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Case Report

Chondrosarcoma secondary to hereditary multiple osteochondromas with spinal cord compression: A case report and systematic review

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ABSTRACT

Background: Hereditary multiple osteochondromas (HMOs) are a rare genetic disorder characterized by the formation of multiple benign osteochondromas that can undergo malignant transformation into chondrosarcoma.

Case Description: A 24-year-old male with a history of HMO and osteochondroma surgery 4 years ago, presented with back pain and paresthesias. The magnetic resonance showed a right paravertebral infiltrating mass at the T12–L1 level causing spinal cord compression. Following *en bloc* resection of the tumor, the patient's symptoms/signs resolved. The final pathological diagnosis was consistent with a chondrosarcoma.

Conclusion: Chondrosarcomas secondary to HMO with spinal cord compression are rare. These patients often presenting with significant myelopathy/cord compression should undergo gross total resection where feasible to achieve the best outcomes.

Keywords: Diaphyseal aclasis, Hereditary multiple exostoses, Hereditary multiple osteochondromas, Secondary chondrosarcoma, Spinal cord compression

INTRODUCTION

Hereditary multiple osteochondromas (HMOs) are a rare autosomal dominant disease characterized by multiple osteochondromas.[1] In 0.5–5% of cases, they can undergo malignant transformation into chondrosarcomas that can cause cord compression.[1] Here, a 24-year-old male with HMO originally presented with osteochondromas that evolved into low-grade chondrosarcoma at the T12/L1 level warranting gross-total resection.

CASE PRESENTATION

A 24-year-old male presented with back pain/paresthesias associated with a growing right-sided, posterior, infiltrating mass magnetic resonance (MR)-documented T12–L1 [Figure 1]. Notably,

4 years ago, the patient underwent a surgical excision of an osteochondroma in the right distal femur. Through T12–L2 laminectomy tumor was found to invade the T12–L1 vertebral pedicles and T12–L2 nerve roots; they were resected. Next, a transpedicular arthrodesis (T11–L2) was performed. In addition, the tumor was removed from the costal cartilage of the 10–12 ribs (i.e., due to invasion of the thoracic cage/right hemithorax) [Figure 2]. The biopsy was consistent with malignant transformation into a low-grade chondrosarcoma [Figure 3]. Postoperatively, the patient had a pneumothorax that was appropriately treated. The postoperative MR imaging confirmed complete tumoral resection [Figure 4]. He was then discharged neurologically intact.

Literature epidemiology

HMO is a rare genetic disorder, with an incidence of 1.5% in the Western population.[2] In our review, 80% have a positive family history and more than 80% of the diagnosis is made

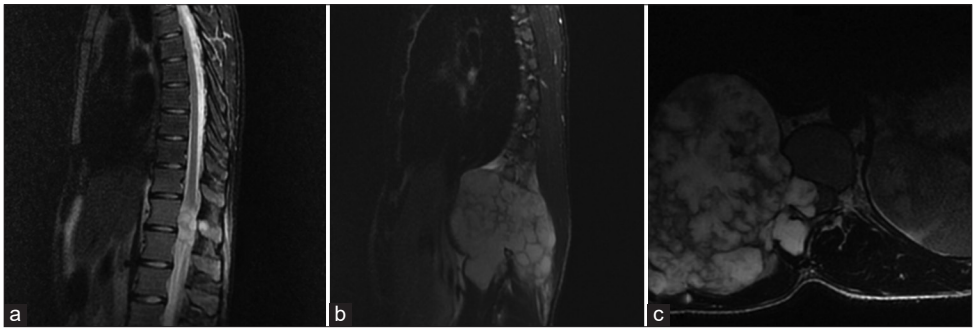

Figure 1: Preoperative magnetic resonance imaging. (a) Midsagittal T2-weighted image (T2WI) with fat suppression revealing a hyperintense lesion causing medullary invasion at the T12–L1 levels. (b) Midsagittal T2WI with fat suppression showing a hyperintense expansive lesion in the right paravertebral area. (c) Axial T2WI with fat suppression revealed a significant spinal cord compression by the tumoral mass.

