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Giant Osteochondroma of Tibia in a Case of Hereditary Multiple Exostoses: A Case Report

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ABSTRACT

Osteochondromas, aka exostoses, are benign neoplasms of the cartilage. Genetic mutations of the EXT1 and EXT2 genes leads to a syndrome causing multiple osteochondromas, namely the hereditary multiple exostoses. Among the complications related to multiple osteochondromas, malignant degeneration into chondrosarcoma has the worst prognosis. Sudden enlargement of size of an osteochondroma or sudden onset of pain at the site of previous osteochondroma in a skeletally mature patient is concerning for malignant transformation. However, the size criterion does not apply for skeletally immature individuals, as observed in the case report here. The imaging of an uncommonly large osteochondroma of the extremity in a skeletally immature patient with hereditary multiple exostoses has been described here. Normal thickness of the cartilage cap (<3cm) proved the benignity of the lesion on imaging, thereby nullifying the size criterion in the skeletally immature.

Keywords: Hereditary multiple exostoses, Osteochondroma, Cartilage cap, Heparan sulphate, Bayonet deformity, Chondrosarcoma.

INTRODUCTION

Osteochondromas (exostoses) are benign tumours of the cartilage consisting of a pedicle or a broad base of normal bone covered by proliferating cartilage cells. Hereditary multiple exostoses (HME, aka diaphyseal aclasis) is an autosomal dominant disorder caused by loss of function mutations of the genes responsible for heparan sulphate synthesis (EXT1 and EXT 2) in turn required for proper cartilage development and long bone growth **[1]**. An average of six exostoses are seen in a HME patient.

Osteochondromas can cause valgus deformities, limb length discrepancies and compensatory scoliosis of the spine. Bayonet deformity of forearm and brachydactyly are seen in the upper limb. Usually the earlier the onset, the bigger the lesion causing more problems such as neurovascular compression and bursitis. In 2 to 4% of cases of HME, malignant degeneration into chondrosarcoma is seen. Solitary osteochondromas have a much lesser risk of malignant degeneration less than one percent **[2]**.

CASE DESCRIPTION

An eleven-year-old female presented with a large painless swelling at the back of her right leg which had progressively increased in size for the past 8 years. There was no history of any constitutional symptoms or previous trauma. Her father had history of multiple painless swellings in the extremities.

Plain radiograph showed a large well defined irregular bony lesion arising from the proximal meta-diaphyseal region of right tibia showing chondroid pattern of matrix calcification within. Few sessile and pedunculated metaphyseal outgrowths were seen centered around the left knee and left ankle joint in the radiograph of the left lower limb. This was suggestive of multiple osteochondromas.

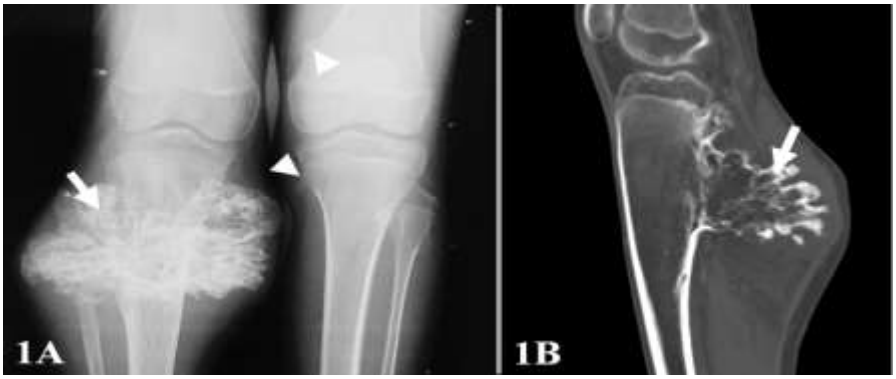

Figure 1: Plain radiograph and computed tomography images in a case of hereditary multiple exostoses - 1A) Anteroposterior radiograph of bilateral knee and upper legs and 1B) Sagittal CT reformat of proximal right leg (in bone window) showing a large cauliflower exostosis (arrows) arising from proximal right tibia. Sessile and pedunculated osteochondromas (arrowheads) arising from metaphyses centered around left knee joint are also observed in the radiograph.

