CASE REPORT

Case Report of Maffucci's Syndrome

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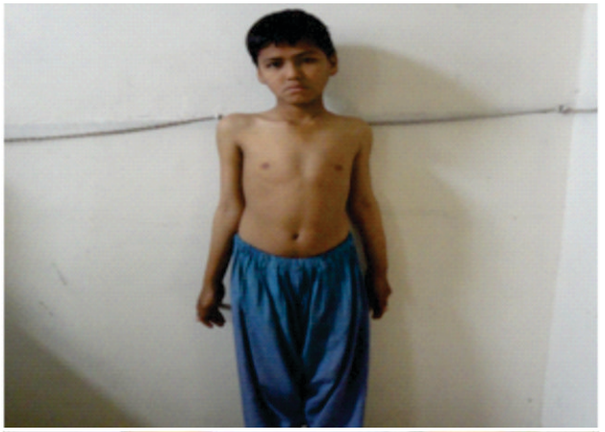
INTRODUCTION:

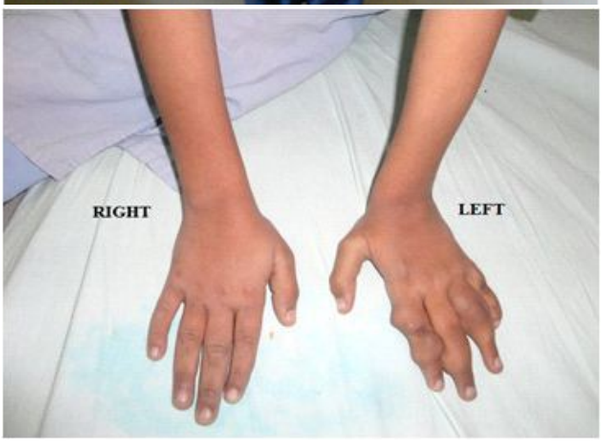
Maffucci's syndrome is a congenital, non-hereditary mesodermal dysplasia manifested by multiple enchondromas and hemangiomas.

Key Words: *maffucci's syndrome, enchondromas, hemangiomas.*

CASE REPORT:

A 9 years old boy presented with multiple swellings of the left hand since last 5 years (Fig. 1).





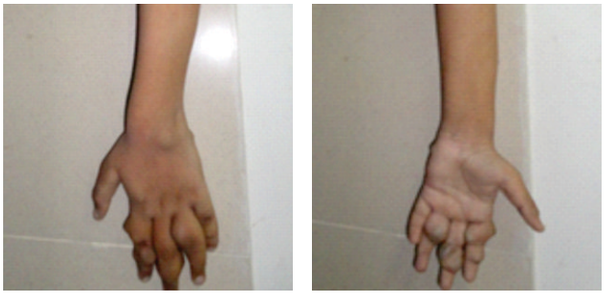


Fig. 1: The patient's presentation

The patient complained of intermittent pain in the swelling and hindrance in the day to day to work due to swellings. On examination swellings were multiple, bluish colored, of variable sized and shaped,non tender, normo-thermic, soft to firm ,compressible, non pulsatile with no bruit and thrill (Fig. 2).

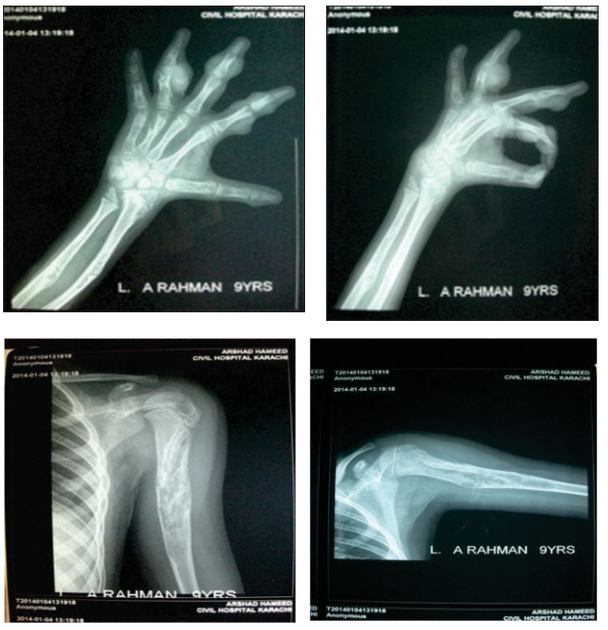


Fig. 2: Radiological findings

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There were palpable phleboliths . Swellings tend to increase with gravity.

