DIAPHYSEAL ACLASIS: A CASE REPORT WITH REVIEW OF LITERATURE

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ABSTRACT

Multiple exostosis (diaphyseal aclasis) is a heritable disorder affecting the endochondral skeleton during the period of growth. It is characterized by thickening and deformity of the growing bone with the formation of numerous cartilage-capped exostoses clustered around the areas of most active growth. Thus, the juxtaepiphyseal regions of the tubular bones, the ribs, pelvis, and scapulae are the most heavily affected areas, while the vertebral bodies, the patellae, and the carpal and tarsal bones are usually unaffected. Hereditary multiple exostosis demonstrates an autosomal dominant inheritance pattern.(1)

A 5-year-old male child presented with a large swelling in his left shoulder. Small swellings were noted in bilateral knee joint, right ankle and also in right humerus. The swelling on the left shoulder was initially small in size which then gradually increased in size. There is a strong family history of similar complaints. Radiographs were performed which showed two well-defined exophytic pedunculated lesions arising from the left scapula along the posterior aspect. On MRI, the lesion shows T2 hyperintensity and T1 hypointensity with cartilage cap covering the entire surface.

KEYWORDS
Hereditary multiple exostosis, Diaphyseal aclasis (Aclasia), Multiple osteochondromas, Familial osteochondromatosis.

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INTRODUCTION: Hereditary multiple exostosis demonstrates an autosomal dominant inheritance pattern. The number of exostosis, the degree and type of angular deformity, and even the rate of malignant transformation vary significantly.(1) Most patients are diagnosed by age 5 years, and virtually all are diagnosed by age 12 years. Patients may be asymptomatic with a few small lesions or may be significantly deformed by multiple large osteochondromas.(2)

CASE REPORT: A 5-year-old male child presented with a large swelling in the left scapular region. Multiple small swellings were also present around both the knee joints, right ankle and also in right scapular region. The swelling in the left scapular region was first smaller in size, around 2-3 cm, gradually increased in size and attained the present size. There is a strong family history of similar complaints. His father, his grandfather and his great grandfather also had the same complaints. No other significant history was noted. On examination, the child was active, obeying commands. His vitals were stable. CVS/RS/CNS/ABD – were normal. Two well-defined swellings were noted in the left scapular region. The swelling was hard in consistency, together measuring around 10 x 10 cm. Multiple small well-defined swellings were noted around the both knee joints, right ankle and also in right scapula.

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X-Ray of the left shoulder was taken which showed (Fig. 1 & 2) two well-defined large exophytic lobulated lesions noted arising from the left scapula. Matrix appears to be cartilaginous. No obvious soft tissue noted. No surface disruption. Visualised humerus shows no cortical disruption. No periosteal reaction. Glenohumeral junction couldn't be adequately evaluated due to bony overlap. Subluxation of acromioclavicular joint is noted. Similar multiple small well-defined swellings were also noted around both knee joints, right ankle and also in right scapula. (Fig. 3, 4, 5 and 6).

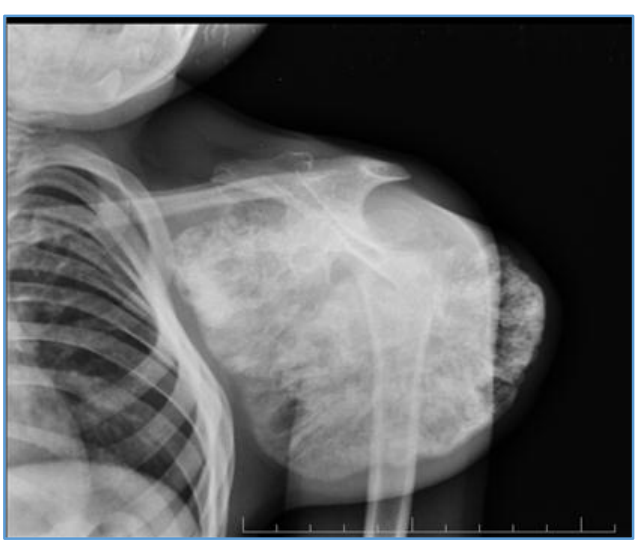

Fig. 1: X-Ray Left Shoulder showing two well-defined large exophytic lobulated lesions noted arising from the left scapula

