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Case Report

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Case Report: A Rare Cause of Severe Osteomalacia: Infiltrating Phosphaturic Mesenchymal Tumor of the Mediastinum

Alexander Gawlitza[1*], Ioana-Medeea Țițu[2,3], Florin Teterea[3], Monica Mleşniţe[3], Alina Silaghi[4,5] and Emanuel Palade[2,3]

[1]*Iuliu Hatieganu University of Medicine and Pharmacy, 400000 Cluj-Napoca, Romania*

[2]*Department of Surgery, Iuliu Hatieganu University of Medicine and Pharmacy, 400000 Cluj-Napoca, Romania*

[3]*Thoracic Surgery Clinic, Leon Daniello Clinical Hospital of Pneumology, 400371 Cluj-Napoca, Romania*

[4]*5th Department of Internal Medicine, Iuliu Hatieganu University of Medicine and Pharmacy, 400348 Cluj-Napoca, Romania*

[5]*Endocrinology Clinic, Cluj County Emergency Clinical Hospital, 400347 Cluj-Napoca, Romania*

[*]Corresponding author

Alexander Gawlitza, student at the University of Medicine and Pharmacy Iuliu Hațieganu, Romania.

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ABSTRACT

Background: Phosphaturic mesenchymal tumors (PMTs) are rare and complex neoplasms that affect multiple systems, with complete surgical excision being the only definitive treatment.

Case: We present a rare case of a PMT with an unusual anatomical location and infiltrative character, further complicated by multiple preexisting comorbidities.

Conclusion: This case underscores the importance of early diagnosis and a multidisciplinary treatment approach, particularly in patients with complex medical histories.

Keywords: Phosphaturic Mesenchymal Tumor, Phosphaturia, Hypophosphatemia, Osteomalacia, Postoperative Complications

Introduction

Phosphaturic mesenchymal tumors (PMTs) are exceptionally rare neoplasms, with approximately 500 confirmed cases reported in the literature as of this writing [1]. Their epidemiology remains incompletely understood, though studies indicate peak incidence in middle-aged adults, with an overall balanced sex distribution despite some variation among tumor subtypes [2,3].

Determining the etiology of PMTs has posed a significant challenge. Recent research has linked their development to genetic factors, particularly the Fibronectin 1 - Fibroblast Growth Factor 1 and Fibronectin 1 - Fibroblast Growth Factor Receptor 1 fusion genes [4]. These tumors induce the paraneoplastic syndrome known as tumor-induced osteomalacia (TIO) via secretion of fibroblast growth factor 23 (FGF23). This hormone reduces renal phosphate reabsorption, leading to phosphaturia, hypophosphatemia, and ultimately, osteomalacia [5].

Clinical manifestations vary but typically include muscle weakness, bone pain, and pathological fractures [2]. Unlike neuroendocrine tumors (NETs), which originate from neuroendocrine cells and express specific markers, or adenomatous tumors derived from glandular epithelial tissue, PMTs arise from mesenchymal cells and lack neuroendocrine differentiation. This distinction is crucial for understanding their biological behavior and treatment strategies.

Although most PMTs are benign and found in extremities, occasional malignant cases have been reported [6]. Radiotherapy and chemotherapy have shown limited efficacy and are rarely used beyond second-line treatment. Supportive therapy,

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including phosphate supplementation and vitamin D analogs, has proven beneficial. However, complete surgical excision remains the only curative treatment, often leading to resolution of the paraneoplastic syndrome with a low recurrence rate [7].

This report presents a rare case of PMT with an unusual posterior mediastinal location and atypical infiltrating characteristics, additionally complicated by metabolic comorbidities.

Case Presentation

A 59-year-old Caucasian woman presented to our thoracic surgery department with severe vertebral column pain, costal pain exacerbated by respiration, cough, and vertigo when in the left lateral decubitus position. Radiological investigation revealed multiple pathological rib fractures (in at least 16 different sites). She also reported mixed-character pain in the coracobrachialis muscle, bilateral hip and knee pain, and localized mechanical lumbago, requiring assistance for ambulation. Her symptoms had progressively worsened over the past year, the chronic pain concomitantly impairing her mobility.

