



SACRAL CHORDOMA: A DIAGNOSTIC PREDICAMENT

*Sunita Bamanikar¹, N.K.Panicker², Nidhi Gupta³, Archana Buch⁴

Department of Pathology, Padmashree Dr.D.Y.Patil Medical College, Hospital and Research Centre, Dr.D.Y.Patil Vidyapeeth, Pimpri, Pune 411018, Maharashtra, India.

Article Received on 19/08/2014

Article Revised on 10/09/2014

Article Accepted on 05/10/2014

*Correspondence for

Author

Sunita Arvind Bamanikar

Department of Pathology,
Padmashree Dr.D.Y.Patil
Medical College, Hospital
and Research Centre,
Dr.D.Y.Patil Vidyapeeth,
Pimpri, Pune 411018,
Maharashtra, India

ABSTRACT

Chordoma is a rare slow growing tumour that arises from the remnants of embryonic notochord. They present mainly in the axial skeleton with a proclivity for the sphenoid-occiput and sacral regions. Chordomas can pose diagnostic challenge and be confused with chondrosarcoma or mucinous carcinoma. We report here a case of classical chordoma in an elderly male patient which was provisionally diagnosed by cytology and frozen section examination and confirmed subsequently by histopathological and immunohistochemical study.

KEY WORDS: Chordoma, immunohistochemistry, sacral.

INTRODUCTION

Chordoma is a rare malignant tumour of the bone. It arises from the embryonic remnants of the primitive notochord and occurs along the midline from the skull base to the sacrum. They account for 1-4% of all malignant primary bone tumours [1]. Three different varieties have been recognized: classical, chondroid and dedifferentiated. Immunohistochemical epithelial markers are classically positive in conventional chordomas.

They appear bland on pathology but are locally aggressive tumours and need prompt intervention at first presentation [2]. We report here a case of sacral chordoma in a 55-year-old male patient diagnosed provisionally on cytology and confirmed by histopathological and immunohistochemical examination.

CASE HISTORY

A 55-year-old male patient presented with lower backache since one year. There was history of acute onset of loss of power in right lower limb associated with inability to walk since 8 days. Tingling and numbness was also present in the right lower limb. He had history of angioplasty done eight months back. There was no history of diabetes, hypertension, asthma or breathlessness. He was afebrile with absence of pallor, oedema or clubbing. On examination, power in the knee, hip and ankle joint was reduced to grade IV. Straight leg raising test (SLR) was 30°. 2D echocardiography was normal with left ventricular ejection fraction (LVEF) more than 60%. Investigations revealed haematological parameters within normal range.

Radiographically a lytic lesion involving the sacral area (S4S5) was detected. On computerized tomography (CT) a lytic lesion involving sacral (S4-S5) with soft tissue involvement having heterogeneous contrast enhancement was seen. Hence, differential diagnosis of tuberculosis, secondary deposits, chondrosarcoma or giant cell tumor of the sacrum was entertained. At surgery an ill-circumscribed partly necrotic soft tissue mass was found, a small piece was sent for frozen section.

The imprint smears of the tissue showed sheets of multivacuolated large cells with abundant cytoplasm in a myxoid background (Figure 1). Frozen section of the tissue also showed cords and sheets of similar cells with mild nuclear atypia and abundant multivacuolated cytoplasm with loose myxochondroid background (Figure 2). A provisional diagnosis of chordoma or mesenchymal chondrosarcoma was given with request to excise the lesion completely.

Subsequently multiple pieces of tissue of gelatinous and fibrous consistency approximately 20 gm were received. Multiple sections from the tissues were processed for routine paraffin sections and stained with hematoxylin and eosin stain. Sections showed a lobulated architecture with sheets and cords of multivacuolated physaliphorous cells embedded in a myxoid stroma, hence a diagnosis of chordoma was suggested (Figure 3). Immunohistochemistry on paraffin sections showed positive staining of tumor cells for S-100 (Figure 4), epithelial membrane antigen (EMA), cytokeratin (CK) and vimentin. These findings confirmed the histopathological diagnosis of chordoma. The subsequent post-operative period was uneventful. There is no evidence of recurrence at review after one year.