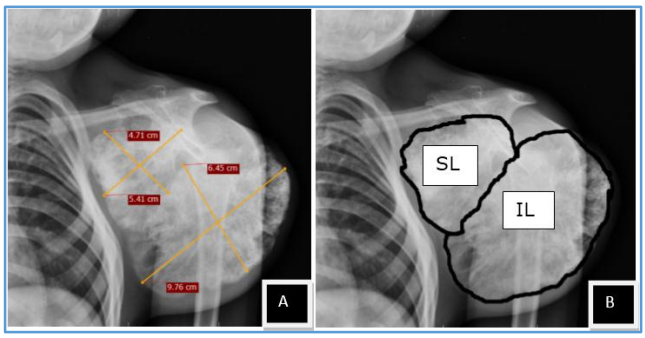

Fig. 2A: X-Ray Left Shoulder showing the measurements of both the exophytic lesion. 2B: X-Ray Left Shoulder showing the superior and the inferior lesion

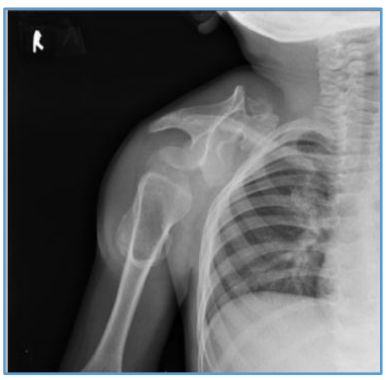

Fig. 3: X-Ray of right shoulder showing sessile exophytic lesion noted in the Right Humerus

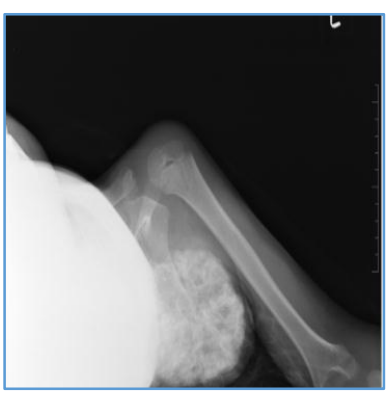

Fig. 4: X-Ray of Lateral view of Left Shoulder showing the exophytic lesion

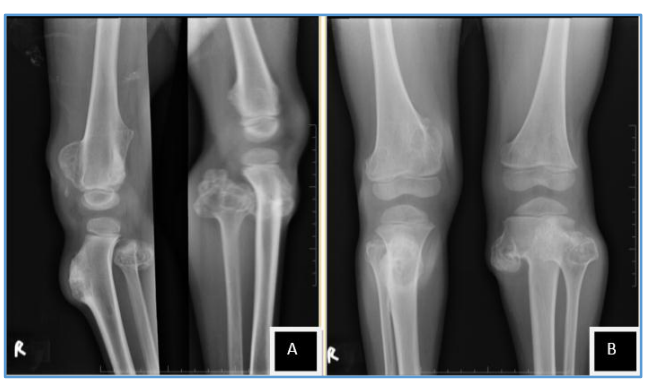

Fig. 5A & B: Shows sessile exophytic lesions noted in bilateral tibia and fibula

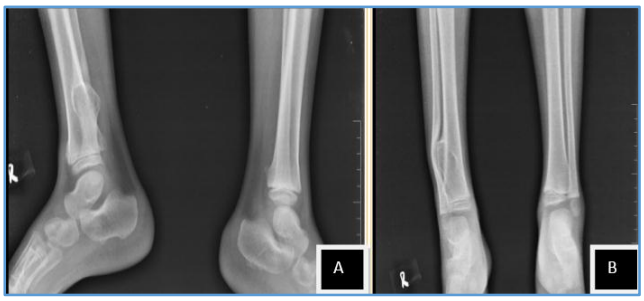

Fig. 6A & B: Shows sessile exophytic lesion noted in the Right lower 1/3rd of Tibia which is seen to indent and deform the contour of lower 1/3rd of Fibula

Patient was then subjected for MRI scan which showed two well-defined multilobulated sessile lesions arising from the posterior surface of the left scapula. Lesions measuring 7.1 x 4.6 and 6 x 4.6 cm. Continuity of the cortex of scapula seen extending to the lesions. Lesion shows T2 hyperintensity and T1 hypointensity cartilage cap covering the entire surface. Average thickness of cartilage cap is around 3-5 mm. Bone marrow within the lesion appears oedematous. A diagnosis of multiple osteochondromas of left scapula was made. (Fig. 7, 8 and 9).