Blood chemistry was normal along with coagulation profile. Radiographs shows multiple expansile osteolytic lesions involving midshaft of radius and ulna and middle phalanx of ring finger and distal end of radius (Fig. 3).

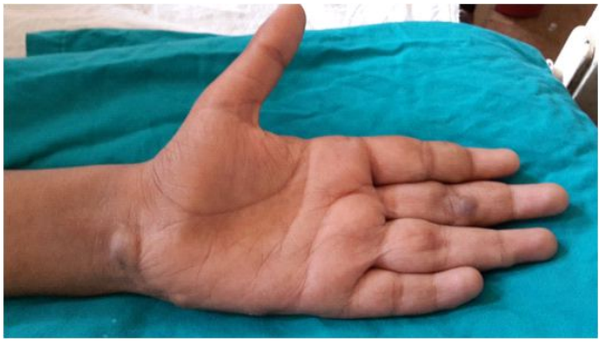

Fig.3 Postoperative result

Multiple soft tissue density lesions of 1st 2nd 3rd 4th fingers with multiple phlebolithsSkeletal survey including x-rays of chest, skull and spine did not reveal any abnormality.

All the lesions were excised and sent for histo-pathological examination (Fig. 4). Patient's parents were counseled about complications of disease especially about risk of malignant transformation.

Histopathology report showedsmall to medium sized blood vessels lined by flattened endothelial cells .Intervening tissue shows areas of congestion with extravasation of red blood cells. No evidence of fibromatosis, granuloma or malignancy was seen. Findings suggestive of vascular malformation. Histopathology of the hard nodules revealed mature lobules of hyaline cartilage in which foci of myxoid degeneration, calcification and endochondral ossification was seen.Findings were consistent with enchondromas.

DISCUSSION:

This rare syndrome was first reported by Maffucciin 1881, after a 40-year-old patient had frequent and severe bleeding that led to amputation of a distal extremity. The patient died of complications secondary to infection. Maffucci described a thorough autopsy and reported all the main points of the syndrome that was to be named after him. Carleton [1] proposed the eponym Maffucci syndrome in 1942 *Maffucci's syndrome is a combined slow flow malformation,denotes the coexistence of exophytic venous anomalies,with bony exostoses &enchondromatoses."*

It's a rare disease, less than 200 cases reported worldwide. It has no familial, racial & sexual predilection. It is not associated mental or psychiatric abnormalities.[2] It can manifests early in life(4-5y) but 78% of them manifest by puberty[3].

Enchondromas exists not only most frequently at the small bones of the hands and feet, the long tubular bones, but also the flat bones, such as pelvis. Enchondromas are usually in close proximity to or in continuity with growth plate cartilage. Consequently, they might be the result from abnormal regulation of proliferation and terminal differentiation of chondrocytes in the adjoining growth plate. The osseous lesions most frequently involve the phalanges, metacarpals and metatarsal. [4]

Maffucci syndrome might be associated with three types of vascular lesions: cavernous hemangiomas, phlebectasiasand lymphangiectasias-lymphangiomas . Clinical problems caused by enchondromas include skeletal deformity and the potential for malignant change, reported in approximately 30% of reported cases.[4]

Complications develop are pathological fractures, growth abnormalities and malignant transformation. Malignant transformation is a common complication and one should look for Radiologic evaluation of suspicious areas . Evidence of malignant transformation includes cortical destruction, endosteal cortical erosion, and zones of lucency within a previously mineralized area.[5]

Chondrosarcomas, the most common

malignant neoplasm associated with Maffucci syndrome, are diagnosed by poorly differentiated pleomorphic chondrocytes.[6] Maffucci syndrome patients have normal life expectancy if there are no complications.

CONCLUSION:

Though Maffucci's syndrome is rare, knowledge of its occurrence and complications to Plastic Surgeon can lead to better management.

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