The patient had multiple comorbidities, including grade 1 obesity, poorly controlled type 2 diabetes mellitus, hypothyroidism, arterial hypertension, lumbar disc herniation, lacunar cerebral ischemia, and psychiatric conditions (depression and schizoaffective schizophrenia).

A thoraco-abdominal-pelvic contrast-enhanced computed-tomography (Figure 1) performed one month prior revealed a fusiform, inhomogeneous mass in the right supradiaphragmatic retrocrural space. The lesion, adjacent to T9–T12, extended medially into the paravertebral soft tissue, raising suspicion of malignancy. Additional findings included multiple retroperitoneal adenopathies, possible osteolysis on the right lateral surface of T11, and a hepatic lesion (segment III), consistent with hemangioma.

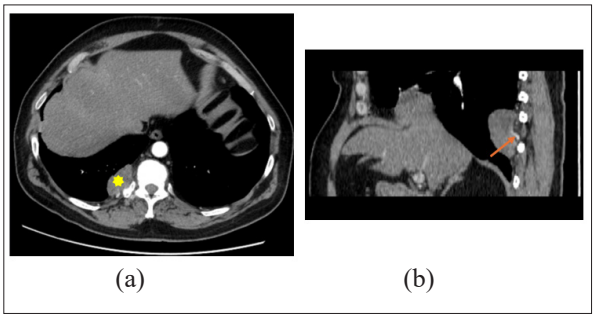

Figure 1: (a) Axial contrast-enhanced CT image showing a hypervascularized tumor (highlighted with a yellow star) located adjacent to the T9–T12 vertebral bodies. The mass extends medially into the paravertebral soft tissues. (b) Sagittal contrast-enhanced CT image demonstrating vascularization of the tumor from surrounding intercostal vessels (marked by the red arrow).

Surgical Intervention

Based on these findings, the patient underwent surgery for resection of the posterior mediastinal mass.

A triportal thoracoscopic approach was initially attempted. However, due to extensive tumor vascularization and high bleeding risk, the procedure was converted to a right anterolateral thoracotomy. The mass was successfully excised along with the corresponding right paravertebral pleura and soft tissue. Intraoperatively, multiple old rib fractures (T2–T12) were noted.

Postoperatively, the patient received analgesic treatment and thrombosis prophylaxis. Lung re-expansion was confirmed on imaging, and the surgical wounds healed per primam intentionem. Her initial favorable and uneventful evolution permitted the removal of a pleural drainage on the second postoperative day allowing for her to be discharged on the fifth day after the intervention.

Histopathological analysis confirmed the diagnosis of a phosphaturic mesenchymal tumor, showing immunohistochemistry profile: strong SATB2 and ERG1 positivity, negative CK AE1/3 and chromogranin, and a Ki67 index below 10%. Although malignization signs could not be confirmed (low mitotic score (below 2 mitoses/2 mm square)), the adjacent adipose tissue was found to contain isolated foci of tumor cells, demonstrating early tumoral spread.

Postoperative Complications & Readmission

Three days after discharge, the patient was readmitted with severe right-sided hemithoracic pain radiating to the ipsilateral hypochondrium and flank.

Neurological evaluation revealed thoracic myelopathy (T6–T10), radicular exteroceptive hypoesthesia in the right T6 territory, and acute urinary retention with overflow incontinence. A non-contrast thoracic CT revealed a small right-sided pleural collection, compressive right basal atelectasis, without signs of pneumonia or recent rib fractures.

Over the span of three weeks, the patient improved with multimodal analgesic therapy, including opioids, anticonvulsants, anti-inflammatories, and dietary supplements (vitamin D3, and B-group vitamins). An epidural catheter was temporarily placed for pain management. At discharge, follow-up recommendations included spinal MRI, contrast-enhanced CT at three months, and physical rehabilitation as active as the patient's state would allow.

