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MULTIPLE HEREDITARY EXOSTOSIS PRESENTING WITH CHONDRO-SARCOMATOUS TRANSFORMATION WITH PARAPARESIS: A CASE REPORT

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Abstract
Chondrosarcoma arising from hereditary multiple exostoses(HME) which is a rare but serious complication that warrants close attention due to its significant impact on patient health. This is a case report of a 54year old male who had been diagnosed with HME, had subsequently developed chondrosarcoma in his lumbar spine. This malignant transformation led to paraparesis, a condition characterized by partial paralysis of the lower limbs. In this manuscript, we delve into the clinical presentation of this patient, outlining the initial symptoms and progression of the disease. The diagnostic workup is detailed, including imaging studies and histopathological examination, which confirmed the presence of chondrosarcoma. Treatment strategies are thoroughly discussed, highlighting surgical interventions, possible adjuvant therapies, and the rationale behind the chosen management approach. The outcomes are also examined, providing insights into the prognosis and recovery process. Emphasis is placed on the challenges faced in managing such complex cases and the principles guiding effective treatment.

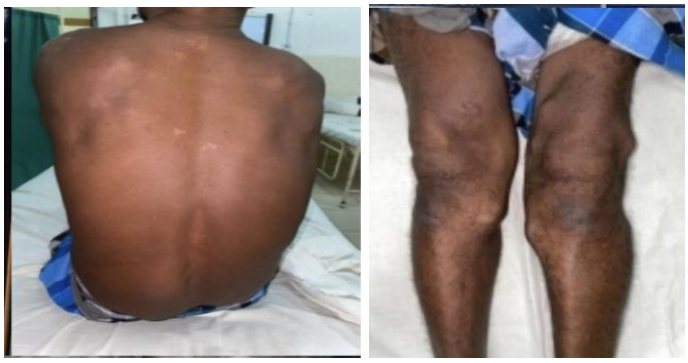
INTRODUCTION

Chondrosarcoma is a malignant neoplasm of cartilaginous origin, accounting for a significant portion of primary bone tumors. It can arise de novo or from pre-existing benign cartilaginous lesions such as osteochondromas, which are characteristic of Hereditary Multiple Exostoses (HME). Hereditary Multiple Exostoses is also called as multiple osteochondromatosis, which is a Genetic disorder where the patient has development of multiple osteochondromas during childhood. 1 in 50,000 people have HME making it one of the most commonest genetic musculoskeletal conditions presenting with an incidence rate of 1 in 50,000 people, where we see multiple cartilage capped exostosis that form in childhood and they ossify during adulthood. less than 10% of all spine chondroma occur in thoracic spine. While typically benign, these lesions have the potential for malignant transformation into chondrosarcomas, posing significant clinical challenges. Compressive neuropathy is common and seen in 22 to 23% of the HME patients [1,2]. It impacts the patients physical activity and quality of life of patient from childhood, especially in female patients[3,4,5].

Case Presentation

Patient Presentation

A 54-year-old male presented with complaints of pain over both lower limbs for the past45 days and progressive weakness, leading to an inability to walk for the past 15 days with no history of trauma/evening rise of temperature/loss of weight and appetite. Clinical examination revealed multiple bony hard swellings consistent with osteochondromas with tenderness over the mass in the left paraspinal region with SLRT and FABER negative in both sides, Motor L2, L3-3/5 on both sides, L4-S1-2/5 on right ,L4- 1/5 on left ,Sensory-L2-2/2-both sides ,L3-S1-1/2 on both sides , lax anal tone and decreased perineal sensation with deep tendon reflexes- normal



Investigations

Laboratory tests showed elevated alkaline phosphatase(137iu/l) and C-reactive protein (19mg/l). In figure 1: Imaging via computed tomography (CT) revealed Large exophytic lesions with irregular sclerotic foci from posterior elements of L4L5 vertebra with soft tissue component in the Left paraspinal region with apparent extension at L3, L4 levels in spinal cord and left neural foramina. Bone scan has no value in the study of HME[6,7].

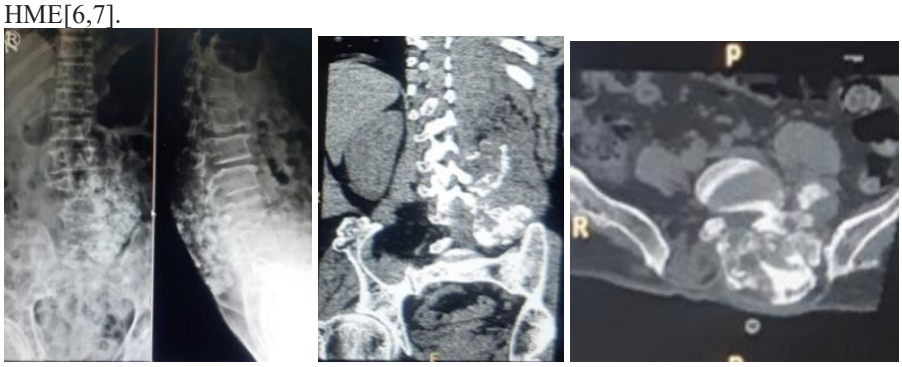


Figure 1: Preoperative radiographs

Treatment

The patient had surgical excision of tumor with decompression of spinal cord, and posterior stabilization at L3L4, L4L5 level. Adjuvant therapy with chemotherapy and radiotherapy was initiated to manage residual disease and prevent recurrence.

