MACRODYSTROPHIA LIPOMATOSA: A CASE REPORT WITH REVIEW OF LITERATURE

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ABSTRACT: Macrodystrophia Lipomatosa is a rare cause of congenital macrodactyly which is characterised by progressive proliferation of all mesenchymal elements, with a disproportionate increase in fibroadipose tissue. [1] It presents as a localised gigantism of hand or foot. This developmental anomaly is reportedly more common in foot than in hand, with a predilection for the plantar and median nerve distribution. Clinical diagnosis is possible with the help of radiography or magnetic resonance imaging(MRI). There are many clinical conditions with similar presentation but imaging and histopathology can lead to confirmatory diagnosis. The treatment option is surgical excision preferably cosmetic surgery.
KEYWORDS: Macrodystrophia Lipomatosa, gigantism, nerve involvement, upper limb.

INTRODUCTION: Macrodystrophia lipomatosa is a rare non-hereditary congenital developmental anomaly leading to disproportionate overgrowth of one or more digits. It may cause local gigantism or involve the entire limb. It has been given various names like, fibrolipoma of nerve, fibrolipomatous hamartoma of nerve and lipomatosis of nerve often described as having a ''coaxial cable-like'' appearance on axial scans.[2]It has also been described as megalodactyly, macrosomia and dactylomegaly.

CASE REPORT: We describe a case of a teenage boy presenting to the dermatology outpatient with history of disproportionately large middle and ring finger of the left hand. On detailed history and examination the findings which were revealed are as follows: swelling of both fingers since birth, no history of any family members with similar complaints not associated with pain. On clinical examination middle finger of left hand measured 9x4x3 cms and index finger ms 8x4x3 cms (figure 1).

No sign of local inflammation or increased vascularity was seen and it was a non tender swelling. Swelling extended upto the medial aspect of the palm (figure2) and slightly to wrist. The overlying skin was pale and thickened. There was no bony deformity noted. Histopathology unit of Pathology department received skin biopsy from representative sites.

Grossly specimen consisted of 3 tiny grey-white irregular bits of tissue largest measuring 0.3 cms. Entire tissue was processed. Microscopy revealed structure of skin with hyperkeratosis; epidermis was unremarkable (figure3). Reticular dermis and subcutis showed bundles of adipose tissue, nerve fibres, blood vessels and muscle bundles. The characteristic findings were infiltration of adipose tissue in the nerve bundle (figure4) and almost replacing the nerve fibres (figure5).Figure 6 shows sheet of adipocytes surrounding a blood vessel. Histopathology report was concluded as Macrodystrophia Lipomatosa. But before finalizing the report a detailed clinical history was taken

from dermatology department and in fact patient was called again for a thorough clinical examination before finalizing the report.

DISCUSSION: Macrodystrophia lipomatosa is a rare congenital disorder of focal gigantism affecting the extremities. Local gigantism has also been described under various names already mentioned earlier in this article.[3]Different authors have given contradicting views regarding the limb involved. According to few authors around 85% of cases show involvement of median nerve and its digital branches in hand, wrist and forearm.[2] It was Feriz in 1925 who first used the term Macrodystrophia lipomatosa for local gigantism of lower extremity. Bursky gave a more detailed description of the disease. He described two dt forms of the true macrodactyly. The first one also called as Static form where the size of enlarged digits increases proportionally in relation to the rest of the body. The other type is progressive in which the growth rate of enlarged digit is disproportionate to the growth of rest of the body and this type is less common. It may be associated with fatty overgrowth.

MDL is non-hereditary condition and shows a slight male preponderance.[4]There are various hypotheses that have been proposed regarding the etiopathogenesis of MDL. These include Lipomatous degeneration, fetal circulation abnormality and errors in the segmentation in intrauterine life and hypertrophy of the concerned nerve. There are various other conditions that have similar presentation.[5]That is why it would be wise to discuss the differential diagnosis of this condition. Hence the differential diagnoses are neurofibromatosis, hemangiomatosis, lymphangiomatosis, proteus syndrome and fibrolipomatous hamartoma. In case of neurofibroma, positive family history, presence of cutaneous lesions and bilaterality favours the diagnosis. Lymphangiomas show characteristic diffuse swelling and pitting edema. In hemangiomatosis a bruit may be palpable on clinical examination. Proteus syndrome presenting with hemihypertrophy may simulate MDL, but associated abnormalities like calvarial changes, pulmonary cysts, pigmented nevi and intra abdominal lipoma, help to arrive at correct diagnosis. According to few authors MDL is a localised form of Proteus syndrome. Fibrolipomatous hamartoma of nerve is a rare tumour like condition in which mature fat infiltrates the neural sheath, with majority of lesions occurring in median nerve. Pathologically in Fibrolipomatous hamartoma, the deposition of fat within the nerve sheath, while in MDL it occurs throughout the involved part of the digit/extremity.[1]Congenital macrodactyly is an independent pathological process and this is consistent with what Barsky postulated i.e. macrodactyly is the result of interaction of two or more intrinsic and extrinsic agents(genetic or environmental)each unable to produce the deformity by itself.[6]Exact etiology is not known though there are many theories which have been attributed to its etiology. Abnormal nerve supply, abnormal blood supply or humoral mechanisms. [7]

