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Thoracic SMARCA4-Deficient Undifferentiated Tumor Presenting as a Giant Mediastinal Mass: A Case Report

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Patient: Male, 67-year-old
Final Diagnosis: Thoracic SMARCA4-deficient undifferentiated tumor
Symptoms: Dyspnea
Clinical Procedure: —
Specialty: Pathology

Objective: Rare disease

Background: Thoracic SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A member 4 (SMARCA4)-deficient undifferentiated tumor (SMARCA4-UT) is a rare and highly aggressive thoracic malignancy. It predominantly affects male smokers and typically arises in the mediastinum, where rapid tumor growth and early metastasis contribute to a poor prognosis. Recent evidence suggests that this tumor exhibits biological heterogeneity and variable responses to therapy, underscoring the need for further clinical characterization.

Case Report: A 67-year-old man presented with dyspnea and superior vena cava syndrome. Computed tomography revealed a 7-cm mass in the anterior and superior mediastinum with suspected lymph node and bone metastases. Histological examination demonstrated a sheet-like proliferation of large atypical cells with rhabdoid features and necrosis. Immunohistochemical analysis showed loss of SMARCA4 (Brahma-related gene 1 [BRG1]) expression, positivity for cluster of differentiation 34 (CD34) and sex-determining region Y-box 2 (SOX2), weak epithelial membrane antigen expression, preserved integrase interactor 1 (INI1) expression, and negativity for other epithelial markers, fulfilling the diagnostic criteria for SMARCA4-UT. Despite palliative radiotherapy and combination immunotherapy with nivolumab and ipilimumab, the tumor rapidly progressed. The patient developed grade 4 drug-induced pneumonitis; transient stabilization was achieved, but his condition deteriorated. He died 6 months after disease onset. Autopsy revealed widespread metastases with minimal therapeutic effect, highlighting the aggressive clinical course and treatment resistance.

Conclusions: SMARCA4-UT is a highly aggressive tumor requiring comprehensive immunohistochemical evaluation for accurate diagnosis. This case highlights the limited efficacy of immune checkpoint inhibitors in a PD-L1-negative setting and underscores the need for more effective therapeutic strategies.

Keywords: Case Reports • Histological Techniques • Immunohistochemistry • Mediastinal Neoplasms • Oncology • SMARCA4 Protein

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Introduction

SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A member 4 (SMARCA4)-deficient undifferentiated tumor (SMARCA4-UT) is a recently established entity characterized by loss of SMARCA4 (Brahma-related gene 1 [BRG1]) expression. Initially described in 2015 as "SMARCA4-deficient thoracic sarcoma," this tumor was noted for its undifferentiated morphology and extremely poor prognosis [1,2]. Subsequent studies revealed that some tumors exhibit partial epithelial differentiation, leading to their reclassification as a distinct entity, "SMARCA4-deficient undifferentiated tumor," in the 5th edition of the World Health Organization (WHO) Classification of Thoracic Tumors [3,4].

Clinically, SMARCA4-UT predominantly affects middle-aged male smokers and most commonly arises in the mediastinum or hilar region [5-7]. Tumors typically grow rapidly, with distant metastases frequently present at diagnosis, including involvement of lymph nodes, bone, and adrenal glands [8]. Patients often exhibit symptoms such as dyspnea, superior vena cava syndrome, or chest pain [9]. Despite increasing recognition of this entity, its clinicopathological characteristics and optimal management strategies remain incompletely understood. Here, we report a case of rapidly progressive SMARCA4-UT arising in the anterior mediastinum and describe its pathological features, clinical course, and autopsy findings in the context of existing literature.

Case Report

A 67-year-old man presented with dyspnea. His medical history included paroxysmal atrial fibrillation, right brainstem infarction, hypertension, and dyslipidemia. He had a smoking history of 10 to 20 cigarettes per day from 20 to 66 years of age (smoking index: approximately 460-920) and no notable family history. One month before admission, he developed supine dyspnea and numbness extending from the left neck to the shoulder. A mediastinal mass was incidentally detected during routine screening. Chest computed tomography (CT) showed a 6-cm anterior mediastinal mass, and he was subsequently referred to our institution (**Figure 1A**).