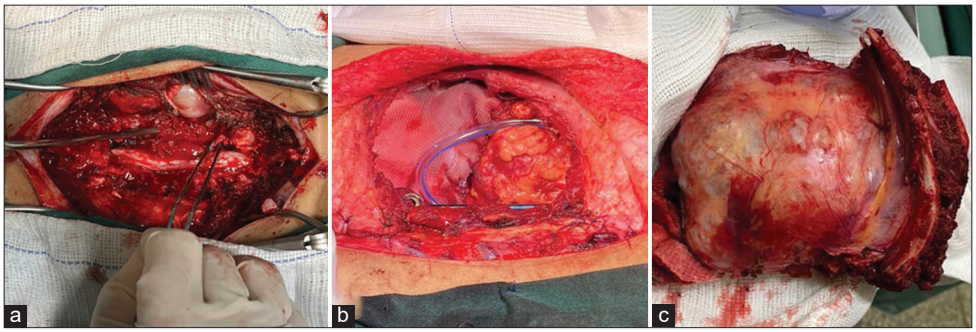

Figure 2: Intraoperative images. (a) View of the tumor after posterior laminectomy. (b) View of the paravertebral area after the tumor resection. (c) Tumoral mass after surgical resection.

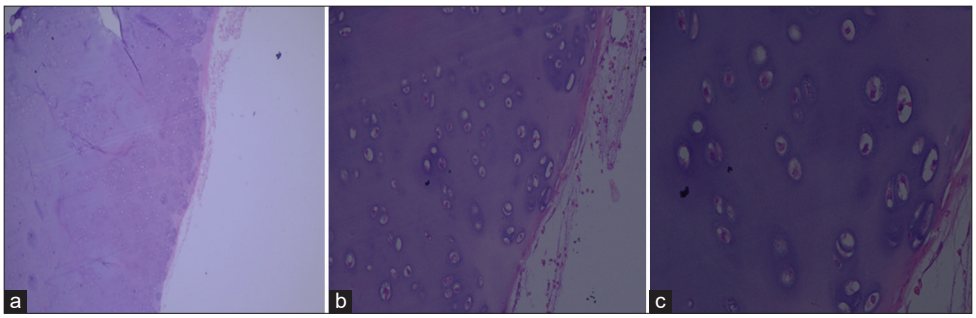

Figure 3: Biopsy examination (histology). (a) Abundant cartilaginous proliferation with chondrocytes embedded in lacunae. (b and c) Minimally increased cellularity and occasional binucleated nuclei and lobular growth pattern.

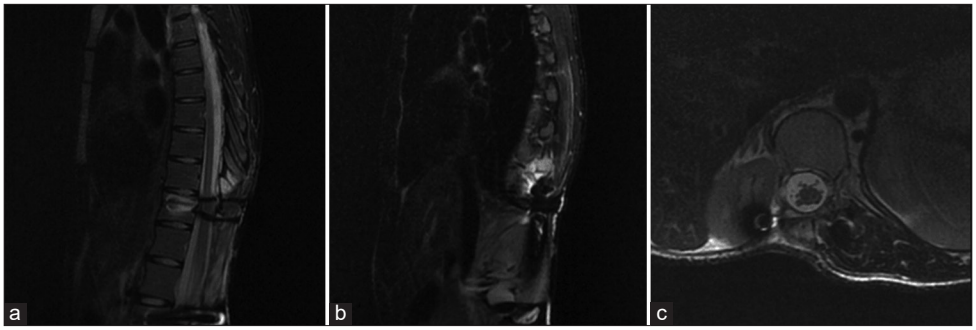

Figure 4: Postoperative magnetic resonance imaging. (a and b) Midsagittal T2-weighted image (T2WI) with fat suppression showing complete resection of the tumoral components. (c) Axial T2WI with fat suppression revealing resolution of the spinal cord compression after the tumor removal, with the right kidney posterior displacement due to surgical procedure.

