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MAFFUCCI SYNDROME WITH CHONDROSARCOMA-A CASE REPORT

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ABSTRACT

Maffucci syndrome is a congenital, non-hereditary mesodermal dysplasia manifested by multiple enchondromas and hemangiomas. Chondrosarcomatous transformation occurs in approximately 30% to 40% of enchondromas. We report a case of a 52 year old female presented with multiple swellings in the hand and a huge swelling in the scapular area. Radiology, cytological and histopathological examination was done. The soft tissue swellings in the hand was found to be hemangioma. Multiple lytic lesions in the hand bones shows picture of enchondroma. The scapular swelling was found to be chondrosarcoma. The case was diagnosed as Maffucci syndrome with chondrosarcoma.

KEYWORDS Enchondroma, haemangioma, chondrosarcoma, Maffucci syndrome

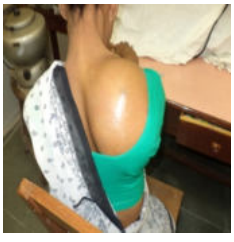
INTRODUCTION

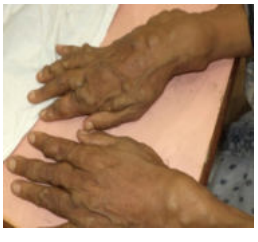
Maffucci syndrome, a congenital mesodermal dysplasia is characterized by multiple enchondromas and hemangiomas and was first described in 1881.[1]Approximately 170 cases have been reported in literature with 40% being published within the last twenty years.[2] The sexes are equally affected. The localization and extent of skeletal involvement varies greatly among individuals, ranging from cases limited to multifocal involvement of a single bone to cases with widespread lesions and crippling deformation. The hand is the most common site. Other common sites are foot, femur, humerus, and forearm bones. In severe cases, the flat bones are also affected. Frequently, the disease is limited to a single extremity or to one side of the body. Many cases, however, have bilateral involvement. Malignant transformation occurs in 25-30% of cases, usually as low grade chondrosarcoma. However, high grade sarcomas, such as osteosarcoma or dedifferentiated chondrosarcoma can also occur.[3]Treatment should aim at symptom relief and early detection of malignancies; no therapy is indicated for asymptomatic patients.[1]

CASE STUDY

A 52 year old female presented in the outpatient department of orthopedics with multiple swellings in the hand (Figure 1) and a huge swelling in the right scapular area (Figure 2).

The hand swellings appeared when the patient was 25 years of age. The swelling were not associated with pain, non-progressive in nature and were asymptomatic. The scapular swelling appeared 3





months before and was gradually increasing in size. It was associated with pain and heaviness. The patient gives history of recent weight loss with decrease in appetite. There was no family history of similar disease. Examination of the multiple hand swellings was done and it revealed that there were two types of swelling. One type was soft, non-tender and fluctuant and the other one was hard, tender on palpation and non-fluctuant in nature. Overlying skin was normal. There was a huge scapular swelling which was firm in consistency, mild tender on palpation and the overlying skin was stretched. Systematic examination was unremarkable.

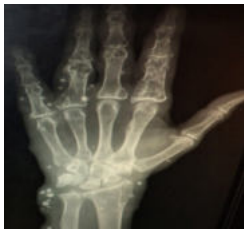
Figure 1: Multiple hand swelling.

Figure 2: Swelling in the scapula.

X ray of multiple swellings in the hand showed radiolucent areas with thickened trabeculae suggestive of enchondroma. Soft tissue calcification was seen in the soft swellings suggestive of phleboliths in hemangioma (Figure 3). Aspiration cytology from the hard hand swellings showed cartilage cells with abundant myxoid ground substance. A diagnosis of enchondroma was rendered. Histopathology confirmed the diagnosis.

Figure 3: X Ray showing osteolytic lesions in phalynx with soft tissue calcification.

Radiology of the scapular swelling showed a large space occupying lesion in scapula with multiple calcification and soft tissue extension (Figure 4).



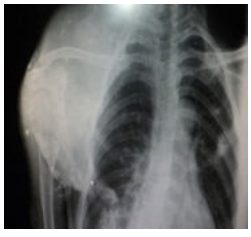
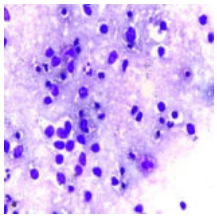


Figure 4: X Ray showing space occupying lesion in scapula with soft tissue extension.

Aspiration from scapular swelling showed enlarged bizarre mononuclear and binucleate cells with finely vacuolated cytoplasm with scant chondromyxoid material. It was diagnosed as chondrosarcoma (Figure 5). Histopathology was done to confirm the diagnosis (Figure 6).



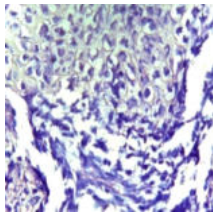


Figure 5: Aspiration from scapular swelling.

Figure 6: Histopathology from scapular swelling.

On the basis of clinical features, radiology, cytological and histopathological examination, a diagnosis of Maffucci syndrome was made.

DISCUSSION

Maffucci syndrome is a nonhereditary developmental disorder characterized by the occurrence of multiple cartilaginous masses, particularly affecting the soft and long tubular bones of the limbs with cutaneous, soft tissue and visceral hemangiomas.[3] It occurs in all races with no sex predominance. Enchondromas and hemangiomas can occur anywhere but hands are most commonly involved. Long bone involvement is common, resulting in progressive deformity and pathologic 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.[4] There is also a likelihood of developing neuro-opthalmological tumors including astrocytomas and gliomas.[5] Hematological malignancies including acute lymphatic leukemia have also been reported.[6]Early detection and surgical management of these tumors form the basis of its treatment and follow up.

CONCLUSIONS

Maffucci syndrome is a rare congenital, non-hereditary mesodermal dysplasia manifested by multiple enchondromas and hemangiomas.[7] Chondrosarcomatous transformation occurs in approximately 30% to 40% of enchondromas.[8]Bone and soft tissue lesions that grow or become painful without a history of trauma should be examined for malignancy and biopsied.

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