Contrast-enhanced CT demonstrated a 7-cm mass extending from the anterior to superior mediastinum, with suspected cervical, supraclavicular, and mediastinal lymphadenopathy. Positron emission tomography-CT revealed intense fluorodeoxyglucose uptake in the mediastinal tumor, right cervical lymph nodes, and left C4 vertebral body (**Figure 1B**). The tumor extended into the cervical region, causing superior vena cava and cervical venous stenosis with upper-body edema, which resulted in superior vena cava syndrome and severe tracheal stenosis. A tracheal stent was subsequently placed. Transbronchial needle aspiration was nondiagnostic; CT-guided biopsy established the diagnosis of SMARCA4-UT approximately 2 months after symptom onset.

Three weeks after admission, palliative radiotherapy (30 Gy) was administered to the mediastinum and cervical spine. Recurrent sputum retention, stent obstruction, and pneumonia complicated respiratory management; a tracheostomy was performed 1 month later. Combination immunotherapy with nivolumab and ipilimumab was initiated. The tumor did not display programmed death-ligand 1 (PD-L1).

Three months after admission, the patient developed grade 4 drug-induced pneumonitis with respiratory failure; he was treated with high-dose methylprednisolone followed by tapering. Although transient disease stabilization was achieved,

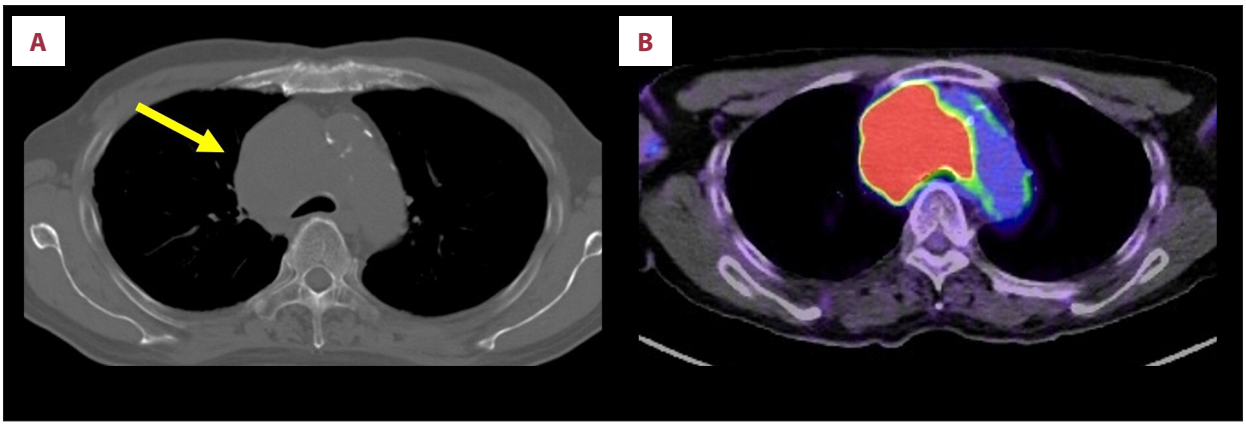

Figure 1. Radiological findings of the mediastinal tumor. (**A**) Non-contrast chest computed tomography showing a large mass measuring up to 7 cm in maximum diameter, extending from the anterior to superior mediastinum (yellow arrow). (**B**) Positron emission tomography-computed tomography demonstrating intense fluorodeoxyglucose uptake (shown in red) in the region corresponding to the anterior mediastinal tumor.

