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Case Report

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Mediastinal PEComa with TFE3 Rearrangement: An Unusual Finding

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Introduction

Perivascular epithelioid cell tumors (PEComas) are rare mesenchymal neoplasms classified by the World Health Organization (WHO) as tumors of uncertain differentiation. They are composed of perivascular epithelioid cells (PECs), which are characterized by their distinctive morphology, close association with blood vessel walls, and dual expression of melanocytic and smooth muscle markers [1].

Case Report

A 46-year-old man with no past medical history presented to the pulmonology department with a dry cough and exertional dyspnea that had been progressing for the past 3 months. These symptoms were accompanied by a persistent chest pain for the past 3 weeks. Clinical examination was unremarkable. A thoracic CT scan revealed a well-defined, heterogeneous, solid-cystic mass measuring 7 × 4.8 × 3.5 cm, located in the right mediastinum. Surgical resection of the mass was performed. (Figure 1)

Histological examination of sections from the mass revealed a tumor proliferation arranged in nests and clusters within a myxoid background. The tumor cells were predominantly spindle-shaped, with abundant eosinophilic cytoplasm. Rare mitotic figures were observed. (Figure 2)

Immunohistochemically, the tumor cells showed positive staining for smooth muscle actin (SMA), H-caldesmon, Melan A, TFE3, and CD34, while staining was negative for HMB-45, AE1/AE3, S100, and EMA. (Figure 3)

Based on the morphological features and immunohistochemical profile, the diagnosis of PEComa (perivascular epithelioid cell tumor) was established.

Figure 1: Macroscopic Appearance of the Resected Mediastinal Tumor

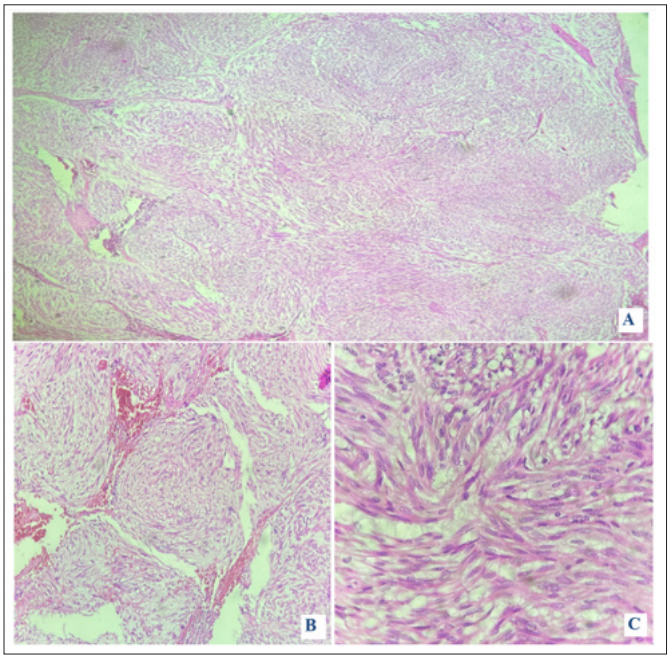

Figure 2: Histological Aspects of PEComa in Hematoxylin and Eosin staining (H&E) Showing a Tumor Proliferation with Spindle-Shaped Cells; (A) H&x10; (B) H&E x 20; (C) H&E x 40

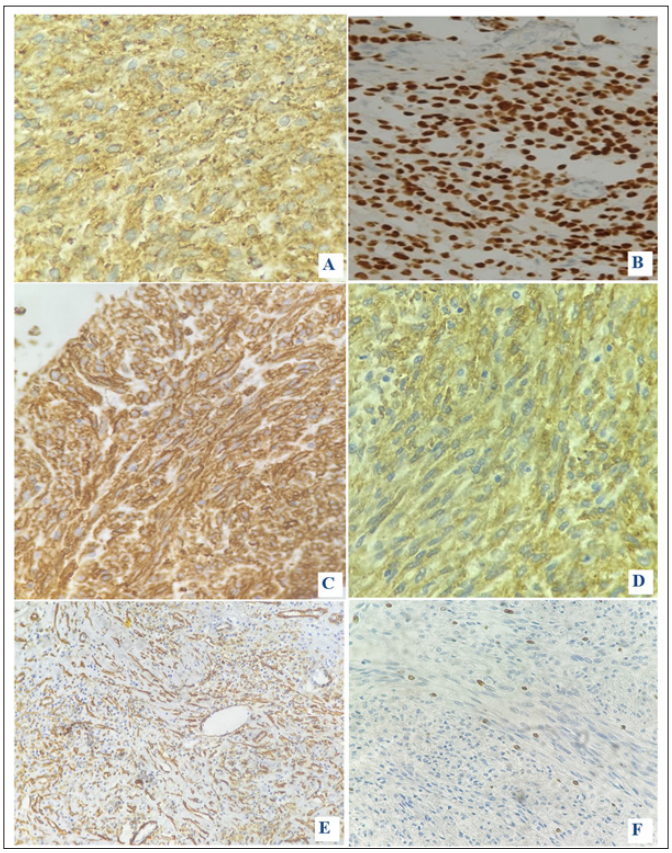

Figure 3: Immunohistochemical Findings: Positive Staining of Tumor Cells with Melan A (A), TFE3 (B), CD34 (C), H-Caldesmone (D) et AML (E), with a low Ki-67 proliferation index estimated at 1% (F)

Discussion

Perivascular epithelioid cell tumors (PEC) are rare mesenchymal neoplasms, most commonly found in the gastrointestinal tract and pelvic organs; however, presentation as a primary mediastinal mass is exceptional, with only 10 cases reported in the literature.

These tumors typically occur in adults between the third and sixth decades of life, with a male-to-female ratio of 3:1. Diagnosis is based on histopathological confirmation along with a characteristic immunohistochemical profile [2].

Unlike typical mesenchymal tumors, our case stands out due to its rarity and immunohistochemical profile: it shows positivity for TFE3 and negativity for HMB-45. Approximately 15% of PEComas exhibit TFE3 gene rearrangements, and these are now considered a distinct entity. TFE3-rearranged PEComas tend to occur in younger patients and often lack expression of smooth muscle markers [3].

Conclusion

This case highlights a rare presentation of a primary mediastinal PEComa, emphasizing the diagnostic challenges associated with such uncommon tumors. Given its rarity and the unique characteristics of TFE3-associated tumors, this case contributes valuable insight to the limited literature and underscores the importance of thorough histological and immunohistochemical evaluation in diagnosing and subclassifying PEComas.

References

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