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INTERNATIONAL JOURNAL OF ADVANCED RESEARCH (IJAR)

Article DOI:10.21474/IJAR01/18408
DOI URL: http://dx.doi.org/10.21474/IJAR01/18408

RESEARCH ARTICLE

MAFFUCCI SYNDROME, UNILATERAL LOWER LIMB INVOLVEMENT WITH GROSS SHORTENING: A CASE REPORT

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Manuscript Info

Manuscript History
Received: 10 January 2024
Final Accepted: 14 February 2024
Published: March 2024

Key words:-
Maffucci Syndrome, Multiple Enchondroma, Hemangioma, Deformity, Non-Union, Shortening

Abstract

Maffucci syndrome is a rare type of multiple enchondromatosis associated with hemangiomas in soft tissues. The presence of hemangiomas and a very high risk of malignant transformation of enchondromas to chondrosarcomas differentiates Maffucci syndrome from Ollier's disease. We report a case of 9 years old girl child with multiple enchondromas and hemangiomas. She had shortening and deformity of left lower limb, fracture of left tibia following a trivial trauma, and a history of surgical interventions to correct valgus deformity at the knee by figure of 8 plating and nailing and bone grafting for fracture tibia that failed to unite. X-rays were suggestive of multiple enchondromas affecting the bones unilaterally, of only the left lower limbs with multiple lesions in the left femur, tibia and fibula. The right lower limb was normal. The resultant shortening was 6.5 inches (16.5 centimeters.). There was, in addition, a painful, warty growth in the lower third of the posterior aspect of the left thigh measuring 1.5 cm x 1.5 cm. An excisional biopsy was done and the histopathology report was a Hemangioma. The child was diagnosed as a case of Maffucci syndrome. The child was fitted with a KAFO with an extension prosthesis (a double decker).

Introduction:-

Maffucci syndrome is a multiple enchondromas with hemangiomas [1]. It is a rare disease. About 160 cases are reported in English literature [2]. It is a non-hereditary disorder caused by a mutation in IDH1 and IDH2 genes [3]. Enchondromas are benign cartilaginous intramedullary tumors that develop near to the physis (growth plate) and are located in the metaphyseal region most often seen in the phalanges, femur and tibia with a tendency to unilateral involvement in about 40 percent of cases[4]. Hemangiomas present as cutaneous lesions, rarely visceral, get calcified and often present as phleboliths visible as opacities on x-ray. In a child diagnosed with Maffucci syndrome the immediate challenge is to make the child ambulatory by correction of the deformities, limb length equalization

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and management of pathological fractures. The long term challenge is to detect and manage the malignant transformation that occurs very frequently in Maffucci syndrome[5]. A child with Maffucci syndrome needs lifelong monitoring to detect and treat a malignant degeneration as soon as it occurs. Pain and rapid increase in the tumor size is an indication for needle biopsy.

Case Report:
A 9 years old girl child presented with gross shortening of the left lower limb, inability to bear weight and deformities of the left lower limb. She also complained of a painful growth on the posterior aspect of the lower third of the thigh on the same side. There was a history of fracture of the upper third of the left tibia following a trivial injury about a year back that was treated with nailing and bone grafting but it failed to unite. Attempts at deformity correction were also done earlier and figure of 8 plating was done to correct valgus at the knee before she presented to us.

On examination, the left lower limb was short by 6.5 inches (16.5 centimeters). There were scars from previous surgeries. The upper third tibia had a non-union. The growth on the post aspect of the lower third of the thigh was 1.5 x 1.5 centimeters, bluish, warty and tender. The right lower limb was normal (Figure 1).

X-ray demonstrated multiple enchondromatosis in the metaphyseal regions of the femur, tibia and fibula on the left side only (unilateral involvement). The involved bones were thin with altered curvatures. The cortices at the involved metaphyseal areas were thin. There was a figure of 8 plate on the medial side of lower end of femur and a tibial nail with evidence of failed grafting and atrophic non-union of the fracture at the upper third of left tibia (Figure 2).