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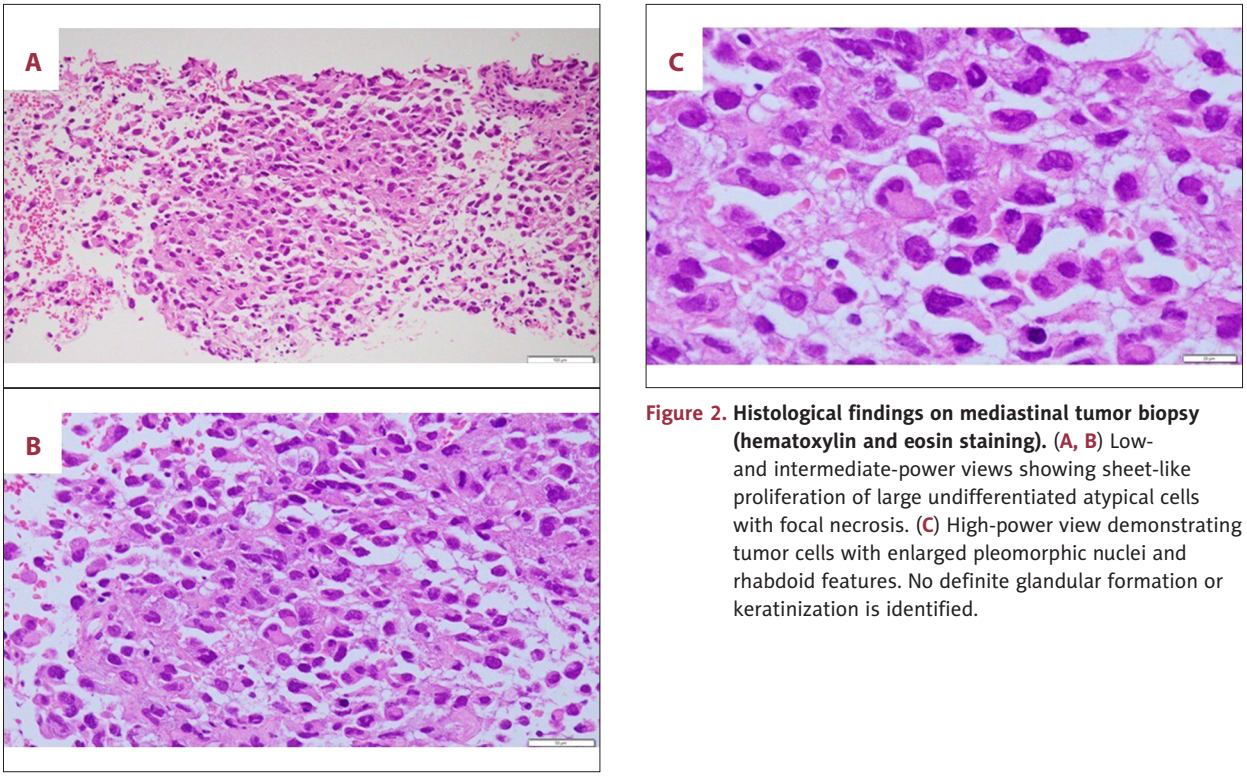

Figure 2. Histological findings on mediastinal tumor biopsy (hematoxylin and eosin staining). (**A, B**) Low- and intermediate-power views showing sheet-like proliferation of large undifferentiated atypical cells with focal necrosis. (**C**) High-power view demonstrating tumor cells with enlarged pleomorphic nuclei and rhabdoid features. No definite glandular formation or keratinization is identified.