Discussion

This case is unique due to the rarity of its presentation. PMTs are already exceptionally rare, but their mediastinal location accounts for fewer than 16% of cases [3], with malignant variants constituting only ~11% of all PMT cases and presenting more frequently in younger individuals [8].

The patient's comorbidities, particularly hypothyroidism and diabetes, likely contributed to the severity of her symptoms. Hypothyroidism affects calcium-phosphate homeostasis and disrupts vitamin D metabolism [9]. Obesity and chronic hyperglycemia impair bone metabolism and promote chronic inflammation, exacerbating bone fragility and muscular weakness [10].

A significant aspect in this case was the intense postoperative pain experienced by the patient. Caused by a combination of the consolidating pathological costal fractures and the thoracotomic intervention itself and further aggravated by the underlying TIO.

It was thus a multifactorial issue and more difficult to manage than most other post surgical pain. Severe bone pain is one of the few symptomatic hallmarks of PMTs, ultimately being caused by the tumor's paraneoplastic influence on bone metabolism. Although the administration of medication aiming at normalization of phosphate levels, like supplements, have shown analgesic efficacy in reduction of discomfort, complete surgical resection is still the only definitive treatment and has proven itself superior for cessation of pain [11]. In the case described above, the patient's severe aches in the early phase following the intervention can be attributed to her multiple still healing rib fractures and her refractory dysregulated bone metabolism. Surgical excision of the tumor can be considered to have prevented the patient from experiencing additional pain in the future.

This highlights the importance of metabolic dysfunctions as potential contributors to PMT symptomatology as well as their role as early warning signs. Given the absence of typical imaging features and the often-concealed location of PMTs, these tumors are frequently diagnosed based on laboratory findings such as low serum phosphate, elevated urinary phosphate, increased FGF23 levels, and high serum alkaline phosphatase. Therefore, regular monitoring of these parameters in suspected cases (patients with unexplained progressive bone pain) is not only essential for early tumor detection but also for identifying recurrence and preventing severe complications. Considering the limitations of conventional imaging methods, this approach could be more effective in managing PMTs, while PET-CT remains the most reliable imaging modality for localizing these tumors [11].

Preoperative embolization may be a valuable adjunct to reduce bleeding risk, potentially enabling less invasive surgical approaches and thus further minimizing the risk of complication occurrence [12].

Conclusion

This extraordinary case underscores the importance of recognizing PMTs in patients with metabolic dysfunctions.

It highlights the significance of FGF23 monitoring, and preoperative embolization to reduce intraoperative bleeding and facilitate surgical resection.

The management of postoperative pain in patients with complex underlying conditions, such as metabolic disturbances, requires a multidisciplinary approach. In this case, the patient's postoperative pain was influenced not only by the surgical trauma but also by her underlying metabolic dysfunction, particularly the phosphaturic imbalance caused by the tumor. This underlines that patients with metabolic disorders may have an increased susceptibility to heightened pain sensitivity, particularly following major surgeries. A tailored multifactorial pain management strategy is essential for effectively addressing both immediate surgical pain and the ongoing systemic metabolic challenges. In complex cases with systemic implications, the anticipation of pre-existing conditions should be a fundamental part of perioperative planning and approached from a broader management perspective-recognizing that surgical pain may be compounded by the patient's underlying disease processes. The need for continued physical rehabilitation and follow-up monitoring (especially regarding bone healing and metabolic levels) is vital for long-term recovery and deserves to be emphasized.

Highlights

- Rare and complex case: PMT with a mediastinal location and infiltrative features.
- Adaptive surgical approach: Demonstrates the necessity of intraoperative flexibility.
- Postoperative management: Emphasizes the role of meticulous rehabilitation in optimizing patient recovery.

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