Since this condition is very rare it is obvious that whenever the dermatologist, pathologist, radiologist and surgeons come across such a case/cases they find it necessary to document it. The curiosity regarding the etiology, as well as proper treatment aspect is always there in the mind of clinician; hence discussions are always welcome in such disorders.

Table 1 summarizes the number of cases, site, histopathological findings, ancillary studies and mode of treatment.

Name of authors	Total number of cases	Location(upper limb, lower limb)	Histopathological findings	Ancillary studies	Treatment & effects
B.V Balakrishna et.al	2	Lower limb	Adipose tissue	Radiogram & Doppler study	Surgical excission.
V.Singhla et.al	2	Upper limb	Adipose tissue with fibrous tissue	Radiogram&MRI	Surgery with tendons protection.
Farhana Ebrahim et.al	2	Arm & Lower limb	Adipose tissue	Radiogram&MRI	Surgical excission.
Rizwaan Khan	4	Lower Limb	Aippose tissue & Proliferation of subcutaneous nerves	Radiogram	Biopsy & excission
Wg Cdr S Sahu et.al	1	Lower limb(great toe)	Adipose tissue in mesh-like fibrous tissue	Ultrasound & Doppler	Cosmetic surgery
Gandage et.al	1	Right hand (little finger)	Fibrofatty tissue & adipose tissue	Radiogram & CT scan	Surgical excision

TABLE-1 showing the number of cases,site histopathology ,ancillary studies & mode of treatment.

B.V Balakrishna and S.R.H Prasad have reported two cases both with lower limb involvement (2005). [8] Anjum Syed et. al have reported five cases, three with involvement of foot and two with hand involvement 2005. V Singhla et.al has reported two cases where upper limb was involved (2008). Farhana ebrahim et.al documented two cases, one in a young kid with left arm involvement and another case of 11 month old girl with right lower limb involvement (2010). Rizwan A Khan et.al has documented four case reports, all cases involving lower limb (2010). Wg Cdr Sahu et.al has reported a case of 18 years old boy with enlargement of right great toe (2011). Gandage et.al have reported a case with hand involvement (2012).Though literature based on certain authors suggest that 85% cases have involvement of median nerve and its digital branches in hand, wrist and forearm [9] but if we glance through the above mentioned few case where 11 cases show lower limb and 5 cases show upper limb involvement by the disorder, it becomes clear that lower limb is commonly involved. Radiological findings do play a major role in correct diagnosis where C.T scan confirms the exact nature of the swelling. Ultimately it is histomorphological features which must be clear and specific so as to reach a correct diagnosis. So it is a team work where clinician, radiologist

and pathologist contribute in making the diagnosis. The patient was advised surgery by the cosmetic surgeon but he did not turn up. Hence we lost the patient for follow-up. Since the approach and treatment modality differs for the different disorders which mimic this condition (MDL), one has to diagnose responsibly.

CONCLUSION: Besides a detailed clinical examination, it is the imaging and histopathology which is crucial in clinching the diagnosis.

Conflict of interest: The authors have no conflict of interest to disclose.

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Figure 1- Clinical presentation with enlargement of left hand index and middle fingers.

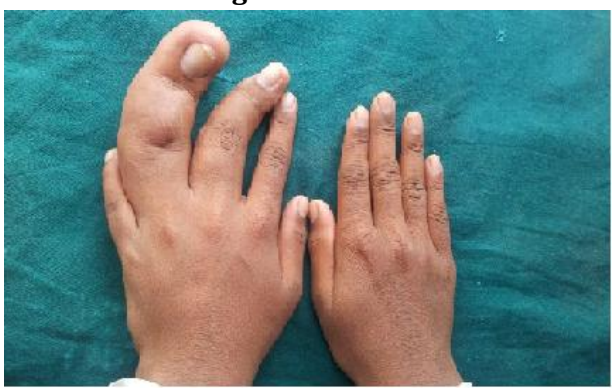


Figure 2- Ventral aspect of left hand showing thickening of palm.