The figure of eight plate was removed (Figure 3). The warty growth on the posterior aspect of the thigh was excised and sent for histopathology (Figure 4,5). The report was a hemangioma. The child was fitted with a Knee Ankle Foot Orthosis with an extension prosthesis (a double decker) and was made ambulatory (Figure 6). The non-union of tibia and limb length inequality remains to be managed by subsequent interventions.

Discussion:-

The condition of multiple enchondromas with hemangiomas was first described by Maffucci in 1881[1]. The disease was termed as Maffucci syndrome by Carlton[6]. There is no sex predilection. It can be diagnosed easily on clinical and radiological grounds. However, biopsy and histopathological examination is imperative when a malignant transformation is suspected, especially when there is a sudden increase in the size of tumour and onset of pain. The tumour is juxta physeal and manifests as longitudinal streaks of cartilage that had failed to ossify in the metapyseal region of long bones. It may occasionally involve epiphysis and may also present in flat bones. Peculiarly, it may involve bones unilaterally in about 40 percent of the patients, as in our case[4]. There are very high chances of malignant transformation to Chondrosarcoma in Maffucci's syndrome as compared to Ollier's, as high as 30 percent as reported by Kaplan et al or even as high as almost 100 percent in the lifetime of a patient with Maffucci syndrome as reported by Schwartz et al[5]. The malignancies may occur in other organs and do not necessarily restrict to bones. Hemangiomas may likewise undergo a malignant transformation. They may get calcified and present as phleboliths visible as opacities on x-ray as in our patient[5].

The unique features that made our case peculiar and also difficult to manage were extensive long bone involvement, unilaterality, massive shortening causing a substantial limb length discrepancy, a fracture that failed to unite and a painful hemangioma.

Conclusion:-

Maffucci syndrome though similar to Ollier's disease, both presenting as multiple enchondromatosis, poses unique challenges because of a considerably high risk of malignant transformation, necessitating lifelong monitoring to detect and manage malignancies as and when detected. Another unique feature is that Maffucci's syndrome is comparatively a very rare disease. No treatment is required for minimal bony involvement in Maffucci syndrome. However, patients with extensive long bone involvement are extremely difficult to manage. The problems of limb length inequality, pathological fractures, non-union and malignant transformation pose problems in the management of these patients. The rapid increase in size of tumor and pain would necessitate a biopsy. Similarly any significant

change in a haemangioma is a hint towards malignant transformation. Lifelong monitoring of the patient to rule out any malignant transformation is imperative.

Figures:

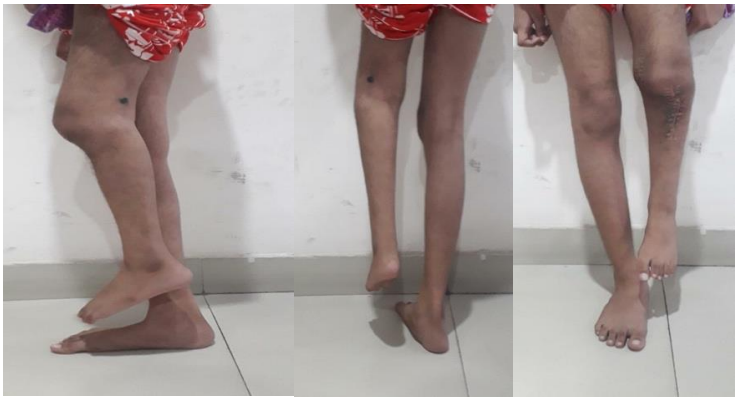

Figure 1:- Shows child at the initial presentation.

