

Case Report

Chordoma of the Lower Back in an Adolescent: A Rare Cytologic Presentation

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ABSTRACT

Chordomas are low-grade malignant tumors of bone that occur almost exclusively in the axial skeleton. Chordomas are rare in children and adolescents and comprise <5% of all cases and the site of development is at the skull base. These tumors are believed to behave more aggressively than chordomas in adults and may have unusual morphology. We herein present a rare case of chordoma in a 20-year-old male with low back mass, diagnosed on fine needle aspiration cytology.

Keywords: Chordoma, Fine Needle Aspiration, Cytology

Introduction

Chordomas are rare malignant bone tumors which involve mainly both ends of the axial skeleton and present as destructive bone lesions with a large soft tissue mass (1). Chordomas develop most frequently in the skull base in <5% children and adolescents (2). These tumors behave more aggressively in children than in adults with unusual morphology (2). Notochordal remnants were the previous believed site of origin of chordomas, but recent studies suggest the tumors to arise from benign

notochordal cell tumors (3). The circumstantial evidence for the same is the location of the tumors (along the neuraxis) with similar immunohistochemical staining patterns (3).

Chordomas are known to occur in the fifth to seventh decades of life (4). It usually presents as pain/tenderness in the low back with constipation or painful bowel movements. Radiographically, chordomas appear as solitary mid-line lesion with destruction of the involved bone. We report a rare case of chordoma in an adolescent male, who presented as low back mass and was diagnosed on fine needle aspiration cytology.

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Case Summary

A 20-year-old male presented in the Surgical Ward with a 10× 8 cm mass in the lower back with tenderness. On plain x-ray, a solitary mid-line lesion with bony destruction and focal sacral calcifications was revealed. The imaging studies (computed tomography and magnetic resonance imaging) helped to demonstrate the soft tissue component, calcifications and sacral destruction. Subsequently, fine needle aspiration of the mass was performed. Smears showed groups of large tumor cells, with small round nuclei and abundant vacuolated cytoplasm suggestive of chordoma (Fig.1).

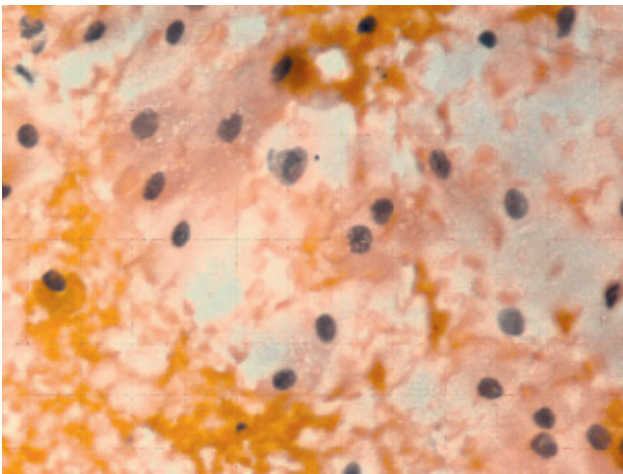


Fig.1: Chordoma: FNA smears showed groups of large tumor cells, with small round nuclei and abundant vacuolated cytoplasm. (Haematoxylin and Eosin × 40)

Surgical excision of the lesion showed an encapsulated mass in the sacral soft tissue that had invaded into the sacrum. The cut surface of the tumor was lobular, greyish and mucoid with areas of focal necrosis. Microscopically, the tumor showed characteristic lobular arrangement with fibrous septations. The malignant cells had eosinophilic cytoplasm and prominent vacuoles pushing the nuclei to the side (physaliphorous cells), confirming the cytological impression. The diagnosis of chordoma was supported by electron

microscopy, which showed that the tumor cells contained numerous mitochondria surrounded by profiles of rough endoplasmic reticulum. Adjuvant treatment in the form of radiation therapy (Co-60,35Gy) was administered to our patient and he is well after 6 months of follow up.

Discussion

Chordoma is a rare malignant tumor that arises from notochord remnants and account for 1 to 4% of all bone tumors (5). We observed a case of chordoma in a 20 year old male, though it occurs in older adults, with the highest prevalence in the fifth to seventh decades of life(4). Chordomas occur in the mid-line of the axial skeleton as they are known to originate from the notochord and can arise from bone in the skull base or anywhere along the spine. The two most common locations are cranially at the clivus and in the sacrum at the bottom of the spine (5). Our patient presented with sacral mass of 10 × 8 cms in size. Other more uncommon sites include transverse processes of vertebrae and the paranasal sinuses.

Though our patient had no family history of a similar lesion, a small number of families have been reported in which multiple relatives have been affected by chordoma (6). In four of these families duplication of the brachyury gene was found to be responsible for causing chordoma (6). A possible association with tuberous sclerosis complex (TSC1 or TSC2) has been suggested (7). Surgical excision of the lesion in our patient, showed an encapsulated mass in the sacral soft tissue that had invaded into the sacrum. But Baboiu and Taylor (8) have reported a case of sacral chordoma, presented as a mobile, encapsulated, benign soft tissue mass without any sacral involvement or lytic lesion. Chordomas are known to be locally invasive with rapid growth, despite optimal treatment, which emphasizes the need to find ways of their aggressiveness. Prolifera-

tion, invasiveness and metastasis in epithelial tumors have been shown to be related to alterations in adhesion proteins. Triana *et al.* (9) analyzed the expression of E-cadherin, N-cadherin, as well as their cytosolic binding proteins alpha-catenin, beta-catenin, and gamma-catenin, in 51 paraffin archived and 17 cryopreserved chordoma specimens and found E-cadherin and N-cadherin expression was inversely correlated, whereas beta-catenin and gamma-catenin expression was directly correlated in the majority of chordomas. Han *et al.* (6) have reported that all sacral chordomas exhibit phosphorylation of Ribosomal protein s6 and EIF4EBP1 by immunohistochemistry with partial or complete PTEN (gene) deficiency.

In one study, the 10-year tumor free survival rate for sacral chordoma was 46% (10). Chondroid chordomas appear to have a more indolent clinical course and in most cases, complete surgical resection with adjuvant radiation therapy offers the best chance of long-term control (11). Incomplete resection of the primary tumor increases the odds of recurrence. Recurrence is reported approximately in 50% cases of sacral and coccygeal chordomas (12). Adjuvant treatments such as radiation and proton beam therapy have been extensively used for difficult, recurrent, or unresectable cases with considerable success. Overall, base of skull chordomas in children and adolescents treated with proton beam radiation have better survival than chordomas in adults (1). Chemotherapy can be used for late stage disease. New drug treatments, including use of a drug called imatinib mesylate are being investigated which may help slow the growth of tumors that cannot be removed (13). Our case of chordoma is disease free, after 35 Gy of Cobalt-60 adjuvant radiation therapy and 6 months of follow up period.

The diagnosis of chordoma or a primary sacral tumour remains uncommon, however, should

be considered in patients with dull lower back or coccygeal pain (typically worse on sitting) especially if associated with an alteration of bowel or urinary habits. Chordoma and other sacral tumours are curable with surgery and early diagnosis by fine needle aspiration cytology may lead to preservation of bladder, bowel, motor and sexual function.

Acknowledgements

The authors declare that there is no conflict of interests.

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