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CASE REPORT PEER REVIEWED | OPEN ACCESS

Unexpected intruder: A rare case of mediastinal synovial sarcoma

Siddhesh Tryambake, Vindhya Vasini, SS Nirni, Chaitanya Kumari, Disha Morzaria, Ravi Wategaonkar

ABSTRACT

Introduction: Synovial sarcomas frequently manifest in the extremities of young people. A primary incidence in the mediastinum is exceedingly rare, with just a limited number of cases documented in global literature. We present a case of mediastinal synovial sarcoma.

Case Report: This report discusses a 37-year-old male who exhibited retrosternal chest discomfort and dyspnea with exertion. Imaging revealed an anterior mediastinal mass. The pathological examination of the excised mass revealed a monophasic tumor characterized by spindle cells possessing elongated oval hyperchromatic nuclei and mild eosinophilic cytoplasm. The tumor had strong staining for TLE-1 and vimentin, validating the diagnosis of monophasic synovial sarcoma.

Conclusion: The mediastinum encompasses a heterogeneous group of neoplasms, including both primary and metastatic, often posing significant diagnostic challenges. Synovial sarcoma, although uncommon in this location, should remain a key consideration in the differential diagnosis, and immunohistochemistry serves as an essential technique in this context. This case highlights the importance of recognizing this rare and aggressive tumor to guide appropriate diagnostic and optimize therapeutic decision making.

Keywords: Adjuvant chemotherapy, Immunohistochemistry, Mediastinal synovial sarcoma, Monophasic spindle cell tumor, SYT-SSX fusion

Siddhesh Tryambake[1], Vindhya Vasini[2], SS Nirni[3], Chaitanya Kumari[4], Disha Morzaria[5], Ravi Wategaonkar[1]

Affiliations: [1]Consultant Medical Oncologist, Department of Medical Oncology, MOC Cancer Care & Research Centre, Pune, India; [2]MD, DM, Senior Consultant, Department of Medical Oncology, Omega Hospitals, Hyderabad, India; [3]MD, DM, Senior Consultant and Head, Department of Medical Oncology, Omega Hospitals, Hyderabad, India; [4]DNB, Senior Consultant and Head, Department of Pathology, Omega Hospitals, Hyderabad, India; [5]Medical Writer, Department of Clinical Research, MOC Cancer Care & Research Centre, Mumbai, India

Corresponding Author: Siddhesh Tryambake, Consultant Medical Oncologist, MOC Cancer Care & Research Centre, Pune, India; Email: dr.srtryambake@gmail.com

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INTRODUCTION

Synovial sarcoma is a malignant mesenchymal tumor characterized by variable epithelial differentiation. It commonly arises in deep soft tissues of extremities particularly among adolescents and young adults [1]. Primary involvement of mediastinum is exceedingly rare, with only sporadic cases documented in the global literature [2]. When arising in mediastinum, synovial sarcoma poses a significant challenge due to its potential for local invasion and distant metastasis. Given its neoplastic clinical features and overlapping radiological features with other mediastinal masses, histopathological examination is essential for definitive diagnosis. We describe this case of a 10 cm primary mediastinal synovial sarcoma found in a 37-year-old male patient emphasizing the diagnostic conundrum associated with rare presentation.