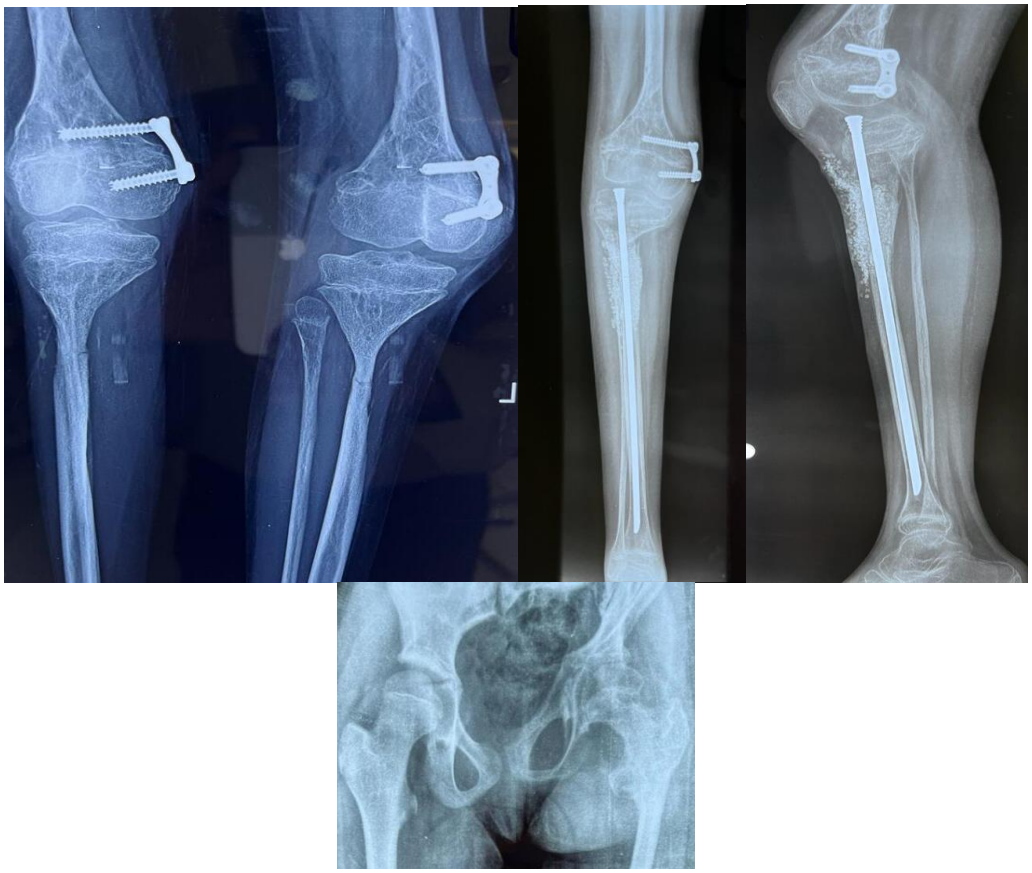

Figure 2:- Shows x-ray at the initial presentation (First x-ray image showing phleboliths (hemangiomas) in the skin.Next two images shows fracture tibia that failed to unite and the last x-ray shows left femoral involvement)

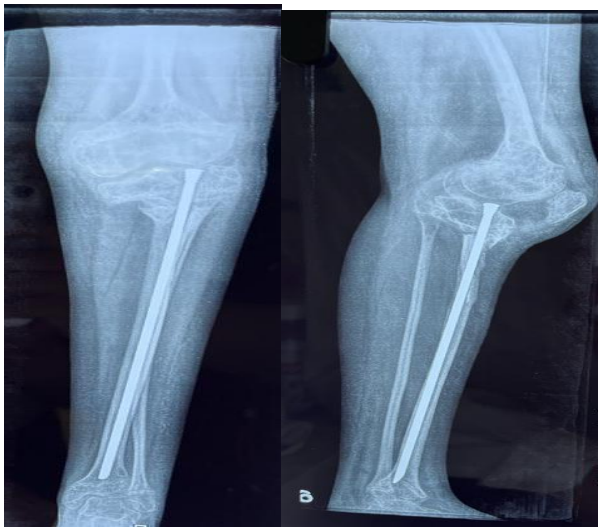

Figure 3:- Shows x-ray after the surgery.

