

Maffucci's Syndrome –a Case Report of Rare Connective Tissue Disorder

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ABSTRACT

Maffucci's syndrome comprises of multiple venous malformations in combination with dyschondroplasia and a variety of cutaneous, vascular, chondro-osseous and other benign and malignant mesodermal growths. This report describes the case of a 20 -year-male patient with a clinical diagnosis of Maffucci syndrome with multiple lobulated soft tissue swelling with phleboliths in left upper and lower limbs. Significant findings were presence of multiple haemangiomas, enchondromas and pathological fractures- those were evaluated by X-ray, skeletal scintigraphy and ultrasound. Skeletal scintigraphy play an important role to see the extent of the disease. Early detection and surgical management of these tumors form the basis of its treatment and desirable outcome.

Key words: Maffucci's syndrome, hemangiomas, enchondromas.

INTRODUCTION

Maffucci's syndrome is a nonhereditary syndrome characterized by early development of enchondromas and various soft tissue tumors, predominantly hemangiomas. It occurs in all races with no sex predominance. A mutation in PTH/PTHrP type 1 receptor was suggested to be the cause of enchondromatosis (1) but the same has not been confirmed in a recent study (2) enchondromas and hemangiomas can occur anywhere but hands are most commonly involved. Long bone involvement is common, resulting in progressive deformity and pathologic fractures (3). A large variety of benign and malignant mesodermal tumors have been reported in this syndrome, chondrosarcomas being the commonest, occurring in 30% of cases (4). Early detection and surgical management of these tumors form the basis of its treatment and desirable outcome. Here, a case of Maffucci's syndrome is presented

CASE REPORT

A patient 20 years male from Barisal came to Sir Salimullah Medical College & Hospital (SSMCH) with complaints of multiple swellings in upper and lower limbs for at least 10 years. There was no history of any seizure, bloody stool or any visual complain. There was no history of any such illness in the family. On examination of skin he had multiple swelling over the left upper and lower limbs which were mostly on distal ends, variable in sizes and consistency ranging from soft to firm to hard, some swelling were compressible and reducible while other were not (Figure 1). On examination, skin was bluish tinged at some sites and normal at other areas. No local tenderness was noticed. Examination of his chest, abdomen and central nervous system did not reveal any abnormality. Radiologically multiple lobulated soft tissue swellings with phleboliths (haemangiomas) noted in the ankle and dorsum of foot. Bony expansile (lytic type) lesions noted in the shaft of the proximal phalanx of 2nd toe (enchondromas) (Figure 1 (a); (d)). Multiple fractures are noted in 3rd, 5th metatarsal bones. On Ultrasound scan of swellings in upper and lower limbs were oval shaped echogenic soft tissue mass like lesions. On color Doppler study both arterial and venous flow seen with low resistance and increased diastolic flow in arterial spectrum (dominant arterial pattern; RI – 0.6, PI-1.4, PSV--18 cm /Sec, EDV-7 cm/sec). Bone scan by 99m Technetium Methylene diphosphonate (99mTc MDP) revealed increased radio tracer uptake in upper shaft of right humerus, lower shaft of right radius, 1st , 2nd 3rd proximal phalanges, lower shaft of left radius, 4th 5th phalanges of left hand, upper shaft of both humerus, lower shaft of both femur, upper shaft of

right tibia, mid shaft of left tibia, lower shaft of both tibia including both ankle joint and tarsal bones of both foot (Figure 2). Few of soft tissue nodules were surgically excised and biopsied. On histological examination these consisted of vascular channels lined with endothelium having intraluminal red blood cells. He was asked to report any rapid increase in size of any further lesions for an early management.



Figure 1(a)



Figure 1(b)



Figure 1(c)



Figure 1(d)

Figure 1 (a): X-ray left foot shows multiple soft tissue swellings with phlebolith (haemangiomas), Figure 1(b): Soft tissue swelling in distal phalanx of left hand, Figure (c): Soft tissue swellings in of fingers in both hands, Figure1(d): X- rays left hand shows multiple expansile lytic lesions (enchondromas) in distal phalanx of fingers.

DISCUSSION

Maffucci's syndrome comprises the association of cutaneous venous malformation with dyschondroplasia. It occurs in all races and there is no sex preponderance. Familial occurrence is not established though occasional reports of disease among the siblings have been reported. Enchondromas are usually in close proximity to, or in continuity with the growth-plate cartilage. Consequently, they may result from abnormal regulation of proliferation and terminal differentiation of chondrocytes in the adjoining growth plate. A mutant PTH/PTHrP type I receptor (PTHR1) has