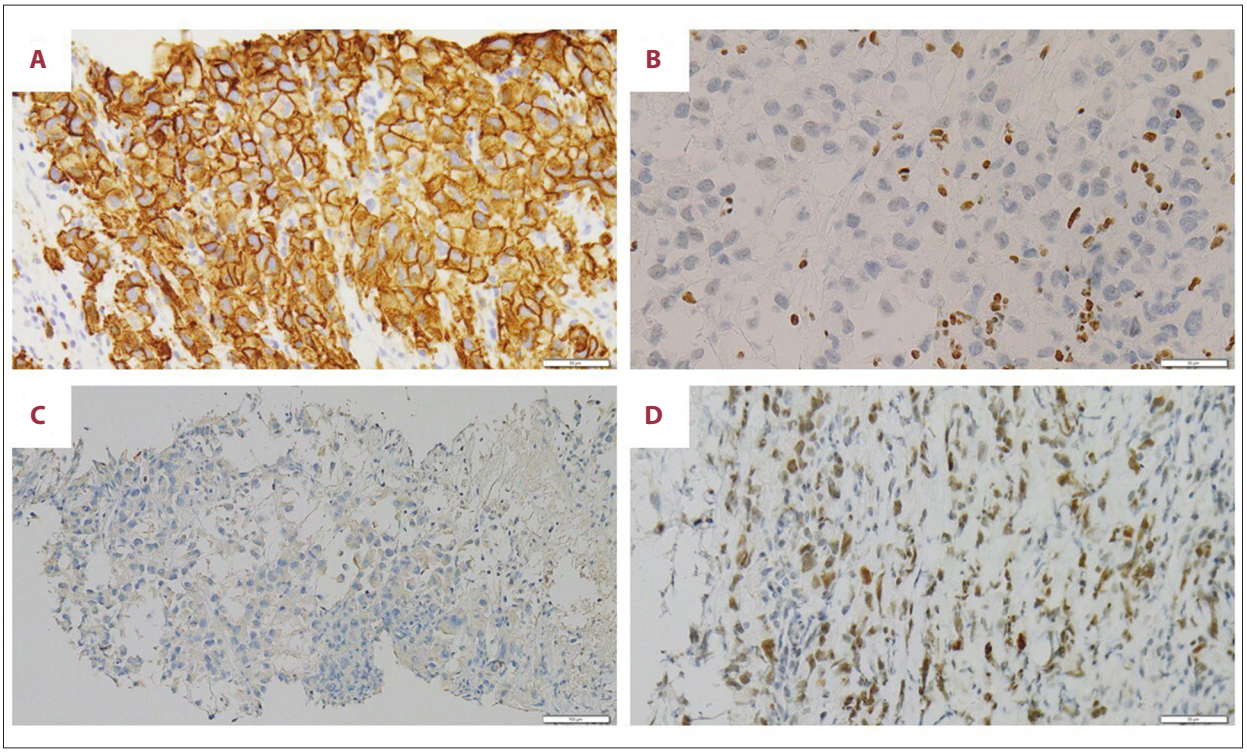

Figure 3. Immunohistochemical findings of the tumor. (**A**) Tumor cells showing diffuse positivity for CD34. (**B**) Complete loss of SMARCA4 (BRG1) expression in tumor cells, with preserved expression in non-neoplastic cells serving as an internal control. (**C**) Tumor cells showing SALL4 negativity. (**D**) Nuclear positivity for SOX2 in tumor cells. Abbreviations: BRG1, Brahma-related gene 1; CD34, cluster of differentiation 34; SALL4, Sal-like protein 4; SMARCA4, SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily A member 4; SOX2, sex-determining region Y-box 2.

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cervical lymphadenopathy, bilateral upper-extremity edema, and pleural effusions progressed 5 months after admission. Thus, palliative care was initiated. No pleural intervention was performed for the progressive pleural effusions. The patient died 6 months after disease onset. Autopsy revealed widespread metastases. Death was attributed to respiratory failure secondary to pneumonia, diffuse alveolar damage, and massive pleural effusion.

Pathological Findings

Histological examination of a biopsy specimen from the superior mediastinal mass showed sheets of large, round-to-polygonal atypical cells with focal necrosis (**Figure 2A, 2B**). The tumor cells demonstrated enlarged pleomorphic nuclei and rhabdoid features (**Figure 2C**). No definite glandular formation or keratinization was observed.

Immunohistochemical analysis revealed positivity for cluster of differentiation 34 (CD34) and sex-determining region Y-box 2 (SOX2), along with complete loss of SMARCA4 (BRG1) expression and preserved integrase interactor 1 (INI1 [SMARCB1]) expression. Sal-like protein 4 (SALL4) was not detected, epithelial membrane antigen showed weak positivity, and most other epithelial markers were absent (**Figure 3A-3D**).

Autopsy Findings

On autopsy, the primary tumor was identified as an ill-defined grayish-white mass in the anterior mediastinum measuring approximately 12×8.5×6 cm. Histologically, the tumor consisted of large round-to-polygonal atypical cells with enlarged pleomorphic nuclei proliferating in alveolar, trabecular, and solid patterns, consistent with findings in the biopsy specimen. Focal necrosis and fibrotic stroma were present. Fibrosis was less prominent in metastatic lesions, where a greater number of viable tumor cells remained.