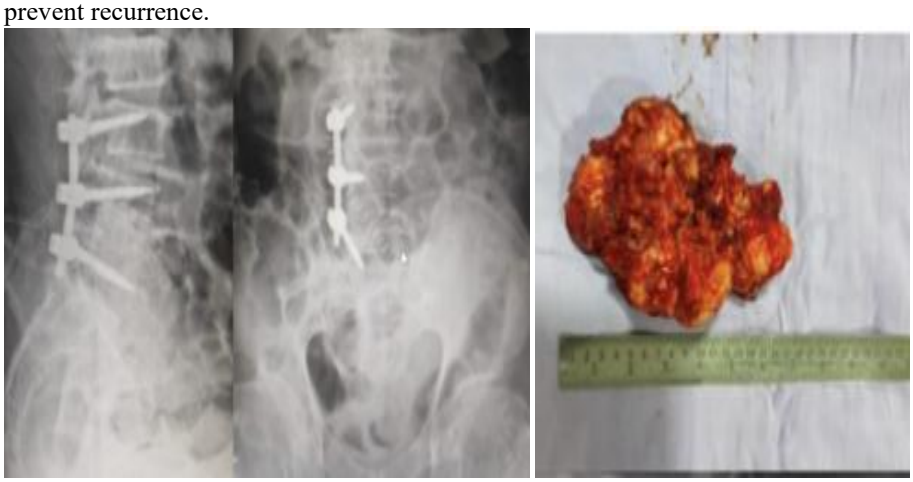


Figure 2: Postoperative radiographs

DISCUSSION

Osteochondromas are not true neoplastic lesions but rather developmental lesions of the bone. Hereditary multiple exostosis[1]. HME patients face a rare but serious risk of malignant transformation of osteochondromas into chondrosarcoma, particularly in the axial skeleton, including the lumbar spine. Lumbar spine involvement presents unique diagnostic and therapeutic challenges due to the critical anatomical structures involved. During the last two decades, incredible progress has been made in clinical examination, surgical techniques, and reconstruction with instrument at all levels of the spine[8].

Patients typically present with symptoms such as localized pain, neurological deficits (e.g., radiculopathy, paresthesia or weakness), and sometimes palpable masses. Radiographic evaluation, including MRI and CT scans, is crucial for assessing the extent of the lesion, identifying features suggestive of malignancy (e.g., increased size, irregular margins, cortical disruption), and planning surgical intervention. Biopsy and histopathological analysis confirm the diagnosis, revealing malignant chondroid cells. Suspicion of malignancy comes into question when its cartilage cap expands more than 1.5–2 cm or exostosis grows in any dimension in adulthood[9,10]

Management primarily involves surgical resection with an emphasis on achieving clear margins to minimize recurrence and preserve neurological function. Due to the complex anatomy of the lumbar spine, this often requires a multidisciplinary approach involving orthopedic surgeons, neurosurgeons, and oncologists. Adjuvant therapies, including chemotherapy and radiation, are generally reserved for high-grade or metastatic chondrosarcomas but remain controversial due to limited efficacy in chondroid tumors. Chondrosarcomas arising from HME, particularly in the spine, are rare and associated with poor outcomes due to their resistance to conventional therapies. Surgical resection remains the cornerstone of treatment, aiming for complete excision whenever feasible. The role of adjuvant therapies, such as radiotherapy and newer modalities like proton-beam therapy, is crucial in improving local control and reducing morbidity. The complex issues related to diagnosis of HME is challenging for doctors and researchers even with advanced progress made in the last few decades[11,12]. Schmale et al states that serial radiographic imaging is warranted for all large osteochondromas, particularly those in high-risk locations to identify cortical invasion by tumor and thickness and metabolic activity of cartilage caps [13].

Vanhoenacker et al stated that conventional radiography is sufficient to confirm the diagnosis and to define the extent and the evolution of the disease, MRI is the imaging technique of choice to evaluate symptomatic lesions

CONCLUSION

In this case report we see the importance of early detection and comprehensive management of patients with HME to mitigate the risk of malignant transformation. Timely intervention involving multidisciplinary approaches, including surgery and adjuvant therapies, is essential for optimizing outcomes and preserving neurological function. Further research is warranted to explore targeted therapies and improve treatment efficacy in rare conditions like spinal chondrosarcomas arising from HME.

Author declaration

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