CASE REPORT

A 37-year-old male exhibited symptoms of retrosternal chest pain, unrelated to physical activity or effort. He additionally reported dyspnea upon effort. There was an absence of cough, hemoptysis, fever, or weight loss in the medical history. No prior interaction with an active tuberculosis case. The physical examination was unremarkable and his initial laboratory tests revealed a hemoglobin level of 11.6 gm/dL, total leukocyte count of 5500 cells/mm³, and platelet count of 260,000/mm³. Serum electrolytes, renal function tests, and liver function tests were within normal limits. Tumor markers including lactate dehydrogenase (LDH), alpha-fetoprotein (AFP), and beta-human chorionic gonadotropin (beta-HCG) were within normal ranges. Contrast computed tomography (CT) of the chest demonstrated a lesion measuring 56 × 93 × 75 mm, involving both the anterior and middle mediastinum at the D2 to D5 level, with normal lung parenchyma present. The positron emission tomography-computed tomography (PET-CT) scan identified a conglomerated nodal mass measuring 6.8 × 9.9 cm, with necrosis in the anterior and middle mediastinum, encasing the mediastinal arteries, with a maximum standardized uptake value (SUV_{max}) of 17.3 and uninvolved lung parenchyma (Figure 1A and B). A CT-guided biopsy was performed. The diagnosis was identified as spindle cell neoplasm. The tumor was histologically comprised of spindle cells (Figure 2A and B). Multiple mitotic figures (exceeding four per ten high-power fields) were observed, along with localized regions of necrosis. Immunohistochemistry exhibited robust positivity for TLE-1 and Vimentin (Figure 3A and B). STAT6, S100, DESMIN, PCK, CD23, MUC 4, and the major pulmonary tumor marker (TTF-1) yielded negative results (Figure 3C illustrates the negatives for PCK). SYT-SSX1/2 was positive, confirming the diagnosis of monophasic synovial sarcoma. Consequently, the integration of histological characteristics and immunohistochemical results led to a diagnosis of primary monophasic synovial sarcoma of the mediastinum. The patient was assessed for surgery and had a median sternotomy with tumor removal. The postoperative recuperation was uneventful. The patient underwent adjuvant treatment with ifosfamide and adriamycin. The patient is undergoing frequent follow-up with no indications of residual or recurrent disease.

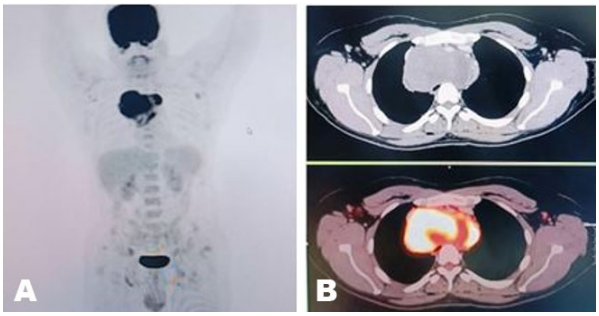

Figure 1: (A) Topographic image of index PET-CT with metabolically active mediastinal mass. (B) Cross-sectional image of index PET-CT s/o FDG avid mediastinal mass.

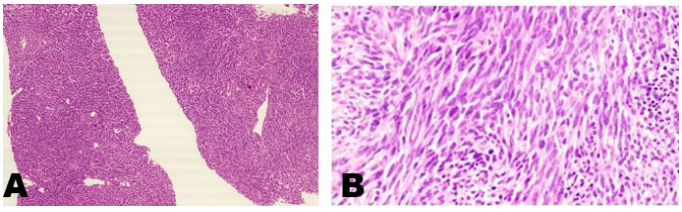

Figure 2: (A) Spindle cells in longitudinal and transverse section, on H and E, 10× (B) Elongated spindle cells on H and E, 40× with interspersed mitotic figures.

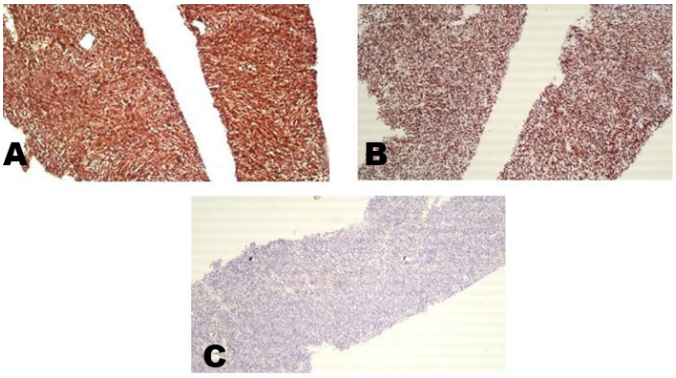

Figure 3: (A) Immunohistochemistry showing positivity for Vimentin, 10×. (B) Immunohistochemistry showing positivity for TLE-1, 10×. (C) Immunohistochemistry showing negativity for PCK, 10×.