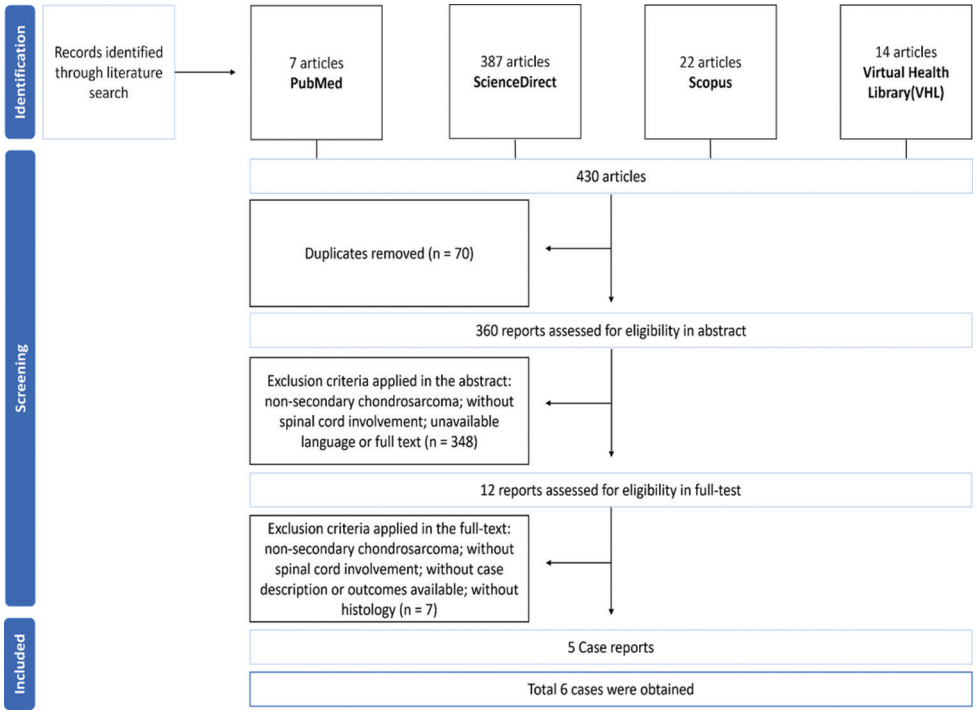

Figure 5: Study flowchart describing the literature search and analysis process.

before the end of the first decade.[2] Five articles met our inclusion criteria and included seven patients [Figure 5]. The mean age was 22 years (range 13-30 years), with four males and three females. Symptoms included pain (five patients) and paresthesia (four patients). The spinal cord was involved in two cases in the cervical spine, two cases in the thoracic spine exclusively, two cases in the lumbar spine exclusively, and one case in the thoracic/lumbar spine. Historically, six of seven patients had osteochondromas previously identified in other regions. Complete resection was performed in six cases, and supplemented with adjuvant chemotherapy in one of these cases. Pathologically, five patients had low-grade chondrosarcomas, while two patients had high-grade lesions. Complete recovery was seen in 3/5 patients, partial recovery in 1/5 patients, and 1/5 patients no recovery [Table 1].

Complications of malignant transformation

Malignant transformation into secondary chondrosarcoma is still the most feared of all complications of HMO.[1,7,9] Paulino Pereira *et al*. reported the following complications occurring up to 90 days after chondrosarcomas resections; dural rupture, respiratory failure, deep infection, fistula (pleuro-incisional), cardiac arrest during surgery, pneumothorax, sepsis, instrumentation failure, recurrence with spinal cord injuries, sternal osteomyelitis, and atrial thrombus.[10]

Table 1: A literature review of reported cases describing patients with spinal cord involvement due to chondrosarcoma secondary to hereditary multiple osteochondromas.

