

Metastatic chordoma detected by endobronchial ultrasound-guided transbronchial needle aspiration

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ABSTRACT

Chordomas are rare, slow-growing malignant bone tumours arising from cellular remnants of the notochord. These tumours are locally invasive but have also a metastatic potential.

Chordomas are characterized by the presence of physaliferous cells in a myxofibrillary stromal background. In cytological aspirates, these characteristic cells are usually absent, revealing only clusters of cells with varying degrees of vacuolation. This makes definitive diagnosis of chordoma difficult as the tumor can mimic other myxoid neoplasms including renal cell carcinomas and well-differentiated chondrosarcomas. In such situations, a confident diagnosis of chordoma requires comparison with histology of the primary tumor.

We describe the first case of metastatic chordoma diagnosed by endobronchial ultrasound-guided transbronchial needle aspiration (EBUS-TBNA).

KEY WORDS

Chordoma; metastasis; endobronchial ultrasound-guided transbronchial needle aspiration

J Thorac Dis 2013;5(1):90-93. DOI: 10.3978/j.issn.2072-1439.2012.12.01

Case report

A 76 year old female was transferred to the authors' institution from an outside hospital with back pain and a presacral mass on a background of progressively worsening fecal and urinary incontinence. Although she had a long history of low back pain and mild stress incontinence for approximately 20 years, this had become significantly worse in the previous few months. Furthermore, in the two weeks prior to presentation, she developed new onset faecal incontinence and her back pain became more severe with radiation to her right leg.

Apart from the above, she was otherwise well, had no other past medical or surgical history and was taking no regular medications. Her family history was positive for colorectal cancer. Her physical examination, hematology, biochemistry and urinalysis were all essentially normal.

An MRI scan of her abdomen and pelvis revealed a 15 cm mass

lesion extending from the lower sacrum inferiorly into the region of the posterior perineum along with extensive bony destruction of the mid and lower sacrum and coccyx (Figure 1A,B). It further showed that mass was causing significant indentation and displacement of the rectum as well as invading the lower spinal canal in the region of the sacrum with involvement of the thecal sac in this area.

A trucut biopsy of the primary mass revealed nests of large vacuolated tumor cells (physaliferous cells) in a prominent extracellular myxoid stroma (Figure 1C). Immunostains showed weak cytoplasmic positivity for S100 protein. The histology and immunohistochemistry were thus consistent with chordoma.

A routine chest x-ray incidentally showed an elevated right hemidiaphragm. A CT thorax further revealed multiple bilateral subcentimetre pulmonary nodules (0.4 to 0.8 cm in diameter) and several enlarged lymph nodes in the right hilum were noted (ranging from 1.1 to 1.4 cm in diameter) as well as a 0.9 cm precarinal node and a 1 cm subcarinal node (Figure 2A). Bronchoscopy and endobronchial ultrasound-guided transbronchial needle aspiration (EBUS-TBNA) of these nodes was therefore performed.

No endobronchial lesion was apparent. Multiple aspirates were taken from stations 4R, 7 and 11R. Four of six specimens collected showed loose aggregates of vacuolated cells (Figure 2B), which were similar in appearance to the physaliferous cells of

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Submitted Nov 04, 2012. Accepted for publication Dec 03, 2012.
Available at www.jthoracdis.com