DISCUSSION

The designation synovial sarcoma is a misnomer, as the tumor does not originate from the synovium; it only mimics synovial tissue under light microscopy. It seems to originate from multipotent stem cells that can differentiate into mesenchymal and/or epithelial structures and do not exhibit synovial differentiation [3, 4]. Fewer than 10–20% of synovial sarcomas occur in sites other than extremities [1, 5]. Soft tissue sarcomas, such as angiosarcoma, leiomyosarcoma (LMS), sarcomatoid mesothelioma, rhabdomyosarcoma (RMS), and synovial sarcoma, constitute fewer than 0.01% of all malignant thoracic neoplasms [6]. A major population-based study indicates that approximately 17% of new soft tissue sarcoma cases, encompassing various histologic subtypes, occur in thoracic regions such as the pleura, lungs, and mediastinum, with an estimated incidence of 6 per million individuals [7]. However, there is a lack of data that precisely documents the incidence of synovial sarcoma cases originating largely in the mediastinum. Synovial sarcomas are morphologically categorized into monophasic and biphasic subgroups. The biphasic type is characterized by the proliferation of bland spindle-shaped cells, accompanied by indications of epithelial development. Monophasic subtypes, conversely, may exhibit solely spindle cells or, on occasion, exclusively

an epithelial component [8]. A poorly differentiated type of synovial sarcoma is also acknowledged. The differential diagnosis of primary synovial sarcoma in the mediastinum is intricate due to the presence of numerous primary and metastatic cancers in this location. Immunohistochemistry is quite beneficial. Immunostains are crucial and corroborative for diagnosing a suspected case of synovial sarcoma. The presence of epithelial markers in both the glandular and spindle cell components substantiates the diagnosis. Epithelial membrane antigen (EMA) is the most common positive marker among the epithelial markers [9]. Pan-cytokeratin and cytokeratins 7 and 19 may also exhibit positivity in epithelial-like component and the spindle cell component [10]. Furthermore, bcl-2 positivity may be observed in certain instances [11]. Precise staging of the disease is essential for effective patient management and necessitates the evaluation of the primary tumor and the assessment for metastatic disease. Magnetic resonance imaging (MRI) has traditionally been the imaging modality for evaluating soft tissue masses; however, soft tissue sarcomas have lately been acknowledged for their heightened 18F-FDG uptake in PET; its added value in assessing metabolic activity, identifying nodal involvement, and aiding in comprehensive staging, which was critical for guiding surgical and therapeutic decisions [12]. Complete surgical resection remains the cornerstone of treatment, with margin-negative excision being the most significant determinant of long-term survival in primary mediastinal synovial sarcomas [13]. Radiotherapy is advised in those with positive margins [14]. The optimal role for chemotherapy administration for this tumor remains inadequately characterized, with scant research available [15]. The combination of adriamycin and ifosfamide has been utilized as adjuvant therapy and for recurrences [15]. Historically, synovial sarcoma has been seen as having a dismal prognosis, with metastases occurring in 50% of cases. The lung is the predominant site for metastasis, succeeded by lymph nodes and bone. Nonetheless, not all synovial sarcomas have the same unfavorable prognosis any longer. Spillane et al. established that a diagnosis age above 20 years and a tumor size greater than 5 cm correlated with a markedly poorer prognosis [16, 17]. Nevertheless, the prognosis with intensive multimodal therapy is favorable, with confirmed survival extending up to 14 years [15].

CONCLUSION

The mediastinum is an atypical location for synovial sarcoma, as seen in the index case, often contributing to diagnostic uncertainty. This case underscores the need to include synovial sarcoma in the differential diagnosis of spindle cell and biphasic mediastinal tumors, and highlights the critical role of timely clinical suspicion, histopathology, and targeted immunohistochemistry in confirming this rare diagnosis. An aggressive multimodal treatment approach is recommended, as demonstrated by our patient's favorable response to surgical resection followed by chemotherapy.