Author, year (country)	Age	Sex	Level	Family history	Other region osteochondromas	Case presentation	Treatment	Histology	Outcome
Crowell and Wepsic, 1972 (USA)[5]	13 years old	M	T1-T2	Yes	Right ulna and bilaterally humerus, radius, and femur	Lower limb paresis	Complete resection (*en bloc*)	Low-grade chondrosarcoma	Partial recovery
Crowell and Wepsic, 1972 (USA)[5]	18 years old	M	T7-T8	Yes	Rights humerus and femurs	Lower limb paresis	Complete resection (*en bloc*)	Low-grade chondrosarcoma	Complete recovery
Hatori *et al*., 2009 (Japan)[7]	28 years old	F	C2-C6	NA	Left scapula, right humerus, and femur	Pain and mild restriction of neck extension	Complete resection and chemotherapy	High-grade chondrosarcoma	No recovery
Anantharamaiah *et al*., 2012 (India)[1]	22 years old	M	L3-L5	No	No	Low back pain	No resection	High-grade chondrosarcoma	NA
Landi 2012 (Italy)[8]	30 years old	F	C6	Yes	Right elbow and left knee	Pain and paresthesia	Complete resection (*en bloc*)	Low-grade chondrosarcoma	Complete recovery
Mesfin *et al*., 2013 (USA)[9]	19 years old	F	L3-L5	NA	Rights femur and tibia	Low back pain	Complete resection (*en bloc*)	Low-grade chondrosarcoma	NA
Present study, 2023 (Brazil)	24 years old	M	T12–L2	Yes	Right femur	Low back pain and paresthesia	Complete resection (*en bloc*)	Low-grade chondrosarcoma	Complete recovery

NA: Not available, M: Male, F: Female

Pathology of HMO

Secondary chondrosarcoma occurs in only 0.5–5% of cases and is characterized by malignant transformation of anterior cartilaginous lesions. Myxoid change may be characterized by the loss of the lacunar pattern with pleomorphic spindle or stellate cells floating in a myxoid stroma.[6] The histological grade determines the prognosis of secondary chondrosarcomas; 10-year survival rates of 83% were reported for Grade I versus 29% for Grade III lesions.[1] We found that the complete resolution rate was higher in patients with low-grade chondrosarcomas.

Genetics of HMO

HMO is a genetically heterogeneous disease caused by tumor suppressor genes Exostosin-1 (EXT1) on chromosome 8, and Exostosin-2 (EXT2) on chromosome 11.[8] The patient's peripheral blood can be screened for germline mutations in EXT1 or EXT2 (i.e., point mutations or crude deletions in almost 90% of HMO patients).[4] Some studies have suggested the involvement of a third gene (EXT3) on chromosome 19, which could also be associated with HMO.[6]

Surgical resection to optimize survival of chondrosarcomas

Optimally, HMO lesions are treated surgically, and best with *en bloc* resection of thoracic tumors. Thoracic surgical options include; anterior, posterior, anterior approach first and posterior second, and finally, the posterior approach first followed by simultaneous anterior/posterior surgery.[3] For low-grade chondrosarcomas (Grade I), the best long-term survival depends on good local control (i.e., *en bloc* resection with negative margins (R0 resection) is currently the best predictor of overall survival). Intermediate-grade (Grade II) and high-grade (Grade III) chondrosarcomas should also be treated with R0 resection. For lesions treated with positive or marginal margins (R1/R2), recurrence is inevitable.[11]

Resistance to radiation therapy (RT)

Chondrosarcomas are resistant to RT. Therefore, RT should only be reserved for those who are not candidates for surgery.[5,11]

CONCLUSION

Malignant transformation to chondrosarcoma may occur in patients with underlying HMO. Once, identified optimal outcomes are associated with gross total *en bloc* excision.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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