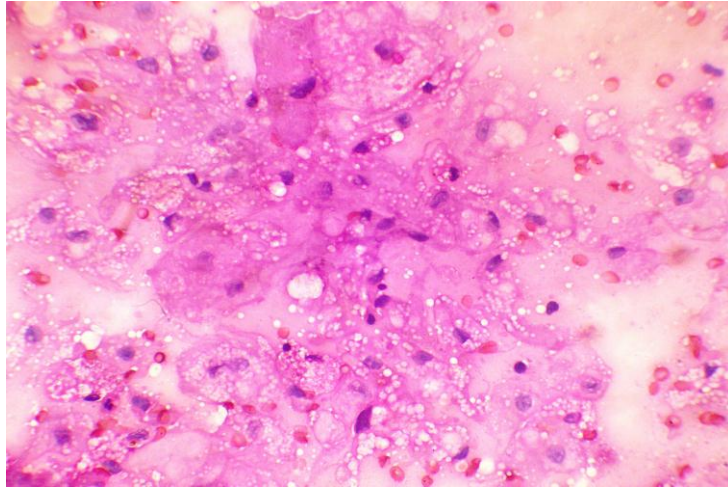


Figure 1: Cytological photomicrograph of the imprint smear shows a number of cells with large vacuoles (arrow) in the cytoplasm; (Rapid H & E, x400).

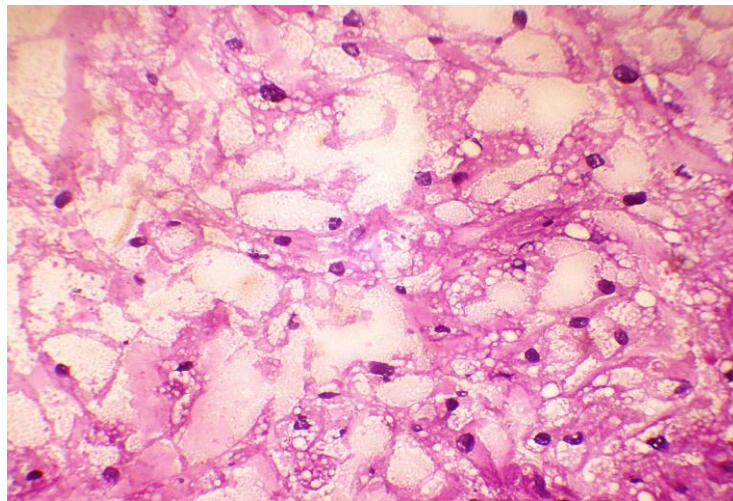


Figure 2: Photomicrograph showing large cells with mild nuclear atypia and multiple vacuoles in the cytoplasm; (H & E, x400).

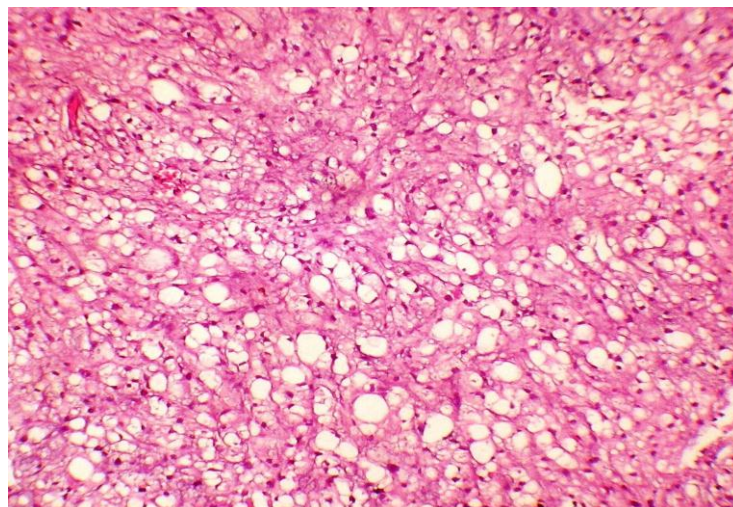


Figure 3: Photomicrograph showing groups and nests of physaliphorous cells embedded in a myxoid background; (H & E, x100).