In view of its large size, the lesion in the right leg was suspected to undergo malignant transformation. MRI of the lesion revealed a large well defined irregular narrow based lesion arising from the posterior aspect of medulla of proximal meta-diaphysis of right tibia showing cortical continuity with the tibia. A cartilaginous cap of ~ 1.2cm in maximum thickness was seen overlying it. The lesion measured ~13 cm in its maximum dimension. No breach of overlying cortex or soft tissue infiltration was seen. The diagnosis of a giant benign osteochondroma with hereditary multiple exostoses was made.

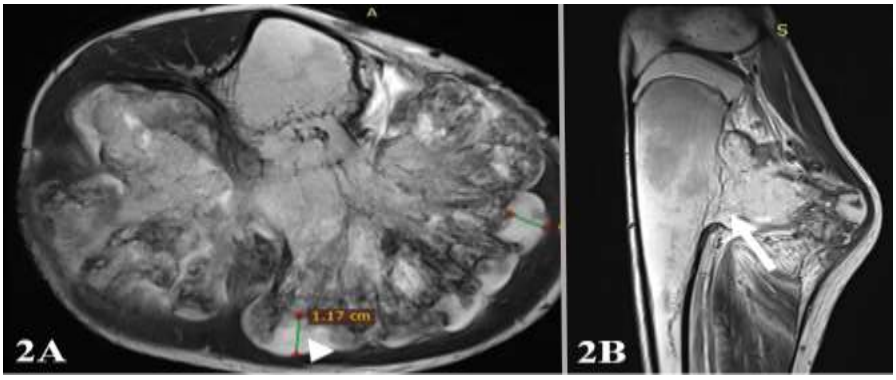

Figure 2: MRI of giant osteochondroma of right tibia 2A) Axial T2 Turbo Spin Echo image shows large cauliflower exostoses with overlying thin hyperintense cartilage cap (white arrowhead). 2B) Sagittal T2 Turbo Spin Echo image shows hypointense cortical and hyperintense medullary continuity (white arrow) of parent bone with the lesion.

CT-angiogram for surgical planning showed a normal popliteal artery bifurcation into anterior tibial artery and tibio-peroneal trunk. However, the vessels at the level of the mass were not well delineated and were seen to reform at the level of mid shaft of right tibia.

DISCUSSION

Malignant degeneration of an osteochondroma is clinically suspected in adults when there is sudden onset of pain or growth of a previously stable exostosis and when the cartilage cap is more than 1.5–2 cm **[2]**. In skeletally immature patients, continued active growth (with cartilage thickness upto 3cm) should not be viewed as a sign of malignant transformation.

Plain films are normally sufficient to diagnose diaphyseal aclasis. Ultrasonography is a good procedure for evaluating the cartilage cap alone. CT

allows optimal depiction of the pathognomonic cortical and marrow continuity of the lesion and parent bone, especially in concealed locations such as ribs.

MR imaging is the best radiologic modality for visualizing the effect of the lesion on surrounding structures and in evaluating the hyaline cartilage cap to look for sarcomatous degeneration **[3]**. On reviewing the available literature, osteochondromas larger than 10 cm have not been reported yet **[4,5]**.

The maximum dimension of the lesion of our concern being around 13cm, complications such as neurovascular compression and malignant degeneration were suspected. Also, in HME, the risk of malignant degeneration is higher (2-4%) compared to the solitary osteochondromas (1%). However, MRI images revealed a cap thickness of ~ 1.2cm which is within the cut off range of 3cm for skeletally immature patients, thereby ruling out malignant degeneration.

However, owing to the potential future growth disturbances that the lesion can cause, surgical excision of the giant osteochondroma in right leg was advised by the orthopedic team. Post resection histopathology of the lesion was done which confirmed the imaging findings. Ultrasound or MRI are hence mandatory in large osteochondromas for assessing the cartilage cap thickness and ruling out sarcomatous transformation.

Treatment of HME is usually conservative unless the osteochondromas become painful, demonstrate rapid or new growth, enlarge after skeletal maturity and/or exhibit signs of malignant transformation following which they are surgically excised. Recurrence is highly suggestive of an aggressive lesion **[4]**.

CONCLUSION

Despite its large size, benignity of the giant osteochondroma in our case was certain on imaging. As described in literature, it is observed that size is not a criterion to suspect malignant degeneration, particularly in skeletally immature patients. Therefore, imaging is mandatory to assess the cartilage cap thickness in suspected cases of malignant transformation.

Declaration by Author
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