

Case Report

Maffucci's syndrome: a case report

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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. Typically these tumors present in early childhood and may lead to significant deformity. We report a 21-year-old young man with 8 years history of multiple soft as well as firm nodules over his feet and shins, which proved to be hemangiomas and enchondromas on radiology and histopathology respectively. The tumors are important to recognize because of their high malignant potential.

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 enchondromatosis¹ 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 follow up.

Case report

A 21-year-old young man presented with 8 years history of progressively enlarging multiple soft to firm skin-coloured nodules over both his feet. Some of them were painful on walking. Along with this he had noticed progressive lateral tilting of his spine. 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 firm skin colored and soft bluish nodules over both his feet (**Figure 1**). A couple of soft lesions showed black-coloured crusts over them. He had prominent scoliosis, although there was no spinal or paraspinal swelling. Besides, there was a large café-au-lait macule over his trunk and a small angiomatous lesion over his right abdominal flank. Examination of his chest, abdomen and central nervous system did not reveal any abnormality. Radiologically, there were multiloculated

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Figure 1 Multiple, firm, soft, skin-colored to bluish nodules over both feet.

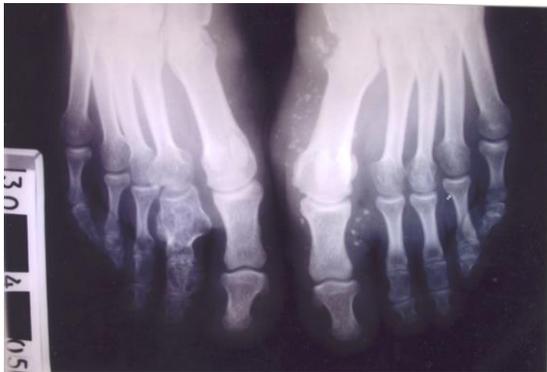


Figure 2 Multiloculated, septate lytic lesions in both proximal and middle phalanges of right 2nd toe.

septate lytic lesions in both proximal and middle phalanges of 2nd toe on right side (**Figure 2**) along with expansion of bones. However, cortex was intact suggesting enchondromas. Moreover multiple soft tissue swellings were noted in both feet especially along medial aspect of left foot and ankle containing speckled calcification suggesting hemangiomas with phleboliths. Further, skeletal survey including x-rays of chest, skull and spine did not reveal any abnormality. Considering the possibility of Maffucci's syndrome two of his enlarging

painful soft bluish lesions 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.

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 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.⁶

Cavernous lymphangiomas are also seen. These vascular lesions have also been reported in gastrointestinal tract.⁷ Besides these lesions patient develops hard nodules arising from bone. These may arise in fingers, toes or metaphysis of long bones. Pathologically, these are enchondromas, which are radiologically translucent. 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 tumors reported are fibrosarcomas, angiosarcomas, lymphangiosarcomas and osteosarcomas. Besides, a number of benign and malignant ovarian tumours have been reported. There is also a likelihood of developing neuro-ophthalmological tumors including astrocytomas and gliomas.⁸ Hematological malignancies including acute lymphatic leukemia have also been reported.⁹

Ollier's disease, which constitutes dyschondroplasia without the cutaneous vascular malformations, is thought to belong to the spectrum of same disease process. Reports of internal vascular anomalies support this theory. Moreover, malignancy is not a feature in Ollier's disease, except for the sarcomatous change in enchondromas.¹⁰

Conclusion

It may be concluded that a high incidence of malignancy in these tumors requires a high index of suspicion for early management.

Other family members must also be checked for any suspected tumors because of unconfirmed reports of a familial incidence.

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