ISSN: 2072-1439

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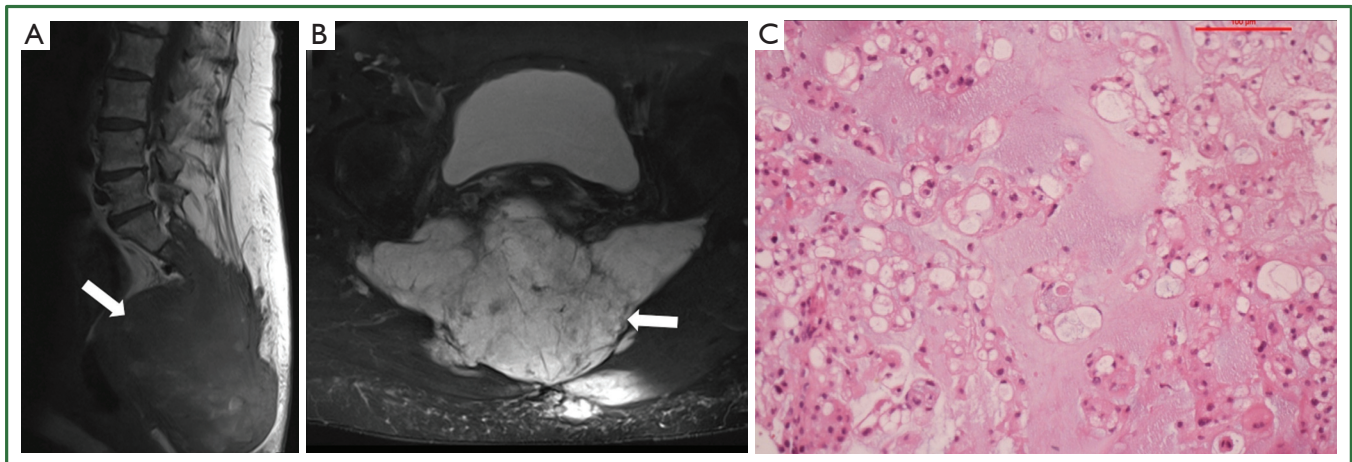


Figure 1. A. Sagittal T1-weighted MR image of the lumbosacral spine showing a large, expansile tumour (arrow) replacing most of the sacrum and extending anteriorly, displacing the rectum; B. transverse T2-weighted, fat-saturated image showing the typical appearances of sacral chordoma (arrow), with a predominantly high-signal mass extending laterally along the piriformis muscles; C. presacral mass biopsy showing the classical appearance of a chordoma with nests of large vacuolated cells (physaliferous cells) in an extracellular myxoid stroma (H&E, $\times 20$).

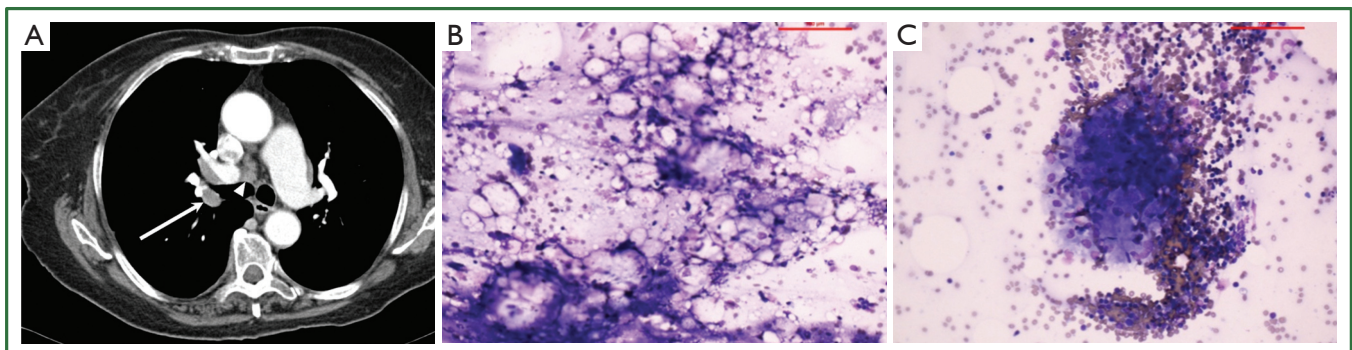


Figure 2. A. Transverse CT image of thorax with i.v. contrast showing a 1.4 cm enlarged right hilar lymph node, station 11R (arrow) and a precarinal lymph node, station 4R (arrowhead); B. EBUS-TBNA smear from hilar lymph node sample showing loose aggregates of vacuolated cells (MGG $\times 20$); C. EBUS-TBNA smear sample smear from hilar lymph node sample showing non-necrotising granuloma (MGG $\times 20$).

