Case Report

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Macrodystrophia Lipomatosa: A Delayed and Rare Presentation in the Hand of a Young Adult

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Abstract

Macrodystrophia lipomatosa (MDL) is a rare developmental anomaly causing localized gigantism. It is due to progressive proliferation of all mesenchymal elements, especially fibroadipose tissue. This developmental anomaly is more common in lower extremity but sometime also found in upper extremity. The disease is almost always unilateral, with an equal incidence in both sex. MDL is usually reposted and diagnosed during childhood. We report a case of macrodystrophia lipomatosa in a young adult which started from a finger and gradually affected the whole hand leading to amputation of hand from wrist joint. This is a case of late presentation. Involvement of whole hand, as seen in our case, is not yet reported in English literature to the best of our knowledge.

Keywords: Local Gigantism; Macrodystrophia Lipomatosa; Adult; Hand.

Introduction

MDL is a rare non-hereditary congenital disorder characterized by three dimensional enlargements of one or more fingers or toes with predominantly fibro-adipose tissue. Cutaneous and systemic manifestations and isolated presentation in a family is the usual presentation. Diagnosis is usually confirmed by imaging and histopathology. Radiological finding is of hypertrophy of soft tissues and bones [1]. Diagnosis of this rare condition is of importance due to its similarity with neurofibromatosis but different clinical course.

Case Report

26 year old boy presented in surgical outpatient with complain of massive enlargement of right hand involving all fingers up to wrist joint. History revealed painless swelling of right hand which started from one finger at the early age of four years. With the increase in age other finger also started growing disproportionately bigger. There was neither pain nor any other medical disease. He never presented to any medical attention for this anomaly and this was his first presentation to any doctor for this problem.

Local examination revealed firm, nontender, nonpulsatile abnormal enlargement of the whole right hand (local gigantism). The skin over the affected part was dry and rough. There was a bluish discoloration above the enlarged hand. This was a recent development and a suspicion of compression of blood vessel was made. Plain radiogram of the hand demonstrated soft tissue enlargement with normal underlying bones. A decision for amputation of affected hand was made to prevent spread of gangrene. Hand was amputated from wrist and sent for pathological examination. Gross examination of amputated hand showed hypertrophied fingers and hand with dry appearance Figure 1. Histopathology showed hypertrophic fibroadipose tissue Figure 2.

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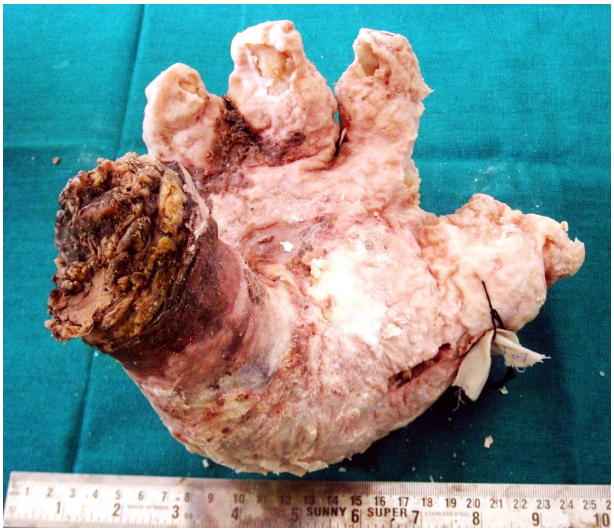

Fig. 1: Photograph of amputated hand showing local gigantism with dry overlying skin

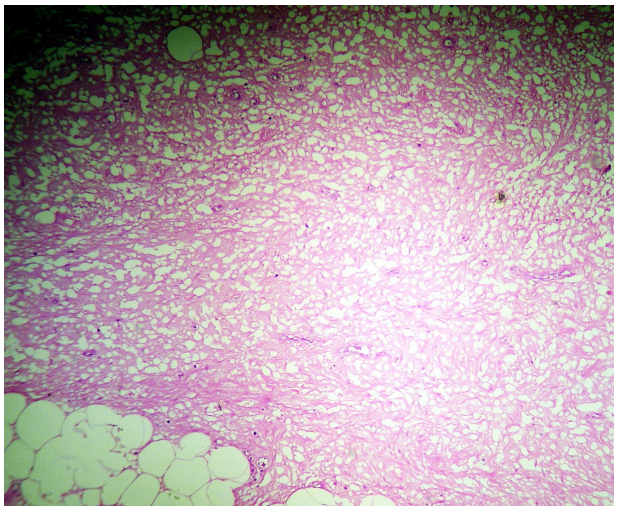

Fig. 2: Microphotograph showing hypertrophy of fibroadipose tissue. (H&E stain; X 100x)

Discussion

Macrodystrophia lipomatosa (MDL) is an unusual entity that predominantly affects children and is classified with other phakomatosis, such as Tuberus sclerosis and neurofibromatosis [1,2]. MDL was first described by Feriz in 1925. MDL is a rare nonhereditary congenital malformation that mainly affects mesenchymal elements especially fibroadipose tissue in a limb [3]. The condition is previously described as macrodactyly, megalodactyly, or localized gigantism. The abnormal tissue is often found along the median nerve in the hand and the plantar nerve in the foot. No causal link between hamartoma of a nerve and soft tissue enlargement has been established, and evidence of nerve involvement is controversial [4]. The disease is almost always unilateral with an equal incidence in males and females, but growth velocity may differ from digit to digit and the abnormal growth usually ceases at puberty. The second ad third digits of the hand and foot are most frequently involved [1-4].

Though, the exact etiology of MDL is not known, various hypothesis exist, including alteration of somatic cells during limb bud development and distributed fetal circulation. It is now believed to be part of generalized hamartomatous disorder known as proteus syndrome [1-6].

MDL presents as localized gigantism of the hand or foot and come to clinical attention because of cosmetic reasons, mechanical problems secondary to degenerative joint disease, or development of neurovascular compression [6,7]. The patient in the present case report had neurovascular compression. This type of presentation of MDL is very rare and not reported in medical literature to the best of our knowledge. This patient came to the doctor for cosmetic reason and neurovascular compression was an incidental finding by treating clinician.

The differential diagnoses include plexiform neurofibromatosis, Klippel-Trenaunay-Weber syndrome (mcrodactyly with hemangioma), lymphangiomatosis, hemagiomatosis, proteus syndrome, and fibrolipomatous hamartomas of the nerve. Clinical examination in conjunction with imaging should easily discriminate between the rests of the differential diagnosis. Cosmetic abnormalities remain a significant reason for surgical intervention in the treatment of MDL [2,4].

Considering the rarity of MDL of hand and in absence of any other clinical features, we can conclude that, "whatever the fundamental lesion" the final pathway was either local deficiency of growth inhibiting factors or excessive local expression of basic intrinsic factors, causing excessive growth of all elements of hand. If this patient would have taken medical opinion early in the course of disease, his hand could have been saved.

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