been detected in human enchondromatosis, that signals abnormally in vitro and causes enchondroma-like lesions in transgenic mice (1). The same has not been confirmed in another study (2). The individuals are generally of normal appearance at birth but multiple cutaneous vascular swellings begin to appear during early infancy. The disease develops slowly, with enlargement of enchondromas and hemangiomas occurring during the first 2 decades of life (5). In the reported case the male person was of 22 years old and history of gradual limb swelling was for last 10 years. At birth he had no complaints taken from parents. The history of the patient was consistent with above mentioned criteria. The skin lesions, which start as soft, bluish, and occasionally tender swellings, show no tendency to regress and grow in proportion with the child. Grotesque masses may grow on the hands and feet. Cavernous lymphangiomas are also seen. Besides these lesions patient develops hard nodules arising from bone. These may arise in fingers, toes or metaphysis of long bones. Enchondromas appear translucent radiologically. The growth of affected bone is delayed and distorted due to retarded growth of the epiphyseal cartilage. The hands and feet may convert into chondromatous masses and may suffer pathological fractures. A variety of other benign and malignant tumours have been reported. Chondrosarcomas occurring in 30% of patients is the commonest. Other malignant mesodermal tumours reported are fibrosarcomas, angiosarcomas, lymphangiosarcomas and osteosarcomas (6). Clinical examination findings, ultrasound imaging, and X-ray findings suggested presence of hemangiomas, endochordoma like lesions in mentioned limbs of the patient and helped in diagnosis of Maffucci's syndrome. Bone scintigraphy is useful to see the extent of the disease (7) as found in the above mentioned case.

CONCLUSION

Clinical presentation, X-ray, and ultrasound imaging are helpful in diagnosis of Maffucci's syndrome. Bone scintigraphy plays an important role to observe the extent of disease surveying the whole body.

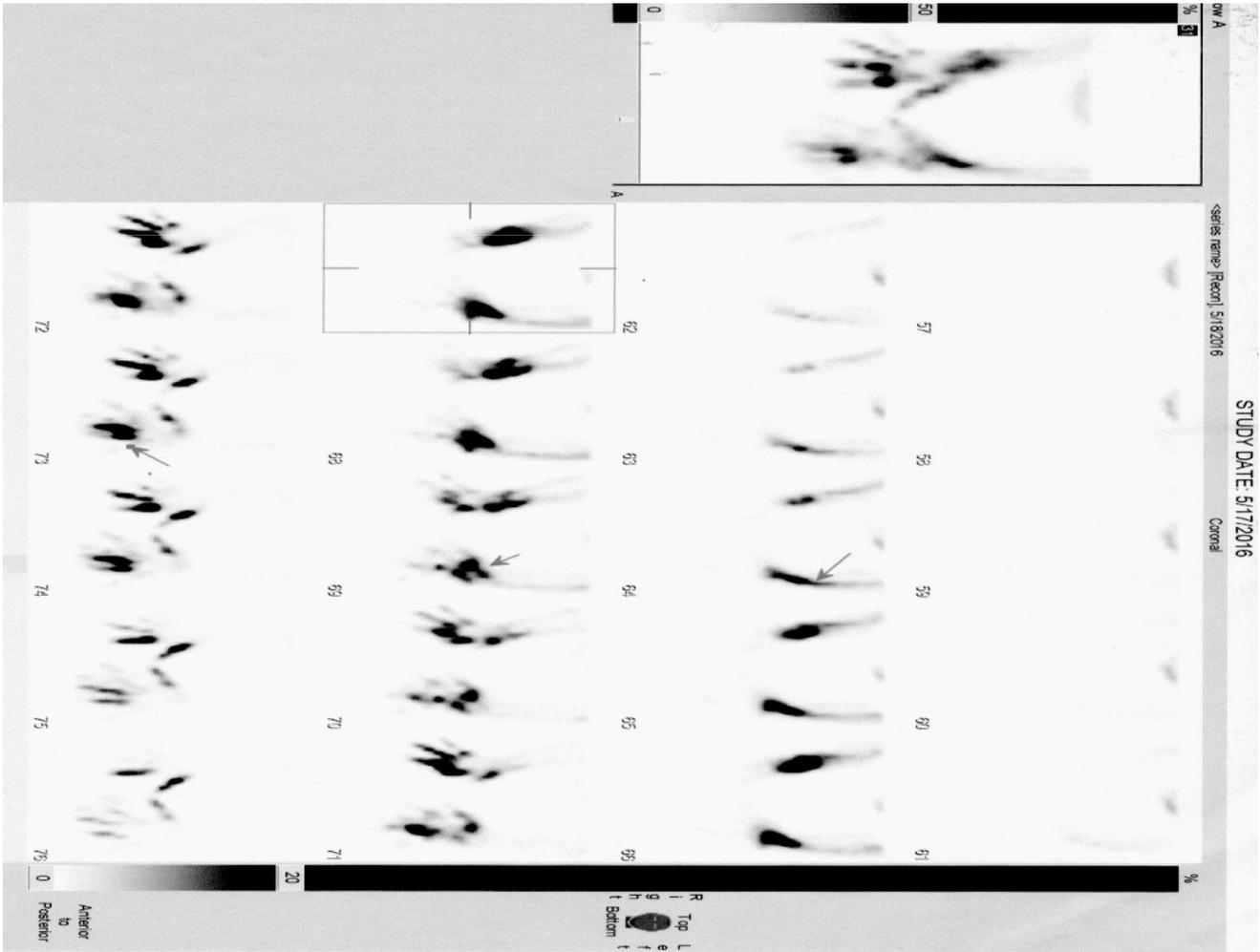


Figure 2: 99m Tc MDP bone scan shows abnormal increased uptake of radio tracer in long bones shaft and distal phalanges of toes and fingers

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