age, type, and extent of tissue involvement. If its static and associated with minimal functional disability, the surgical intervention can be delayed but if it is progressive and has higher degree of functional disability, the

surgical intervention should not be delayed. The mainstay of treatment remains surgical which is a debulking or liposuction surgery. Debulking surgery can be a one stage procedure or multiple stage procedures [5]. There can be a need for partial or complete amputation depending on the extent of the tissue involvement. But preserving the nerve and muscles is the aim while performing this extensive surgery to maintain the function of the limb. The recurrence rate is 33-60% with these multiple debulking procedures therefore reconstructive surgeries can also be preferred [6].

Conclusion

Macrodystrophia lipomatosa is an overgrowth syndrome characterized predominantly by uninhibited proliferation of fibroadipose tissue. It presents with disproportionate enlargement of the digits or limbs causing varying degree of functional and aesthetic disability. It presents in childhood which is a sensitive and impressionable age group therefore, early diagnosis and intervention is prudent to avoid low self-esteem, minimize the functional disability and have a near normal life.

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