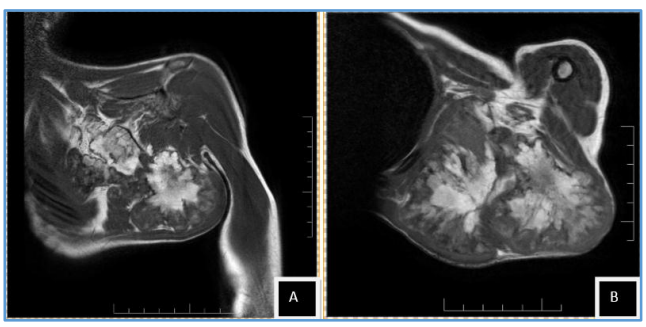

Fig. 7A & B: MRI T1 Sequence of coronal and axial view showing two well-defined multilobulated sessile lesions arising from the posterior surface of the left scapula with hypointensity cartilage cap covering the entire surface

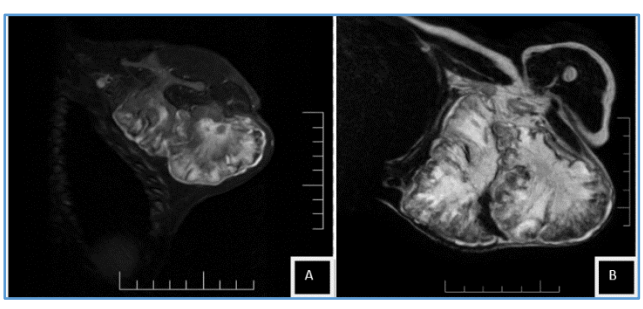

Fig. 8A & B: MRI T2 Sequence of coronal and axial view showing two well-defined multilobulated sessile lesions arising from the posterior surface of the left scapula with hyperintensity cartilage cap covering the entire surface

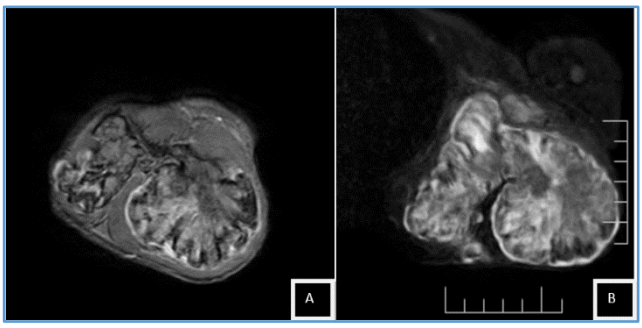

Fig. 9A & B: MRI STIR Sequence of coronal and axial view showing two well-defined multilobulated sessile lesions arising from the posterior surface of the left scapula with average thickness of cartilage cap is around 3-5 mm

The child was operated and a sample was sent for histopath analysis which was reported to be osteochondroma. (Fig. 10)

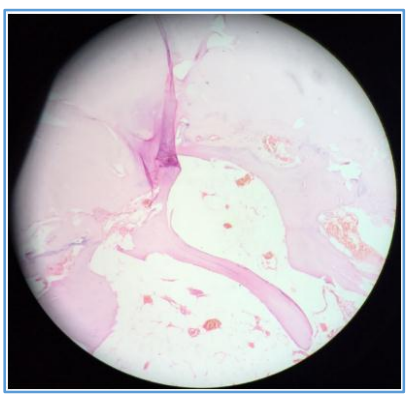

Fig. 10: High resolution microscopic image – H&E stain: Section shows mature cartilaginous cap with endochondral ossification with bony trabecular spaces with marrow elements also seen

DISCUSSION: Osteochondroma is the most common benign bone tumour. The tumour is often diagnosed as an incidental finding. Osteochondromas account for approximately 35% of benign bone tumours and 9% of all bone tumours. Most are asymptomatic, but they can cause mechanical symptoms depending on their location and size.

