

MAFFUCCI'S SYNDROME

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Maffucci's syndrome (MS) is a rare congenital nonhereditary disorder manifested by endochromatosis (dyschondroplasia) and soft tissue hemangiomas,¹ and having a high incidence of malignant transformation. Until now, no more than 120 cases have been reported in the literature. The presence of cyst-like bone lesions and multiple phleboliths constitute the characteristic plain roentgenographic appearance. An earlier report² suggested that this entity probably represents a mesenchymal dysplasia. We report a case in a Saudi seen in Arar Central Hospital, Arar, and discuss the clinical aspects and management of the condition with a review of the relevant literature.

Case Report

A 62-year-old Saudi man was referred for evaluation of a mild pruritic eruption and localized pain on the right hand of about three-months' duration. He had a life-long history of enlargement of the right hand, with multiple swellings scattered all over the body. The past medical history was unremarkable. There was no history of similar lesions in the family, including his offsprings. The patient was in good health except for a limping gait. On physical examination, the right hand had a grotesque appearance, with multiple compressible, soft blue swellings (Figure 1). Some areas were tender with localized sweating. A few nodules were scattered all over the body.

Routine laboratory testing showed a normal full blood count, blood sugar, electrolytes and urea, liver enzymes and urinalysis. Histopathological examination of one of the small nodules showed dilated blood-filled vascular spaces that were lined by flattened endothelial cells, typical of a cavernous hemangioma (Figure 2). Many of the vascular spaces showed recent as well as old thrombi.

FIGURE 1. Grotesque appearance of right hand showing multiple soft knob-like swellings which on histopathology showed cavernous hemangiomas.

Radiological examination of the right hand showed radiolucent lesions at the base of the proximal phalanx of the thumb (15 x 15 mm), with thinning of the cortex. Similar small subtle lesions were seen at the middle phalanx and the neck of the proximal phalanx of the index finger. Multiple phleboliths were seen scattered all over the hand (Figure 3). Ultrasonographic and computed tomographic examinations ruled out any associated vascular lesions of the liver, spleen, and both kidneys.

Based on the findings of soft tissue hemangioma and the radiological appearance of endochromatosis, a diagnosis of Maffucci's syndrome with mild eczema/dermatitis was made. The patient was placed on analgesics, mild topical steroids and oral antihistamines. The pain and dermatitis subsided with treatment. Although cosmetic surgery was considered after due consultation with the plastic surgeon, it was thought to be inappropriate in this case. The patient is still in good health, and has been having periodic examinations for early detection of any malignant degeneration.

Discussion

Multiple enchondromas (dyschondroplasia, Ollier's disease) with co-existing soft tissue hemangiomas is termed Maffucci's syndrome (MS). Both these conditions

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Accepted for publication 29 January 1997. Received 2 October 1996.

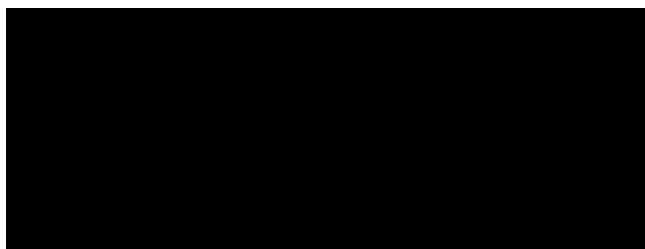


FIGURE 2. Cavemous hemangiomas with dilated blood-filled vascular spaces with thrombi (hematoxylin and eosin, 100x).