Tumor invasion and metastases were observed in the trachea, cervical soft tissues, thyroid gland, infrahyoid muscles, liver, bilateral adrenal glands, bilateral lungs, right atrial appendage, small intestine, and multiple lymph nodes, including mediastinal, bilateral cervical, and mesenteric nodes. Pulmonary tumor involvement was limited, supporting the mediastinum as the primary site. Treatment-related necrosis was minimal, indicating only a mild therapeutic effect.

Associated findings included intra-alveolar aggregates of foamy macrophages, consistent with recurrent pneumonia. Acute neutrophil-predominant bronchopneumonia was present in the right middle lobe, with focal keratinous debris suggestive of aspiration. Partial hyaline membrane formation in both lungs indicated diffuse alveolar damage.

A large volume of clear yellow pleural effusion was present without pleural tumor involvement, suggesting a nonmalignant etiology related to circulatory disturbance or reduced oncotic pressure. The cause of death was respiratory failure due to pneumonia, accompanied by massive pleural effusion and diffuse alveolar damage.

Discussion

This report describes a case of SMARCA4-UT arising in the anterior mediastinum. This recently recognized thoracic malignancy is characterized by loss of SMARCA4 (BRG1), a catalytic adenosine triphosphatase subunit of the SWItch/Sucrose Non-Fermentable (SWI/SNF) chromatin remodeling complex [1,2,5]. The SWI/SNF complex regulates chromatin accessibility and transcriptional activity through adenosine-triphosphate-dependent nucleosome remodeling and plays a crucial role in cellular differentiation, proliferation, and DNA repair [10]. Loss of SMARCA4 disrupts these regulatory processes and promotes dedifferentiation, genomic instability, and aggressive tumor behavior [1,5]. Additionally, SMARCA4 deficiency has been associated with alterations in the tumor immune microenvironment, which may partly explain the reported responsiveness of some tumors to immune checkpoint inhibitors [11].

SMARCA4-UT predominantly affects middle-aged male smokers and most commonly arises in the mediastinum, lung hilum, or pleura. It is characterized by rapid growth, early metastasis, and an extremely poor prognosis [5,7,12]. Radiologic findings are nonspecific and require differentiation from thymic epithelial tumors, germ cell tumors, malignant lymphoma, and other mediastinal malignancies [5,6]. Histologically, SMARCA4-UT typically shows sheets of large atypical cells with prominent nucleoli, abundant cytoplasm, and frequent rhabdoid features and is often accompanied by extensive necrosis and high mitotic activity [1,3]. The defining feature is loss of SMARCA4 (BRG1) expression, often accompanied by CD34 and SOX2 expression and preserved INI1 (SMARCB1) expression [2,3,5]. Additional markers reported to be useful in the diagnostic evaluation include SMARCA2, SALL4, and claudin-4 [2,5,6]. We note that SMARCA2 immunohistochemistry analysis was not performed in the present case. In the present case, histological findings led to a broad differential diagnosis, including undifferentiated carcinoma, malignant mesothelioma, malignant melanoma, malignant lymphoma, granulocytic sarcoma, rhabdomyosarcoma, germ cell tumor, epithelioid sarcoma, and thymic tumor. However, these entities were excluded based on the immunohistochemical profile.

Previous reports and case series indicate that the mediastinum or pleura is the most common primary site and outcomes are uniformly poor (**Table 1**) [1,2,6-8,12,13]. Median

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Table 1. Clinical features, treatment strategies, and outcomes of SMARCA4-deficient tumors reported in the literature.

