ORIGINAL RESEARCH PAPER

Pathology

MACRODYSTROPHIA LIPOMATOSA: ENSUE AS LOCALIZED GIGANTISM (TWO CASES)

KEY WORDS: Macrodystrophia Lipomatosa, gigantism, adipose tissue, non-hereditary.

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ABSTRACT

Background - Macrodystrophia lipomatosa is a rare non -hereditary congenital disease. The exact etiology is unknown. The specific body region; sclerotome is involved followed by mesenchymal overgrowth in that region. Cosmetic disfigurement and mechanical dysfunction are two problems associated with Macrodystrophia lipomatosa. The characteristic histopathology finding in Macrodystrophia lipomatosa is increased deposition of adipose tissue in periosteum, nerve sheath, muscle, subcutaneous tissue as well as bone marrow.

Case series – Two cases of macrodystrophia lipomatosa are reported. Cases presented as gradual painless macrodactyly since birth. Involvement of left middle finger observed in case one whilst, in case two involvement of right thumb, index and middle finger noted. On clinical examination the hypertrophied digits were soft, non-tender and non-pulsatile. The radiology investigation revealed increased soft tissue shadow as well as bone hypertrophy in both the cases. The histopathology examination confirmed the excessive deposition of adipose tissue not only in in soft tissue planes but also in inter-trabecular spaces.

Conclusion – Macrodystrophia lipomatosa is rare non-hereditary condition leading to macrodactyly. The histopathology examination is the gold standard for diagnosis. The mode of management is surgical but does not ensure a favorable outcome.

Introduction

Macrodystrophia lipomatosa (ML) is a rare non-hereditary congenital disease. This uncommon developmental condition is characterized by disproportionate overgrowth of all mesenchymal elements particularly adipose tissue leading to localized gigantism of the limb. [1] This overgrowth generally comes to halt at puberty. Here we report two cases of macrodystrophia lipomatosa in pediatric patients.

Case one

A one year old male child presented with macrodactyly of left middle finger in department of plastic surgery. The enlargement was gradual and painless since birth. The middle finger was nonfunctional. Clinical examination revealed significantly enlarged left middle finger (Figure 1). The swelling was soft, non-tender and non-pulsatile. The radiology examination showed the hypertrophy of phalanges as well as increased soft tissue component. There was no other significant clinical history or examination finding. Amputation of the middle finger was done and sent for histopathology examination. Gross examination of the middle finger revealed a dorsally curved finger over lined by corrugated skin and significant enlargement of size measuring 8cm in length and 2.5cm in maximum diameter. The microscopic examination showed stretched thinned out over lined skin with notable increase in underlying adipose tissue. The adipose tissue is seen encircling the nerve bundles (Figure 3b) and skin adnexal structures. The partially ossified bone is seen with inter-trabecular hematopoietic tissue (Figure 3c).

Case two

A five year old female child presented with painless progressive enlargement of right thumb, index and middle finger since birth. The child's birth and developmental history is unremarkable. On examination the child was left hand dominant because of hypertrophied non – functional right hand fingers. The length of right forearm was more than left one. The plain radiograph showed enlarged bones of phalanges and metacarpal (Figure 2). The amputation of the right middle and index finger were performed and sent for histopathology examination. Gross examination revealed significantly enlarged soft middle and index finger with dorsal curve. The overlying skin was corrugated. On microscopic examination there was seen remarkable increase in adipose tissue with entrapment of nerve bundles within (Figure 3b). The bony tissue with intertrabecular hematopoietic tissue (Figure 3c) is also noted.

