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Conservative management approach in Maffucci syndrome: A case series

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Abstract
Maffucci syndrome is a rare, non-hereditary condition characterized by multiple enchondromas, soft tissue hemangiomas and skeletal abnormalities. It is associated with diverse secondary musculoskeletal deformities, which are exceedingly rare. Patients with this syndrome demonstrate a normal intellect and generally live a productive life. Rarely, it presents with fracture of long bones, where conservative modality is better, surgical treatment is unsafe due to risk of bleeding. We have reported 4 cases of maffucci syndrome over 6 years duration at 4 institutes in central India from 2015 to 2021. Some are associated with bony fractures, and managed conservatively (6 months follow up). Careful surveillance for malignant degeneration of both skeletal and non-skeletal tumors, especially in the brain and abdomen is essential [1].

Keywords: Maffucci syndrome, enchondroma, fracture, conservative management

Introduction
Maffucci syndrome is a rare clinical diagnosis characterized by multiple enchondromas and hemangiomas. It was first described in 1881 [1]. It consists of combination of multiple enchondromas and vascular tumors [2, 5, 6]. In 78% of the cases, the symptoms start before puberty [2]. Enchondromas are benign cartilaginous tumors that may occur at any site, but most commonly is seen in phalanges and long bones. Patients with this syndrome demonstrate a normal intellect and generally live a productive life. Complications include spontaneous fracture through area of advanced rarefaction in 26% and sarcomatous degeneration of enchondroma in 15-30% [1]. Chondrosarcomatous transformation occurs in approximately 30% to 40% of enchondromas [5]. Most frequent location of hemangiomas is in the dermis and subcutaneous fat adjacent to areas of enchondromatosis or elsewhere. In this case series, we present 4 patients of maffucci syndrome over 6 years duration at 4 institutes in central India-SSMC (Rewa), Medanta (Gurgaon), ESI Hospital (Okhla-New Delhi) & NSCB medical college (Jabalpur). All of them have been managed with conservative modality of treatment without any surgical means.

Materials and Methods
Case 1: A 38 years female presented with multiple soft compressible swelling on left feet, left thigh involving knee joint for 10 years, with difficulty in walking for 3 year. Diagnosed with multiple enchondroma with no fractures on x-rays.

Case 2: A 20 years male presented with complaints of weakness, fatigue, multiple compressible tender and irregular swelling in back, right forearm, hand and right chest wall. Diagnosed with multiple enchondroma with no fractures on x-rays.

Case 3: A 16 year old male reported to us following history of fall on ground with severe pain and swelling in the left lower thigh with inability to bear weight. Physical examination revealed severe tenderness and irregular soft tissue swelling in thigh with numerous non-tender immobile nodules in the thigh.

Joint movements (flexion and extension) at the knee joint was significantly restricted with pain & spasm. Radiographs of left hip with thigh and knee showed fracture distal femur with multiple well-defined, irregular and radiolucent lesions corresponding to intra-osseous benign cartilaginous tumors or enchondromas with associated with soft tissue swelling. (Figure 1 a,b)

Case 4: A 40 year old female presented with complaints of bone pain, multiple compressible and tender swellings in right lower limb, right forearm, hand and right upper back. Diagnosis of multiple enchondroma was established with no fractures on xrays.

All the patients underwent conservative management in the form of rest, analgesics with traction splint. Bohler braun splint with calcaneal skeletal traction with 2 kg weight was used in patient with distal femur fracture (figure 1c). Physiotherapy was started as required after radiological & clinical union of fracture.

All patients had regular follow-up for 6 months. Serial x-rays were taken and examined clinically at follow up. Protective weight bearing was started in patient with fracture femur after 6 weeks and converted to complete weight bearing in 6 months.

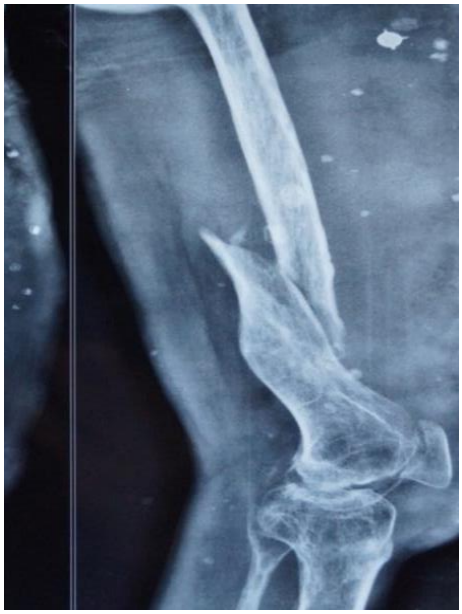

Fig 1A: Radiograph of patient with distal femur fracture at the time of admission

Fig 1B: Bohler braun splint with skeletal traction (to align the fragments)