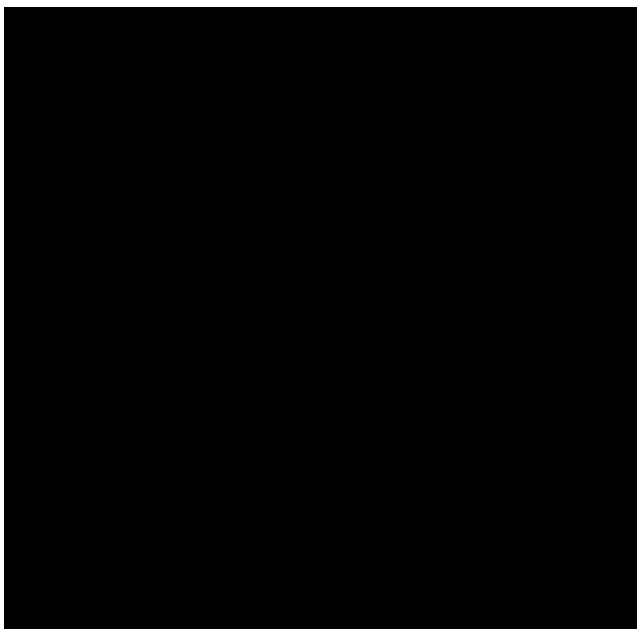


FIGURE 3. Enchondromas seen at base of proximal phalanx of thumb, middle and proximal phalanx of index finger. Phleboliths are also seen scattered all over.

tend to involve one side of the body more than the other,¹ but the incidence of progressive deformity and malignant transformation is greater in MS.³ Maffucci first reported this condition in 1881,⁴ although possible cases have been known to exist since 1835. This rare entity has also been termed Kast's syndrome,⁴ and until 1965 only 56 cases had been reported.⁵ Since then, only a few more cases have been reported.^{2,5-9}

Both sexes are equally affected in MS, and the individuals are usually normal at birth. The lesions develop progressively after five years of age and by puberty, they are invariably evident.^{3,4} The bony lesions predominantly involve the short tubular bones which are usually severely affected, with those of the hand being the preferred site.

Soft tissue involvement is usually limited to the subcutaneous tissue and is mostly a cavernous hemangioma, which typically harbors multiple phleboliths. These hemangiomas overlie normal as well as abnormal bones. The bony and soft tissue lesions eventually lead to a grotesque appearance of the affected limb. As in our case, internal manifestations are uncommon,⁴ despite isolated reports of coexisting visceral hemangiomas.⁸ Plain roentgenographic findings are characteristic.³ Computed tomography and magnetic resonance imaging are helpful in detecting the extent of involvement when malignant degeneration has occurred or is suspected.⁹ The management is surgical and depends on the size and location of the lesions. Severe affection usually leads to amputation.

The lesions of MS have a marked propensity to undergo malignant changes. This is in contrast to single endochroma of tubular bones, which rarely become malignant. Sudden increase in size and the presence of pain in the absence of a fracture should arouse the suspicion of malignancy. The incidence of malignant transformation after the age of 40 years is more than 25%.³ Seven secondary chondrosarcomas and four other malignancies have been reported by Johnson et al.¹⁰ In Berlin's series,⁵ 8 of the 56 cases died of malignancy. Schwartz et al.⁸ mentioned that malignant degeneration is almost a certainty. In his series of seven cases, four patients had chondrosarcomas and one osteosarcoma; none died of skeletal sarcoma. There were, however, no reports of any malignancy in younger age groups. Besides secondary chondrosarcoma, other malignancies which have been reported in MS include angiosarcoma, pancreatic adenocarcinoma and ovarian teratoma.³ Periodic examination is thus essential for the early detection of malignant degeneration.

In our case, the predominantly disproportionate soft tissue abnormality (hemangioma with phleboliths) almost completely masked the subtle osseous lesions which were limited to only three tubular bones of the hand. Such inconspicuous osseous lesions have not been previously reported in MS. This is also contrary to previous reports which mentioned that the vascular component was not a predominant feature of MS.¹¹ Bender and Yunis⁶ reported a case of MS where, instead of hyaline cartilage, fibrocartilage was present in a patient with soft tissue hemangioma and lipoma. Loewinger et al.² reported a patient who had lymphangiomatosis at birth, and subsequently developed hemangioma with osteochondromas. They suggested that MS represented a mesenchymal dysplasia. From the aforementioned, it is likely that MS represents a type of mesenchymal dyschondroplasia with associated dyschondroplasia.

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