Discussion

The Macrodystrophia lipomatosa (ML) is uncommon congenital anomaly denoted by mesenchymal tissue overgrowth leading to gigantism of the entire limb, single or multiple digits. The term Macrodystrophia lipomatosa was coined by Feriz in 1925. However, in 1967 Barsky described the entity local gigantism in detail and differentiated it in two forms, namely static and progressive. [2]The exact etiology is unknown. [3]The condition is non-hereditary and usually unilateral however, there are few case reports suggesting bilateral involvement. [4] There is slight male preponderance. [2] The age of clinical presentation varies between neonatal period to adulthood. [2] Involvement of lower limb is more frequent compare to upper limb. The specific body region; sclerotome is involved followed by mesenchymal overgrowth in that region. In general, the medial aspect of lower limb and lateral aspect of the upper limb is commonly affected. [5, 6] Till puberty the overgrowth continues after that it reaches plateau. The characteristic histopathology finding in ML is increased deposition of adipose tissue in periosteum, nerve sheath, muscle, subcutaneous tissue as well as bone marrow. [7] If nerve is involved the commonest being median nerve in upper limb and plantar nerve in lower limb. Distal phalanges of involved limb or digit gives mushroom like appearance due to proliferation of the nodules of osteoblast, osteoclasts and chondrocytes residing in the periosteum. [8] Cosmetic disfigurement and mechanical dysfunction are two problems associated with ML. Cosmetic problem is there at any age of presentation while, mechanical difficulties starts at later age due to degenerative changes. Calvarial abnormalities, syndactyly, polydactyly, brachydactyly, symphalangism, pigmented nevus, lipomatous lesions in the intestine and pulmonary cysts are other associated lesions seen in patients with ML. [9]

Different radiology investigations play vital role in the diagnosis of ML but gold standard is histopathology. [10] The characteristic X-ray findings are increased soft tissue as well as osseous tissue and radiolucency due to increased adipose tissue. The CT scan shows two characteristic findings i.e. adipocytes proliferation in between muscle fibers and bone overgrowth in the region innervated by involved nerve.[2] Ollier disease, Proteus Syndrome, neurofibromatosis type 1 (Plexiform neurofibroma), fibrolipomatous hamartoma (FLH), hemangiomatosis, Klippel-Trenaunay-Weber syndrome, lymphangiomatosis and Mafucci syndrome comprises of differential diagnosis of ML. All the above diseases are hereditary and have both cutaneous as well as systemic organs involvement. [2]

Conclusion

There are many etiologies leading to macrodactyly. The histopathological examination and imaging studies especially MRI can clinch the diagnosis of the rare non-hereditary condition i.e. ML. The definitive management is surgical however; it does not ensure a satisfactory outcome.

Consent

Author declared that they have taken in-formed written consent, for publication of this report along with clinical material, from the legal guardian of the patient with an understanding that every effort will be made to conceal the identity of the patient however it cannot be guaranteed.

Authors' Contribution

Author has contributed in concept, literature review, drafting of the manuscript and approved the final version of this manuscript.

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Figure 1 The middle finger (Case one) showing hypertrophy and noticeable dorsal curve with corrugated overlying skin (Palmar and dorsal surface).

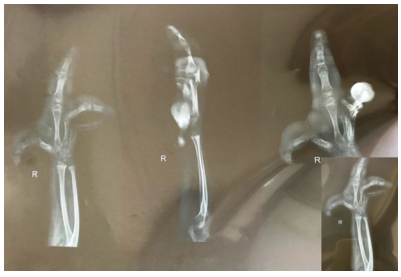

Figure 2 The X-ray (Case two) showing involved right middle and index finger with increased soft tissue shadow and elongated phalanges.

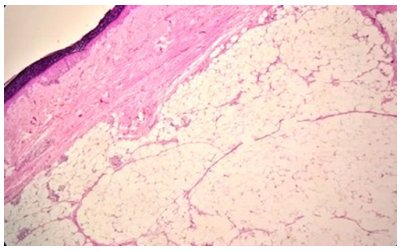

Figure 3 a) The microscopic image showing increased dermal adipose tissue (40x, H and E).

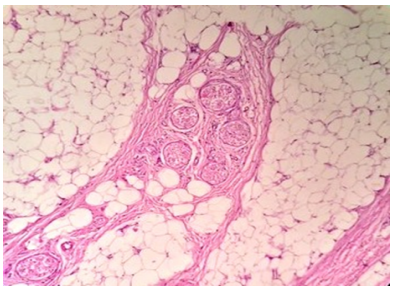

Figure 3 b) Hypertrophied nerve bundles with surrounding adipose tissue (400x, H and E).

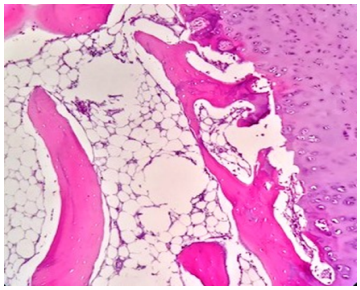

Figure 3 c) Increased inter - trabecular adipose tissue of bone (400x, H and E).

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