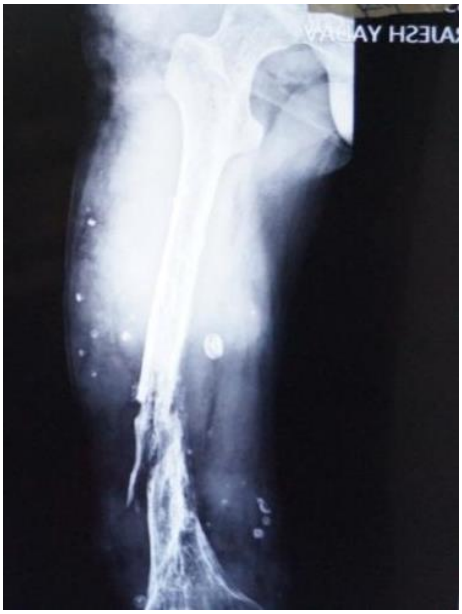

Fig 1C: Follow up at 3 weeks

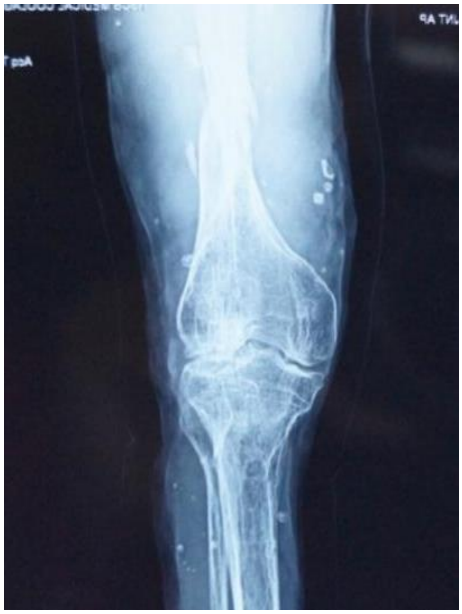

Fig 1D: follow up at 6 months

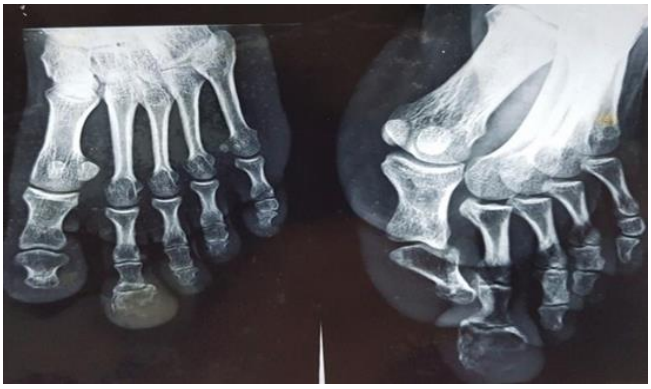

Fig 2: enchondroma of left foot distal phalynx great toe & 2nd toe causing hallux valgus

Discussion

Maffucci's syndrome comprises the association of cutaneous venous malformations with dyschondroplasia. Enchondromas exists not only most frequently at the small bones of the hands and feet, the long tubular bones, but also the flat bones, such as pelvis [1]. They are commonly in close proximity to or in continuity with growth plate cartilage. They might be the consequence of abnormal regulation of proliferation and terminal differentiation of chondrocytes in the adjoining growth plate. The osseous lesions most frequently involves the phalanges, metacarpals and metatarsals. Maffucci syndrome might be associated with three types of vascular lesions: cavernous hemangiomas, phlebectasias and lymphangiectasias-lymphangiomas [2, 5].

The tumors appear as nodular outgrowth and can cause a fracture leading to further complication such as shortened or unequal limb length [1, 6].

It occurs in all the races without any sex preponderance. Familial occurrence has not been established though occasional reports of disease among the siblings have been reported. Chondrosarcoma is the most common neoplasm (25-30%) associated with this syndrome & therefore an early diagnosis is crucial [4].

Long bone fracture in maffucci syndrome is rare and modality for treatment for that is

1. **Conservative:** With rest, high protein diet, skeletal traction, physiotherapy and regular physical examination in order to evaluate changes in the skin and the bone lesions that may suggest malignancy.
2. **Surgical:** Hemangioma can be treated with sclerotherapy, laser therapy, irradiation and open surgery followed by fracture fixation by plating, nailing or external fixation, corrective osteotomy and lengthening of the limb can also be done, but due to blood loss and poor bone quality it is rarely indicated. Curettage and bone packing with bone graft material is another surgical method [1]. Sometimes amputation could be indicated in malignancy (chondrosarcoma).

If absence of clinical features or major signs on examination, no treatment is needed [8, 9] Surgery is indicated only in the case with complications, such as pathological fractures, and malignant transformation [8, 9]. The purpose of surgery is to remove the tumor mass and make histological diagnosis. If necessary, chondrosarcoma needs adjunctive therapies[8], including sclerotherapy, irradiation and laser therapy. In our case series, the patients were managed conservatively and followed very closely to look for any malignant changes[10] and other possible complications. Therefore, it was an appropriate modality of treatment.

All enchondromas and hemangiomas were located in multiple areas which were asymptomatic and unrelated with each other. X-ray showed irregularly shaped, radiolucent areas with stippled calcification & phlebolith in soft tissue[1]. In view of large hemangioma present in left thigh of 3rd patient, surgical management was not found to be a suitable mode of treatment (due to the high risk of bleeding) and attempts were taken to manage it conservatively.

Conclusion

In case of large hemangioma with long bone fracture, conservative management is the better modality of treatment. It not only leads to fracture union but can correct or minimize deformities as well. Risk of bleeding can be avoided which could be life threatening if surgical intervention is taken. The key point of follow-up is to master the progress of multiple hemangiomas and malignancy [3], if such changes are seen then biopsy is indicated to make pathologic conclusion.

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