Study	Year	No. of cases	Age, years (median, range)	Sex (male/female)	Smoking history	Primary site	PD-L1 expression	Treatment	Outcome
Le Loarer et al [1]	2015	19	41 (28-72)	16/3	17/19 smokers	Mediastinum/pleura/lung	ND	Surgical resection; chemotherapy	Median OS: 7 months
Yoshida et al [2]	2017	12	39 (27-82)	11/1	Predominantly smokers	Chest wall/thoracic cavity/mediastinum/lung	ND	Surgical resection; chemotherapy; radiotherapy	Median OS: 7 months
Perret et al [6]	2019	30	48 (28-90)	27/3	Predominantly smokers	Mediastinum/pleura/lung	ND	Chemotherapy	Median OS: 6 months
Zhou et al [7]	2024	20	61 (39-72)	20/0	Predominantly smokers	Lung/mediastinum/pleura/chest wall	ND	Various systemic therapies	Median OS: 5.6 months
Rekhtman et al [8]	2020	22	58 (30-80)	16/6	Predominantly smokers	Lung/mediastinum/lymph node/bone	ND	ND	Median OS: 5.2 months
Ohta et al [12]	2025	Systematic review (160)	58 (18-88)	112/48	Frequent	Thoracic (40%); digestive (17.5%); gynecologic (15.6%)	TPS ≥1%: 28/33; <1%: 4/33; ND: 1/33	Chemotherapy; immunotherapy; surgery	Median OS: 5.0 months
Shinno et al [13]	2022	18	53 (32-80)	14/4	18/18 smokers	Thoracic	TPS ≥50%: 2/11; 1-49%: 5/11; < 1%: 4/11	Immune checkpoint inhibitors; chemotherapy	Median PFS: 2.4 months
Present case	–	1	67	1/0	Smoker	Anterior mediastinum	Negative	Radiotherapy + nivolumab/ipilimumab	OS: 6 months

Abbreviations: ND, not described/not available; OS, overall survival; PD-L1, programmed death-ligand 1; PFS, progression-free survival; TPS, tumor proportion score.

overall survival is approximately 5 to 7 months, reflecting the extremely aggressive nature of this disease. In the present case, a rapidly enlarging tumor arose in the anterior mediastinum, causing superior vena cava syndrome and severe tracheal stenosis. Histologically, the tumor showed sheet-like proliferation of large undifferentiated cells with rhabdoid features and extensive necrosis. Immunohistochemical analysis demonstrated loss of SMARCA4 expression, accompanied by CD34 and SOX2 expression and preserved INI1 expression, consistent with previously reported features of SMARCA4-UT [2,5,6]. Given the morphological overlap with various undifferentiated malignancies, diagnosis can be challenging; however, the combination of epithelial marker negativity and CD34 positivity provides important diagnostic clues.

The differential diagnosis of mediastinal SMARCA4-UT includes several malignant tumors [2,3,5,6]. Thymic epithelial tumors, particularly thymic carcinoma, may show overlapping clinical and radiologic features; however, these tumors usually retain SMARCA4 expression and demonstrate positivity for epithelial markers such as cytokeratins and claudin-4 [3,5]. Mediastinal germ cell tumors also represent an important differential diagnosis, especially when tumors exhibit undifferentiated morphology; they typically express germ cell markers such as SALL4, octamer-binding transcription factor 3/4 (OCT3/4), or placental alkaline phosphatase [3,5]. Malignant lymphoma may also manifest as a rapidly enlarging mediastinal mass but can be distinguished by expression of hematolymphoid markers such as CD45 [3]. Nuclear protein in testis (NUT) carcinoma should also be considered and can be identified by diffuse

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nuclear positivity on NUT immunostaining [14]. Epithelioid sarcoma and malignant rhabdoid tumor may also exhibit rhabdoid morphology; however, these tumors typically show loss of INI1 (SMARCB1) expression, whereas INI1 expression is preserved in SMARCA4-UT [3,5,15].

According to the 5th edition of the WHO Classification of Thoracic Tumors, SMARCA4-UT is defined as an adult-onset intrathoracic tumor composed of discohesive round-to-epithelioid cells growing in sheets, lacking definite epithelial differentiation, and demonstrating loss of SMARCA4 expression [3]. Desirable features include loss of SMARCA2 and expression of CD34 and SOX2. In the present case, the absence of epithelial differentiation along with characteristic immunohistochemical findings fulfilled the WHO diagnostic criteria, supporting a diagnosis of SMARCA4-UT.