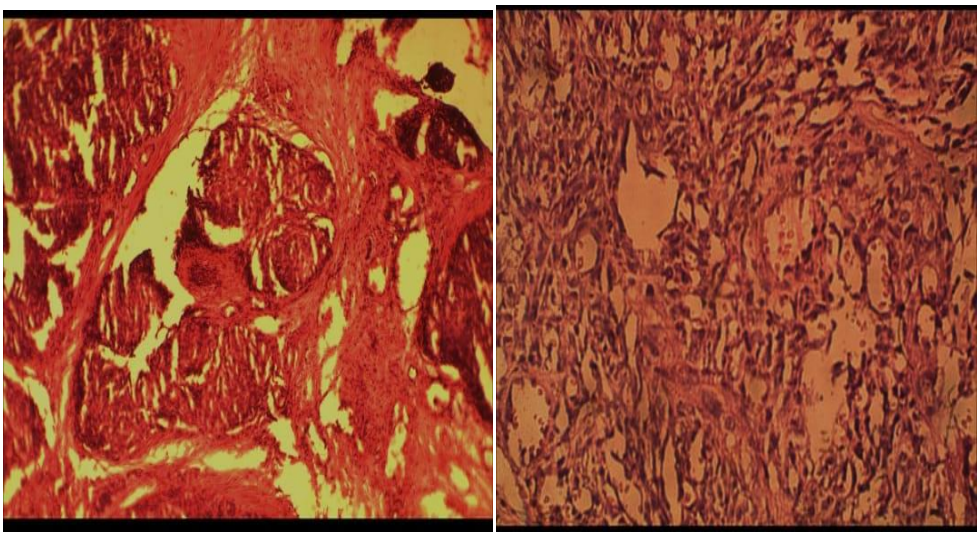

Figure 4:- Shows benign cavernous haemangioma with hemosiderin pigmentation. No malignant pathology seen.

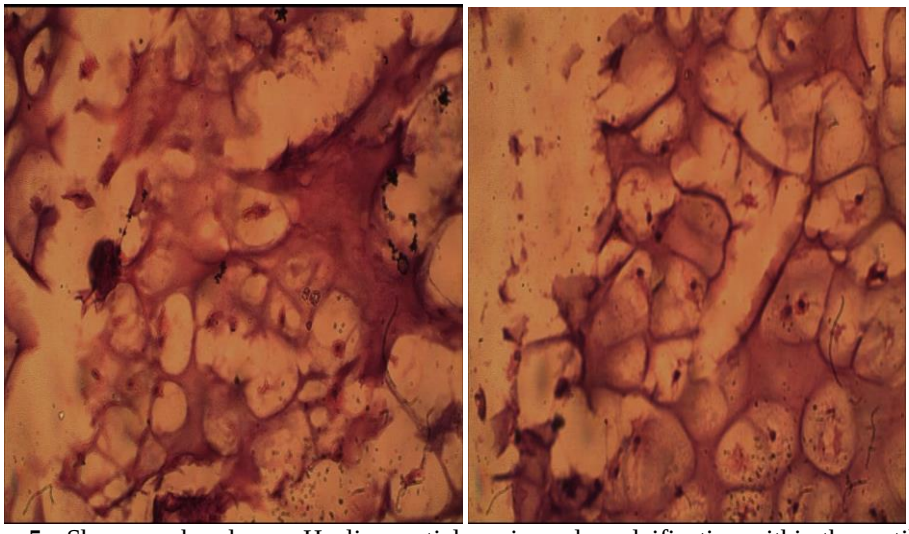

Figure 5:- Shows enchondroma. Hyaline cartialage, irregular calcification within the cartilage.

Figure 6:- Shows clinical picture after applying double decker KAFO.

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