Case Report

Diaphyseal aclasis : study of imaging pattern and associated deformities

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Hereditary multiple exostosis, also known as Diaphyseal aclasis is characterised by development of multiple exostosis. We report two cases of diaphyseal aclasis who presented to the outpatient orthopaedics department of our institution with clinico-radiological correlation of the diagnosis with due emphasis on the associated deformity as encountered. The multiplicity of lesion and associated deformity makes the diagnosis of diaphyseal aclasis at an early stage than its solitary counterpart. Most of the patients presented with short stature as the chief complaint. The spectrum of the radiological features of osteochondroma, its variants and complications are a direct refelction of its pathological appearances. Identification of these features are important in guiding therapy and thus improving the patients management.

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***Key* words : Osteochondroma, hereditary multiple exostosis, diaphyseal aclasis.**

Osteochondroma is the most common benign tumor or tumour-like condition with characteristic radiological features. It may be solitary or multiple with latter being associated with Hereditary multiple exostosis or diaphyseal aclasis. Treatment of hereditary multiple exostosis is complex and is often directed to correct the associated deformities rather than restricted to exostosis alone (Figs 1-5).

Case 1 :

A twenty-five year old male patient presented with chief complaints of short stature and multiple bony swellings around the joints of upper and lower extremity since childhood. There was no history of similar complaints in the family. General and systemic physical examination was normal. On Local examination, multiple hard bony outgrowths fixed to the underlying bone were noted in the region of bilateral wrist joint, bilateral knee and ankle join with deformity at bilateral wrist joint and forearms with shorening of right lower extremity. Skeletal survey of the patient was done which revealed multiple sessile and pedunculated bony outgrowths continuing with the underlying bones at multiple sites. Radiograph of bilateral wrist with forearm revealed sessile bony outgrowths composed of cortical and trabeculated component continuing with the underlying shaft of the forearm bones. There was shortening of the bilateral ulna with outward bowing of radius and subluxation of bilateral radiocarpal joints classically termed as the Bayonet hand deformity. Radiograph of bilateral knee joint in frontal and lateral preojections shows undertubulation of ends of femur termed as Erlenmeyer flask deformity. Multiple pedunculated bony outgrowths from bilateral lower end of femur and upper end of tibia and fibula projecting away from the joint space, a deformity termed as coat hangers exostosis at the distal end femur. Radiograph of bilateral ankle joint, frontal projection showed similar pedunculated exostosis at the distal end of tibia and fibula with pressure erosion over the medial end of distal right fibula with formation of interlocking exostosis. Diagnosis of Diaphyseal aclasis was confirmed on the evaluation of the skeletal survey of the patient which revealed characteristic sessile and pedunculated multiple exostosis with associated deformities. The patient was treated with reconstructive operation for bilateral bayonet deformity.

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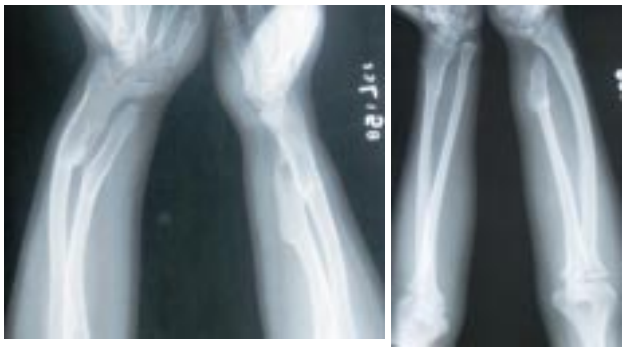

Fig 1(a) Fig 1(b)

Figs 1a & 1b — Radiograph bilateral forearms with wrist joint (Frontal projection):Multiple sessile bony outgrowths continuous with the undelying long bone noted at the lower end of bilateral radius-ulna with shortening of ulna and outward bowing of radius with subluxation of radio-carpal joint ,classically termed as Bayonet deformity

Fig 2 — Radiograph bilateral knee joint, frontal projection: Multiple sessile and pedunculated bony outgrowths consistent with multiple exostosis

Case 2 :

A thirty two year old male patient presented with chief complaints of multiple hard bony swelling around wrist and knee joint, associated with swelling over the back on the left side since childhood. There was no associated pain. Family history of similar swellings in elder brother was also noted. On examination, multiple bony swellings fixed to the underlying bones were noted in the region of wrist, bilateral knee, right shoulder and left scapular region. Radiograph of the right shoulder

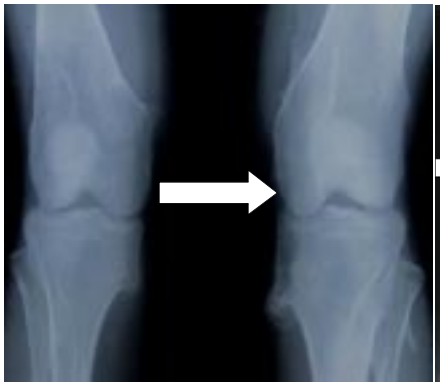

Fig 3 — Radiograph ,bilateral Knee : Undertubulation of lower end of bilateral femur(Erlenmeyer flask deformity) with multiple pedunculated exostosis growing away from the joint space (Coat hangers exostosis)