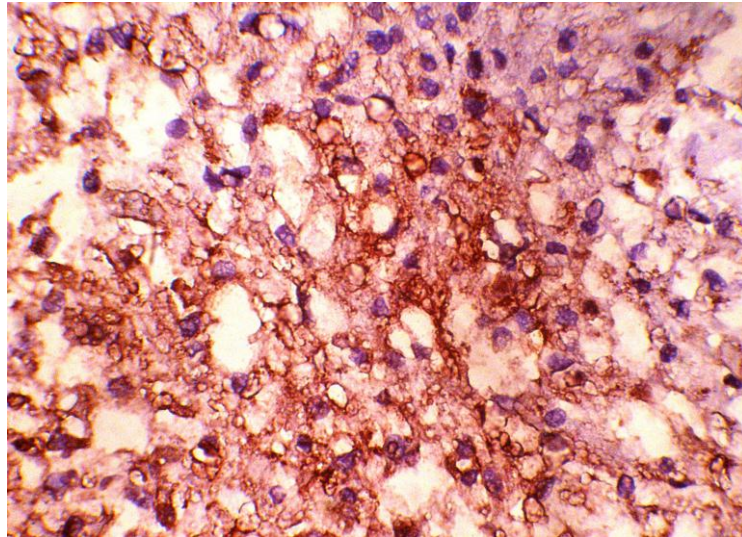


Figure 4: Immunohistochemical stain, the physaliphorus cells show positivity to antibodies against S100 protein; (x400).

DISCUSSION

Chordomas were first described by Virchow in 1857 as tumours made up of vacuolated or physaliferous cells derived from nests of embryonic notochord along the midline central nervous system axis. Chordomas are locally invasive and in advanced disease they can metastasize to bony structures like the sacrococcyx, skull base and vertebrae [2]. The incidence of chordoma metastasis in literature ranges between 3% and 48% in sites such as the spine, skin and other soft tissues, lymph nodes, lung, bone, liver and other intra-abdominal organs [1]. Some series have reported high rates and early occurrence of metastasis [3].

Chordomas affect males twice as often as females with pain being the main presenting symptom. They are distinctly uncommon in patients younger than 20 years of age, but occasional cases (<5%) in children have been described [4]. A study by comparative genomic hybridisation and fluorescent in-situ hybridization (FISH) suggested the involvement of tumour suppressor genes at 1p31 and 3p14 and an oncogene at 7q36 in the pathogenesis of chordoma [5].

Roentgenograms show irregular areas of destruction involving the midline of the sacrum, the body of the vertebrae and the clivus [6]. Grossly, chordomas tend to be soft, lobulated and have a gelatinous appearance. Rarely chordoma shows cystic changes [7]. Histologically lobulation and cording of tumour cells along with presence of physaliferous cells is considered diagnostic. Chordomas with chondroid differentiation have a better prognosis [8].

Although histopathologic features of chordomas are characteristic, at times it presents a diagnostic challenge especially in small biopsies, in its objective differentiation from other tumors such as chondrosarcoma, mucinous carcinoma or myxoid liposarcoma ^[9].

The provisional cytological diagnosis is possible in representative location as in our case when the characteristic multivacuolated physaliferous-like cells are present. However, a differential diagnosis of mesenchymal chondrosarcoma could not be excluded on imprint cytology and frozen section. But in metastatic sites, these cells can be confused with a variety of epithelial and mesenchymal tumors ^[10]. Immunohistochemistry (IHC) can help to distinguish chordomas from chondrosarcomas and other mucinous tumors. Chordomas express EMA, cytokeratin and S-100 as was seen in our case while chondrosarcomas are generally negative for EMA and cytokeratin. Brachyury appears to be a very sensitive and specific IHC marker for chordoma ^[11].

Transformation of a chordoma into a high grade spindle cell sarcoma has been described with irradiation ^[12]. The prognosis of patients with chordomas used to be poor because of the location of the tumours. Treatment in general consists of surgical excision or combined radiotherapy and surgery ^[1].

CONCLUSION

Intraoperative cytological and frozen section diagnosis of a rare neoplasm was an impetus for an attempt through excision in this case. Subsequent confirmation by immunohistochemistry on paraffin sections for the management and follow up. The most important tumors needing exclusion in this case were chondrosarcoma and other mucinous tumors.

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