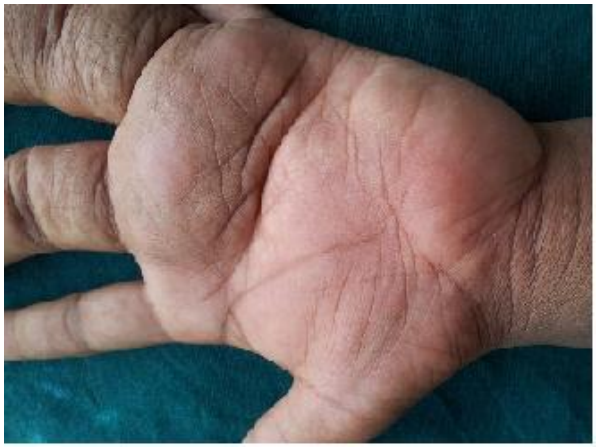


Figure 3 - Shows hyperkeratosis and thickened epidermis.

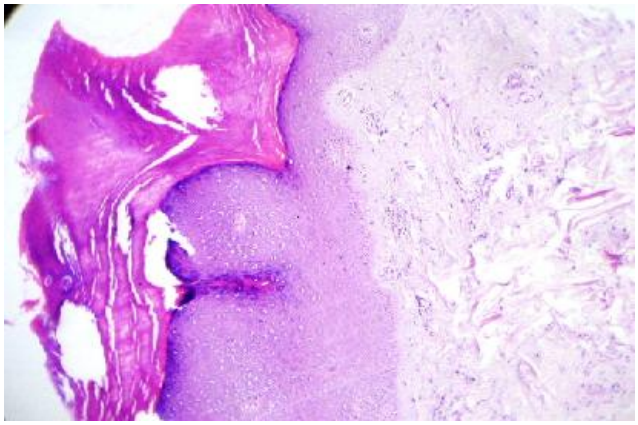


Figure 4 - Shows nerve bundle surrounded by adipose tissue

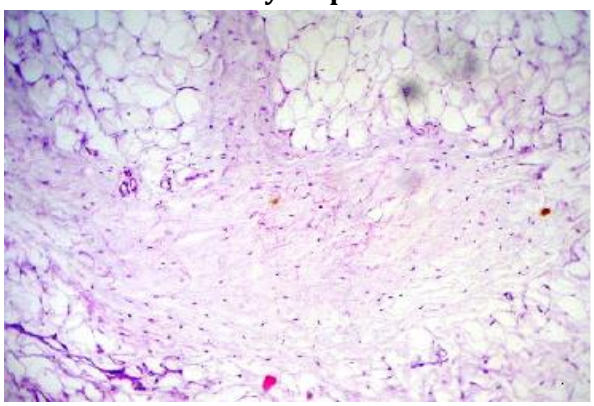


Figure 5 - Scanner view showing abundant fatty tissue invading the nerve tissue

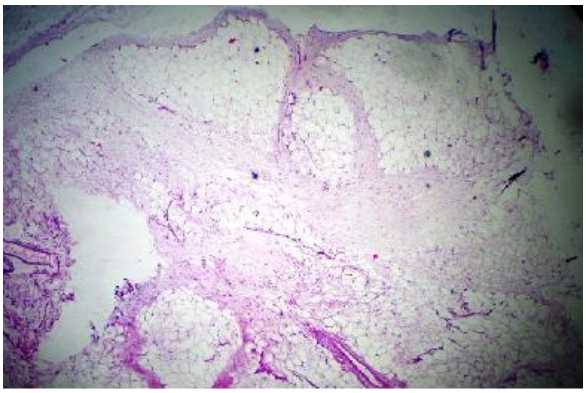
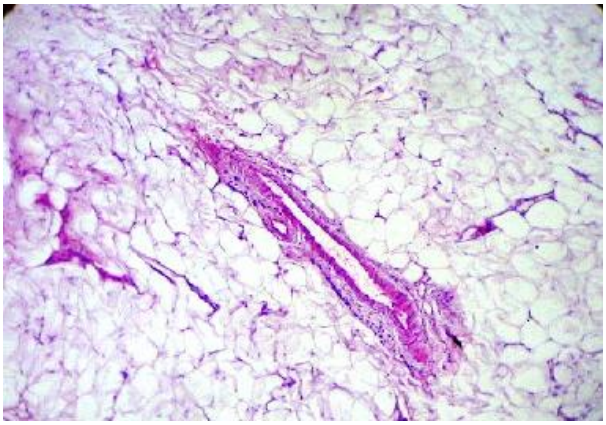


Figure 6 - Shows abundant fatty tissue around a blood vessel.



CASE REPORT

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