

Case Report

Maffucci's Syndrome - first case report from Pakistan

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Introduction :

Maffucci's syndrome is characterized by multiple enchondromata, cutaneous hemangiomata and more recently has been associated with spindle cell hemangioendothelioma.¹ It is a rare disease. The first case of this syndrome was described by Maffucci in 1881 and eponym, Maffucci's syndrome was later proposed by Carlton et al.² Approximately 170 cases have been reported in literature with 40% being published within the last twenty years.³ We report the first case from Pakistan who required amputation of the left hand due to massive tumors and pain. The clinical, histopathological and radiological features are discussed along with the complications of this rare condition.

Case Report

A thirty years old female presented in outpatient department with multiple swellings on upper and lower extremities. These swellings appeared fifteen years back, when she was fifteen years of age. The swellings were soft, asymptomatic and developed from the fingers of left hand followed by appearance of painful bony hard swellings on the wrist. Both soft and hard swellings progressed with age along with appearance of similar swellings on left elbow, left ankle and feet and toes of the right foot. There were no associated systemic complaints. The patient was single with no family history of similar disease.

Examination revealed multiple hard and soft nodules on left elbow, left ankle and foot and the toes of the right foot. They were ten to fifteen in number, of variable sizes, ovoid in shape and smooth surfaced. Some of the nodules were hard and whitish in colour and non fluctuant and tender on palpation, while other nodules were soft, bluish, fluctuant and non tender (Figure 1). Both type of nodules were asymmetrical in distribution and not fixed to overlying skin. Systemic examination was unremarkable. Above wrist amputation of the left hand was performed.

The roentgenogram of the foot (Figure 2) showed multiple rounded areas of decreased density in the bones with thick trabeculae suggestive of enchondromas. Small rounded areas of soft tissue calcification are seen on the medial side of foot representing phleboliths in the hemangiomas (arrow). Biopsy of the soft cutaneous nodules was done and histopathology (Figure 3) revealed thick and thin walled dermal and subcutaneous vascular spaces with a single endothelial lining. The findings were consistent with vascular malformation. Histopathology of the hard nodules (Figure 4) revealed mature lobules of hyaline cartilage in which foci of myxoid degeneration, calcification and endochondral ossification was seen. Findings were consistent with enchondromas.

On the basis of clinical features and histopathological appearance, a diagnosis of Maffucci's syndrome was made. The small vascular lesions were excised and the patient was informed about the risk of malignancy and the need for a regular follow-up.

Discussion

Maffucci's syndrome does not have a familial pattern of inheritance. The symptoms may begin early in life, at an average age of 4 to 5 years with 25% being congenital. There is no racial or gender predisposition and fertility is not affected. The patients are typically of normal intellect and majority of them can live a reasonably normal life³, but some patients have to endure years of orthopedic complications. The prognosis is grim in some patients when either skeletal or extra skeletal malignancy develops by the end of the fourth decade.⁴

Enchondromas represent an abnormality in the development of the cartilage (dyschondroplasia).³ As bone grows and lengthens, cartilage is left behind and grows irregularly to form expanded cartilaginous tumors (enchondromas).⁵ Clinically, these tumors appear as hard nodules arising from bone and on radiography as multiple irregular cystic lucencies in the growth region. Enchondromas are most frequent on hands (88%) followed by tibia/fibula (56%), foot (51%), femur (47%), humerus (39%), radius/ulna (37%), ribs (30%), pelvis (23%) and head (12%).⁶ They are also reported intracranially at tuberculum sella⁷ and cerebello-pontine angle.⁸ Irregular expansion of enchondromas may prevent normal skeletal growths which cause deformities like short stature, scoliosis, leg-length discrepancy and pathological fractures with delayed union or healing with significant deformity.³

The hemangiomas in Maffucci's syndrome appear as reddish blue, soft, compressible, occasionally tender, subcutaneous nodules that grow proportional to body growth and have no tendency to resolve. The distribution of hemangiomas is likely to be much more extensive on one side of the body. They are most commonly located on hands and feet, 57% and 41% respectively.³ One third of the patients have hemangiomas on arms, legs or trunk. They can essentially occur anywhere and have been reported in the leptomeninges, eyes, pulmonary tree, throughout gastrointestinal tract from the oral to anal mucosa, cervical spine, orbital region and mediastinum. Histologically the hemangiomas are often of the complex venous type but, capillary, and mixed venous and capillary type can occur. They are located in the mid to lower dermis and subcutaneous fat. In the dilated vessels and vascular spaces, there is a tendency for thrombosis and subsequent calcification (Phleboliths) which can be identified by radiography³ as in our patient.

Lymphangiomas represent another rare aspect of the mesodermal dysplasia in Maffucci's syndrome. The existence of lymphangiomas without hemangioma has been proposed as a sub form of Maffucci's syndrome.⁹

Although, Maffucci syndrome has been considered as a generalized mesodermal dysplasia, malignant and benign tumors of both mesodermal and non-mesodermal origin has been associated with it.³ An overall malignancy rate of 37%⁶ has been reported. By far the commonest tumor is chondrosarcoma which occur in 30% of patients⁶ and is reported at unusual sites like cranial base¹⁰ and nasal septum.¹¹ The average age of development of chondrosarcoma is 40 years but the age has ranged from 13 to 69 years. A majority of chondrosarcomas are of low histologic grade and can often be treated with adequate surgical resection. Other malignant neoplasms include astrocytoma¹², ovarian tumors¹³, pancreatic carcinoma³, hemangiosarcoma¹⁴, lymphangiosarcoma¹⁵, acute myelocytic leukemia, fibrosarcoma, osteosarcoma, angiosarcoma, hepatic adenocarcinoma, and acute lymphoid leukemias.³

Benign tumors associated with Maffucci's syndrome include pituitary adenoma, adrenal cortical adenoma, parathyroid adenoma, breast fibroadenoma, thyroid adenoma, ovarian thecoma, uterine fibroid, and testicular mesothelioma.³ Once a neoplasm develops in a patient, the likelihood of developing another neoplasm is increased.³

The condition has to be differentiated from Ollier's disease in which there are dyschondroplasias without the cutaneous malformation seen in Maffucci's syndrome and Gorham's disease in which bone is replaced by vascular malformation, generally of venous type, and there may be associated cutaneous and soft tissue vascular malformations.

There is no treatment to slow the progression of disease. Therefore, management consists mainly of symptomatic relief and regular follow-up of these patients to assess for development of these com-

plications.

We report the first case of Maffuci's syndrome from Pakistan. Our patient required amputation of hand due to exuberant tumors and pain illustrating the need for regular orthopedic follow up and intervention in these cases.

References

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