Although SMARCA4 alterations occur in a subset of non-small cell lung cancers [16], most represent conventional carcinomas, highlighting the need for comprehensive immunohistochemical assessment to distinguish SMARCA4-UT from other SMARCA4-altered tumors. Additionally, SMARCA4-deficient lung carcinoma may share similar molecular alterations; however, these tumors usually demonstrate more evident epithelial differentiation and are associated with a primary lesion in the lung parenchyma [2,4,7,8]. Thus, the diagnosis of SMARCA4-UT requires careful integration of morphologic findings, tumor location, and comprehensive immunohistochemical evaluation [3,5,6].

Recent evidence suggests that the tumor immune microenvironment of SMARCA4-deficient tumors is heterogeneous and can be broadly categorized into "immune-desert" and "immune-active" phenotypes [11,17]. Immune-desert tumors are characterized by minimal infiltration of cytotoxic T lymphocytes and frequently exhibit low PD-L1 expression, which may result in poor responses to immune checkpoint inhibitors [11,17]. Conversely, immune-active tumors demonstrate increased immune cell infiltration and may exhibit greater sensitivity to immunotherapy [11,13,17]. Differences in the tumor immune microenvironment may partly explain the heterogeneous responses to immune checkpoint inhibitors documented among patients with SMARCA4-deficient tumors [11,13]. This concept may also explain the limited responses to nivolumab and ipilimumab observed in the present case.

Although immune checkpoint inhibitors have shown activity in some cases, including PD-L1-negative tumors [11,13,18], the response in the present case was limited, underscoring the aggressive and treatment-resistant nature of SMARCA4-UT [5,6]. Investigators have recently proposed combination therapeutic strategies incorporating immune checkpoint inhibitors, anti-angiogenic agents, and chemotherapy for highly aggressive thoracic malignancies, including SMARCA4-deficient tumors [19-21]. Reports suggest that such combination approaches can provide superior clinical outcomes relative to dual immunotherapy alone. These multi-agent regimens, sometimes referred to as "quadruplet therapy," may enhance antitumor effects by improving tumor vascular normalization and immune activation [19-21]. Although rapid clinical deterioration in the present case limited the feasibility of intensive treatment, such strategies may represent therapeutic options for selected patients in future clinical practice [19]. Autopsy findings concerning our patient revealed extensive local invasion centered in the mediastinum, accompanied by multiple metastatic lesions involving lymph nodes, bone, liver, adrenal glands, and other organs. These findings further support the highly invasive and metastatic nature of SMARCA4-UT. Additional cases and molecular studies are warranted to refine the diagnostic criteria and develop effective therapies [5,12,18]. Overall, the present case illustrates the typical clinical and pathological features of SMARCA4-UT and reinforces the extremely aggressive clinical course associated with this tumor.

Conclusions

We report a rare case of SMARCA4-UT arising in the anterior mediastinum that was associated with an aggressive clinical course and poor prognosis. Accurate diagnosis required comprehensive immunohistochemical evaluation, particularly demonstration of SMARCA4 (BRG1) loss along with CD34 positivity and the absence of epithelial marker expression. The limited therapeutic response in this case underscores the highly aggressive and treatment-resistant nature of SMARCA4-UT. Accumulation of additional cases is essential to improve diagnostic accuracy and establish more effective therapeutic strategies for this rare entity.

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Patient Consent

Written informed consent was obtained from the patient (or the patient's next of kin) for publication of this case report and any accompanying images.

Declaration of Figures' Authenticity

All figures submitted have been created by the authors who confirm that the images are original with no duplication and have not been previously published in whole or in part.

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References:

1. Le Loarer F, Watson S, Pierron G, et al. SMARCA4 inactivation defines a group of undifferentiated thoracic malignancies transcriptionally related to BAF-deficient sarcomas. Nat Genet. 2015;47(10):1200-5
2. Yoshida A, Kobayashi E, Kubo T, et al. Clinicopathological and molecular characterization of SMARCA4-deficient thoracic sarcomas with comparison to potentially related entities. Mod Pathol. 2017;30(6):797-809
3. Yoshida A. Thoracic SMARCA4-deficient undifferentiated tumour. In: WHO Classification of Tumours Editorial Board, editors. WHO Classification of Tumours of the Lung, Pleura, Thymus and Heart. 5th ed. Lyon: IARC; 2021;111-14
4. Nicholson AG, Tsao MS, Beasley MB, et al. The 2021 WHO classification of lung tumors: Impact of advances since 2015. J Thorac Oncol. 2022;17(3):362-87
5. Nambirajan A, Jain D. Recent updates in thoracic SMARCA4-deficient undifferentiated tumor. Semin Diagn Pathol. 2021;38(5):83-89
6. Perret R, Chalabreysse L, Watson S, et al. SMARCA4-deficient thoracic sarcomas: Clinicopathologic study of 30 cases with an emphasis on their nosology and differential diagnoses. Am J Surg Pathol. 2019;43(4):455-65
7. Zhou P, Fu Y, Tang Y, et al. Thoracic SMARCA4-deficient tumors: A clinicopathological analysis of 52 cases with SMARCA4-deficient non-small cell lung cancer and 20 cases with thoracic SMARCA4-deficient undifferentiated tumor. PeerJ. 2024;12:e16923
8. Rekhtman N, Montecalvo J, Chang JC, et al. SMARCA4-deficient thoracic sarcomatoid tumors represent primarily smoking-related undifferentiated carcinomas rather than primary thoracic sarcomas. J Thorac Oncol. 2020;15(2):231-47
9. Jiang J, Chen Z, Gong J, et al. Thoracic SMARCA4-deficient undifferentiated tumor. Discov Oncol. 2023;14(1):51
10. Reddy D, Bhattacharya S, Workman JL. (mis)-Targeting of SWI/SNF complex(es) in cancer. Cancer Metastasis Rev. 2023;42(2):455-70
11. Gantzer J, Davidson G, Vokshi B, et al. Immune-desert tumor microenvironment in thoracic SMARCA4-deficient undifferentiated tumors with limited efficacy of immune checkpoint inhibitors. Oncologist. 2022;27(6): 501-11. doi: 10.1093/oncolo/oyac040
12. Ohta R, Yamamoto N, Tanaka K, Sano C, Hayashi H. Clinical characteristics and outcomes of SMARCA4-mutated or deficient malignancies: A systematic review of case reports and series. Cancers (Basel). 2025;17(16):2675
13. Shinno Y, Yoshida A, Masuda K, et al. Efficacy of immune checkpoint inhibitors in SMARCA4-deficient thoracic tumor. Clin Lung Cancer. 2022;23(5):386-92
14. French CA. NUT carcinoma: Clinicopathologic features, pathogenesis, and treatment. Pathol Int. 2018;68(11):583-95
15. Agaimy A. The expanding family of SMARCB1(INI1)-deficient neoplasia: Implications of phenotypic, biological, and molecular heterogeneity. Adv Anat Pathol. 2014;21(6):394-410
16. Schoenfeld AJ, Bandlamudi C, Lavery JA, et al. The genomic landscape of SMARCA4 alterations and associations with outcomes in patients with lung cancer. Clin Cancer Res. 2020;26(21):5701-8
17. Wang X, Shi W, Xiao Y, et al. The immune desert tumor microenvironment in thoracic SMARCA4-deficient undifferentiated tumors: Spatial distribution characteristics and prognostic significance. Lung Cancer. 2025;210:108817
18. Shinno Y, Ohe Y; Lung Cancer Study Group of the Japan Clinical Oncology Group (JCOG). Thoracic SMARCA4-deficient undifferentiated tumor: Current knowledge and future perspectives. Jpn J Clin Oncol. 2024;54(3):265-70
19. Longo V, Catino A, Montrone M, et al. Treatment of thoracic SMARCA4-deficient undifferentiated tumors: Where we are and where we will go. Int J Mol Sci. 2024;25(6):3237
20. Shi M, Chen X, Lin T, et al. Efficacy of antiangiogenic therapy in patients with advanced SMARCA4-deficient thoracic tumor. Lung Cancer. 2025;202:108498
21. Liu B, Wang D, Li Y, et al. Potential efficacy of anti-angiogenic therapy combined with immunotherapy for advanced SMARCA4-deficient thoracic tumor: A retrospective study. Transl Lung Cancer Res. 2025;14(9):3430-43

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