the primary specimen (Figure 2A), with associated sarcoid-like reaction (Figure 2C). Hence, the diagnosis of metastatic chordoma was made via EBUS-TBNA. In the evaluations of both the primary tumour and the EBUS-TBNA samples, there was agreement among two pairs of experienced consultant histopathologists, and the diagnosis of metastatic chordoma was facilitated by comparison with the primary tumour. Following discussions at a multidisciplinary team meeting, it was felt that the patient was not a candidate for surgery, and she received palliative radiation therapy. She passed away two months following the diagnosis.

Discussion

Chordomas are rare, slow-growing malignant bone tumors arising from remnants of the notochord (1). They account for 1-4% of primary malignant bone tumours and typically arise in the axial

skeleton, most often in the sacrococcygeal region or skull base (spheno-occipital chordoma) (2). These tumors are locally invasive but reported rates of metastasis vary between 10% and 42% of cases, though it has been suggested that misdiagnosis of mucus-producing carcinomas of the rectum as chordomas may have occurred in some chordoma case series (3-5). Metastases to the liver (6), brain (7), heart (8), skin (9), orbit (10) and ovaries (11) have all been reported, with rare documentation of metastases to the lung or mediastinum (12-16). Only two of these thoracic cases utilized cytological aspirates as the means of histological diagnosis: one via transcutaneous fine needle aspiration cytology (12) and one via exfoliative cytology from a sputum sample (13).

Chordomas are characterized by the presence of nests and cords of physaliferous cells in a myxofibrillary stromal background (17). In cytological aspirates, however, these characteristic cells are often absent, revealing only clusters of cells with varying degrees

of vacuolation. This makes definitive diagnosis of chordoma difficult as the tumor can mimic other myxoid neoplasms including renal cell carcinomas, myxoid chondrosarcomas, myxoid liposarcomas, metastatic mucinous adenocarcinomas, and myxopapillary ependymomas (18-21). Adjunctive histochemical, immunocytochemical (e.g., cytokeratin, epithelial membrane antigen, vimentin, and S-100 protein), and ultrastructural techniques can assist in supporting the cytological diagnosis of chordoma, but in aspirates where the characteristic physaliferous cells are absent (unlike in the presented case) a definitive diagnosis of metastatic chordoma requires comparison with histology of the primary tumor (which, in any event, was conducted in the current case) (21). The delay in diagnosis outlined in the current case, to the extreme of having widely metastatic disease at the time of diagnosis, likely reflects masking of the slow growth of the chordoma by the long standing history of back pain and stress urinary incontinence.

This is the first case report in the literature of metastatic chordoma diagnosed by EBUS-TBNA. EBUS-TBNA has become an established method for diagnosing mediastinal nodal metastases in lung cancer (22). However, this minimally invasive modality has increasingly been used in the diagnosis of mediastinal metastases of extrathoracic malignancies, including thyroid, renal, oesophageal cancers and rarer tumors such as chondrosarcomas (23-27). Moreover, improvements in adequacy of tissue sampling and specimen handling has allowed EBUS-TBNA specimens to be used, not only for histological and immunocytochemical analyses but also for multiple molecular analyses such as EGFR, ALK and KRAS mutations (28). In spite of these advances, definitive histological diagnosis is often not possible, especially in cases of rare tumors. However, as EBUS technology continues to evolve towards incorporated use of needle forceps and histology needles, the role of EBUS-TBNA in rare thoracic tumor sampling should continue to grow.

Acknowledgements

Disclosure: The authors declare no conflict of interest.

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Cite this article as: Fearon C, Fabre A, Heffernan EJ, Skehan SJ, Swan N, Keane MP, Butler MW. Metastatic chordoma detected by endobronchial ultrasound-guided transbronchial needle aspiration. *J Thorac Dis* 2013;5(1):90-93. DOI: 10.3978/j.issn.2072-1439.2012.12.01