Hereditary multiple exostosis also called as diaphyseal aclasis (aclasia), multiple osteochondromas, familial osteochondromatosis. Hereditary multiple exostosis (HME) is an inherited autosomal dominant disorder where multiple osteochondromas throughout the skeleton are present.[1,3] It is genetically heterogeneous, with three loci, currently identified on chromosomes 8q24.1, 11pl3, and 19q.[4] Prevalence is around 1:50,000-1:100,000. Age of onset is found to be between 2 and 10 years with male predominance (2:1). Patients with HME may present with short stature and asymmetric growth at the knees and ankles, which may lead to deformities. The osteochondromas are located close to the metaphyses, and they may be sessile or pedunculated. The cortex of the lesion is continuous with the cortex of the bone with a homogeneous continuation of the medulla which is a characteristic feature. Usually, a painless mass near joints. Mechanical limitation of joint movement may occur. The affected individuals have a positive family history.[5,6]

Location is usually multiple and bilateral. Common sites are knee, elbow, scapula, pelvis, ribs. Sites being metaphyses of long bones near epiphyseal plate (distance to epiphyseal line increases with growth); always point away from joint and toward centre of shaft. Occasionally, small punctate calcifications are seen in cartilaginous cap. The skeletal distribution of lesions varies, which can be bilateral and symmetric or unilateral.[2,7]

Specific sites of involvement include: Scapula and ribs (40% of cases), humerus (50%–98%), elbow (35%–40%), wrist (30%–60%), hands (20%–30%), pelvis (5%–15%), hips (30%–90%), knees (70%–98%), ankles (25%–54%) and feet (10%–25%).[3,6,8]

The conventional radiograph of an osteochondroma can be either sessile or pedunculated, and is seen in the metaphyseal region typically projecting away from the epiphysis. There is often associated broadening of the metaphysis from which it arises. The cartilage cap is variable in appearance. It may be thin and difficult to identify, or thick with rings and arcs calcification and irregular subchondral bone. New cortical irregularity or continued growth after skeletal maturity has been reached, as well as frankly aggressive features (e.g. bony destruction, large soft tissue component, and metastases) are all worrying for malignant transformation.[4,5,6]

Ultrasonography enables accurate assessment of the cartilage cap of exostosis (as a hypoechoic region bounded by bone on its deep surface and muscle/fat superficially). The detection rate and measurement accuracy of ultrasound is higher than with computed tomography and comparable to magnetic resonance imaging. Ultrasonography appears to be a good procedure for evaluating the cartilage cap, which is usually thin for a benign exostosis and thick for a malignancy.[7,8]

CT demonstrates the same findings as on radiograph, but is better able to demonstrate medullary continuity and the cartilage cap.[5,8]

MRI is the best imaging modality to assess cartilage thickness (and thus assessing for malignant transformation), presence of oedema in bone or adjacent soft tissues, and visualising neurovascular structures in the vicinity. The cartilage cap of osteochondromas appears the same as cartilage elsewhere, with intermediate-to-low signal on T1 and high signal on T2 weighted images. A cartilage cap of over 1.5 cm in thickness is suspicious for malignant degeneration. With intravenous gadolinium administration, enhancement of benign lesions is normally seen in the tissue that covers the cartilaginous cap which is fibrovascular in nature; however, the cartilaginous cap itself should not enhance.[6,8]

Treatment of HME is surgical, usually for cosmetic rather than symptomatic reasons. Recurrence of the tumour following adequate excision is highly suggestive of an aggressive lesion. Another reason for surgery is for the removal of the osteochondroma in the event of sarcomatous degeneration.[8,9]

CONCLUSION: Hereditary multiple exostosis is an inherited disorder characterized by multiple osteochondromas.[1] It is genetically heterogeneous, with three loci, currently identified on chromosomes 8q24.1, 11pl3, and 19q.[4] The chief complaint is the discovery of single or multiple hard painless masses near joints. The distribution is usually bilateral and may be symmetrical leading to dwarfing and deformities. MRI is the best imaging modality to assess cartilage thickness.[6,8] Malignant degeneration into chondrosarcoma occurs in 5-25%. It is therefore important to monitor all cases of HME especially if the patient complains of pain or growth of an osteochondroma.[9]

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