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Author Contributions

Siddhesh Tryambake – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Vindhya Vasini – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

SS Nirni – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Chaitanya Kumari – Conception of the work, Acquisition of data, Interpretation of data, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Disha Morzaria – Interpretation of data, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Ravi Wategaonkar – Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

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Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.

ABOUT THE AUTHORS

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Siddhesh Tryambake is a Consultant Medical Oncologist and Hemato-Oncologist at MOC Cancer Care & Research Center, Pune, India. He earned his undergraduate degree in M.B.B.S. from B.J. Government Medical College, Pune, and postgraduate degree M.D. in Radiation Oncology from Tata Memorial Hospital, Mumbai. He further obtained DrNB in Medical Oncology from the National Board of Examinations in Medical Sciences (NBEMS), New Delhi, and completed fellowships in Medical Oncology, Palliative Medicine, and Precision Radiation Oncology. He has published more than 10 research papers in national and international academic journals. His research interests include immunotherapy, targeted therapies, and integrated multimodality cancer care. He intends to pursue academic research and clinical excellence in solid and hematological malignancies in the future.
Email: dr.siddheshtryambake@gmail.com

Vindhya A Vasini, Best Medical Oncologist in Hyderabad. Expert in Breast Cancer, Head & Neck Tumors, Chemotherapy & Immunotherapy. Dr. Vindhya A Vasini, MBBS, MD (Radiotherapy), DM (Medical Oncology), is one of the top medical oncologists in Hyderabad, practicing at Omega Hospitals, Gachibowli. With years of experience in precision oncology, she provides personalized, evidence-based cancer care with compassion and clarity.
Email: drvindhya@gmail.com

SS Nirni, Best Hemato-Oncologist in Hyderabad. Expert in Blood Cancer, Chemotherapy & Bone Marrow Transplantation. Dr. SS Nirni, MBBS, MD (Internal Medicine), DM (Hemato-Oncology), is a leading medical oncologist and hemato-oncologist at Omega Hospitals, Gachibowli. With years of expertise in diagnosing and treating complex cancers, he is considered one of the top hemato-oncologists in Hyderabad, especially known for his skill in managing blood cancers and bone marrow disorders.
Email: Drssnirni123@gmail.com

Chaitanya Kumari is a renowned Pathologist with MD and DNB in pathology. She is certified in digital pathology with varied awards on her name. One of the most well-known pathologists in Hyderabad and Omega Hospitals, Gachibowli, Dr Chaitanya has keen interest in diagnostic precision oncology.

Disha Morzaria is Manager of Retrospective Research at MOC Cancer Care & Research Centre, Mumbai, India. She earned her undergraduate degree in Bachelor of Homeopathic Medicine and Surgery (BHMS) from Maharashtra University of Health Sciences, Nashik, and postgraduate certifications in Clinical Medicine, Gynecology & Obstetrics, and Child Health from the Maharashtra Board of Health Science, Pune. She further trained in Biostatistics and Clinical Research Methodology at Tata Medical Center and Scientific Writing in Health Research at ICMR-NIE. She has co-authored multiple abstracts and publications in national and international journals. She intends to advance oncology research and clinical data science.
Email: drdisharesearch@mocindia.co.in

Ravikumar Wategaonkar is a Senior Consultant in Medical Oncology at MOC Cancer Care & Research Centre, Pune, and also practices at Onco-Life Cancer Centre, Satara. He completed his DM in Medical Oncology from Gujarat Cancer Research Institute, Ahmedabad, and MD in Internal Medicine from Byramjee Jeejeebhoy Medical College, Pune. With multiple national and international publications and contributing in 20+ clinical trials as Principal investigator, his research interests include to advance cancer care through clinical research, innovative therapies, and patient-centered oncology practice.
Email: drraviwategaonkar@mocindia.co.in