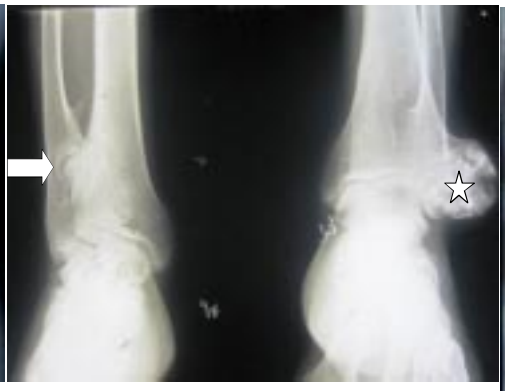

Fig 4 — Radiograph bilateral ankle joint : Multiple pedunculated exostosis noted at with calcified hyaline cap ,Cauliflower exostoses(star).Interlocking exostosis of right sided distal tibia-fibula with pressure erosion on distal medial shaft of right fibula.(arrow)

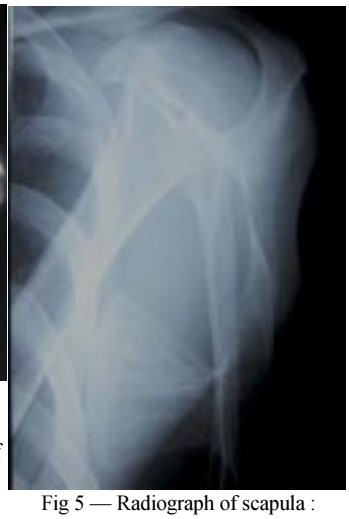

Fig 5 — Radiograph of scapula : Pedunculated exostosis arising from the infero-medial margin of left scapula

and humerus showed multiple sessile bony outgrowths from the proximal half of right humerus. Radiograph of the left scapula revealed similar exostosis arising from the lower medial border of the scapula. Radiograph of bilateral wrist with forearm revealed similar bony outgrowths with right sided bayonet deformity. Radiograph of bilateral knee joints showed multiple bony exostosis with undertubulation of the ends of femur and tibia-fibula. Diagnosis of hereditary multiple exostosis was made. The patient underwent reconstructive operation for the right side bayonet deformity with excision of the associated exostosis under general anaesthesia. Postoperative period was uneventful and physiotherapy was started on 4th postoperative day.

Discussion

Hereditary multiple exostosis, also known as familial osteochondromatosis or diaphyseal aclasis, is characterised by the development of multiple osteochondromas. It has an autosomal dominant inheritance with incomplete penetrance in females. The number of exostosis, the degree and type of deformities and the risk of complications varies significantly even within the family[1].

The genetic basis of HME is on chromosome[8], the locus being EXT-1 and additional locus on chromosome 11 and 19, referred as EXT2 and EXT3[2].

The skeletal distribution of lesions of HME is characteristically described as bilaterally syymmetric involvement with involvement of any bones developing from enchondral ossification with most frequent involvement of bilateral lower extremity in 50% cases.

On imaging, the individual osteochondromas in HME is identical to that of solitary lesions. Carroll *et al* noted that the amount of involvement and deformity of the forearm and distal leg is a measure of the overall disease extent. In addition, they found that the percentage of sessile osteochondromas correlated with the extent of deformities[3].

Cosmetic deformity caused by an osteochondroma is the most common clinical presentation. Bony deformities include both growth sequelae as a result of failure of normal tubulation and local effects such as osseous bowing and malalignment. Extrinsic pressure erosion of an adjacent osteochondroma is commoner with large lesions where paired bones lie juxtaposed. Other complications include Fracture of the osteochondral fragment, Vascular compromise, Neurological sequelae, bursal formation and rarely malignant degeneration, most commonly into chondrosarcoma. The risk of malignant degeneration is about 1% in solitary osteochondroma to upto 25% in HME.

Radiographic features suggestive of malignant degeneration include[4].

(1) Growth of osteochondroma in a skeletally mature patient.

(2) Irregurity of the surface of the lesion.

(3) Focal regions of radiolucency in the interior of lesion

(4) Erosion or destruction of adjacent bone. And

(5) Significant soft tissue mass with irregular calcification.

(6) A cartilage cap thickness of more than 1.5 cm should be considered suspicious for malignant transformation.

Fig 6 — Radiograph of right shoulder with proximal humerus: Multiple sessile exostoses noted arising from proximal humerus

Treatment of patients with HME is complex and controversial. Most of the surgical treatment is directed to correct the associated deformity along with excision of exostosis[5]. Patients with HME require continuous surveillance for the progression of deformity and development of complications. The overall recurrence rate of osteochondromas is estimated to be 2% with most of these related to inadequate excision of the overlying perichondrium.

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