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Case Report

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A Rare Evolution: Mediastinal Mature Teratoma Undergoing Somatic-Type Malignant Transformation to Rhabdomyosarcoma

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ABSTRACT

Mediastinal mature teratomas are typically benign tumours that predominantly occur in the anterior mediastinum. However, rare cases of malignant transformation within mature teratomas have been documented, most commonly involving somatic-type malignancies. This case report presents a 46-year-old female with productive cough and persistent right lung consolidation despite a course antibiotic and Mycobacterium tuberculosis workout negative. Histopathological examination (HPE) revealed a high-grade epithelioid tumour with myogenic and neuroendocrine differentiation, consistent with alveolar rhabdomyosarcoma with neuroendocrine features. The patient underwent chemotherapy. This case report discusses a unique presentation of a mediastinal mature teratoma with somatic-type malignant transformation into rhabdomyosarcoma in a middle-aged female and highlights the importance of considering rare malignancies, such as rhabdomyosarcoma, in the differential diagnosis of a persistent mediastinal mass, though rare, can significantly alter prognosis and treatment strategies.

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Introduction

Mediastinal teratomas are germ cell tumours typically found in the anterior mediastinum, commonly diagnosed in young adults. These tumours are usually benign and composed of well-differentiated tissues from all three germ layers (ectoderm, mesoderm, and endoderm) [1].

Mature teratomas, in particular, are generally asymptomatic and can be incidentally discovered during imaging studies. However, in rare cases, these tumours undergo malignant transformation, most often involving somatic-type malignancies such as carcinoma, sarcoma, or another aggressive histologist [2].

One of the more unusual and aggressive forms of malignant transformation within a mature teratoma is the development of rhabdomyosarcoma, a rare mesenchymal tumour of striated muscle origin. Rhabdomyosarcoma is a highly malignant soft tissue sarcoma that can present diagnostic and therapeutic challenges. The transformation of a benign teratoma into rhabdomyosarcoma, particularly in the mediastinum, is an exceedingly rare event, with few documented cases in the literature [3].

Case Report

A 46-year-old female with a history of hypertension, prediabetic mellitus, and a haemorrhagic stroke in 2023 presented with a three-month history of a productive cough and progressive dyspnoea on exertion. Her symptoms began in November 2024. Despite being treated for pneumonia at a district hospital, the patient's condition did not improve after completing two courses of antibiotics. Serial chest radiographs revealed persistent consolidation in the right lung. Investigations for Mycobacterium tuberculosis, including sputum TB Gene Xpert and culture, were negative. Given the persistence of symptoms, the patient was referred for specialist evaluation.

On examination, the patient exhibited reduced air entry and dullness to percussion over the right upper and middle zones of the chest with reduced vocal resonance. Her vital signs were stable, and laboratory tests, including full blood count (FBC), liver function tests (LFT), and renal function tests, returned normal results. A chest X-ray revealed a homogenous opacity in the right upper to middle zones, suggesting a mass or consolidation (Figure 1).

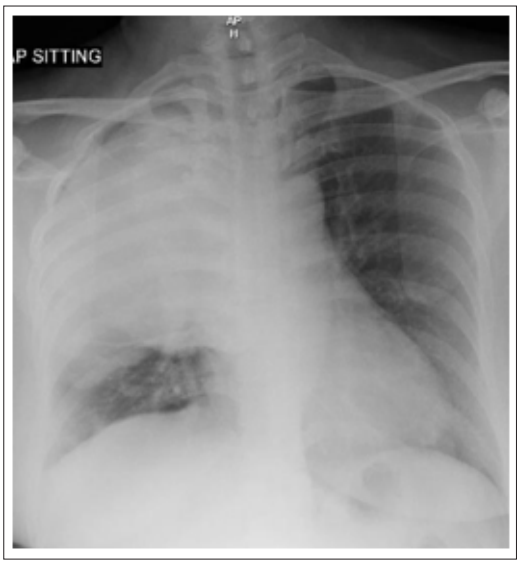

Figure 1: Chest X-Ray: Right Upper to Middle Zone Opacity

A Contrast-Enhanced Computed Tomography (CECT) scan of the thorax was performed, revealing a large mass located in the right upper lobe of the lung (Figure 2 and Figure 3). The mass extended across the fissure into the right middle and lower lobes,

with significant mediastinal extension. Notably, the mass exerted pressure on vital structures, including the superior vena cava, trachea, and bilateral main bronchi. The mass appeared to have a mixed composition, raising the concern for a complex pathology.

An Endoscopic Ultrasound (EUS) biopsy was performed, revealing fragmented cartilaginous tissue with atypical chondrocytes. The differential diagnosis included chondrosarcoma, carcinosarcoma, and malignant teratoma. A resection of the mass was suggested for definitive diagnosis, leading to a repeat CECT scan. This scan demonstrated an enlarging right lung mass with mixed tissue components, compressing the right brachiocephalic vein and involving the posterior right T5 rib. The lesion had extended beyond the rib cage, complicating the surgical approach. The multidisciplinary team (MDT) decided that a Chamberlain procedure, a thoracic surgery technique for accessing the lung and mediastinum, was necessary to obtain a biopsy for further evaluation.

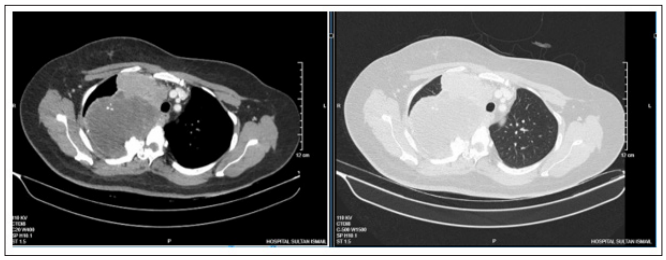

Figure 2 & 3: CECT Thorax: Large Mass Located in the Right Upper Lobe of the Lung Compressing Right Brachiocephalic Vein

The histopathological examination (HPE) of the biopsy revealed a high-grade epithelioid tumour with both myogenic and neuroendocrine differentiation, consistent with a diagnosis of alveolar rhabdomyosarcoma with neuroendocrine differentiation (Figure 4, Figure 5, Figure 6 and Figure 7). Immunohistochemical staining was performed, revealing strong, diffuse positivity for myogenin, vimentin, CD56, and CD99, which are markers characteristic of rhabdomyosarcoma. The tumour cells also showed rare, heterogeneous positivity for synaptophysin, INSM1, desmin, TLE1, and SATB2. However, the tumour was negative for a wide range of other markers, including cytokeratin (MNF116, AE1/AE3), EMA, chromogranin, WT1, and others, which helped exclude other malignancies such as carcinomas or neuroendocrine tumour.

The loss of RB1 nuclear expression was notable, further supporting the diagnosis of rhabdomyosarcoma, as RB1 gene loss is frequently observed in this tumour type.

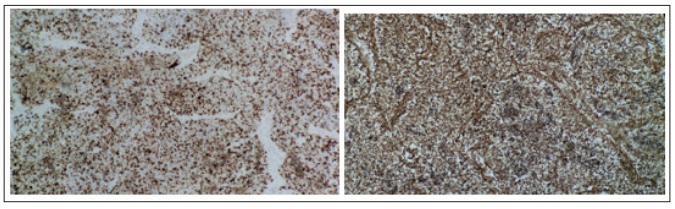

Figure 4: Myogenin Positive **Figure 5:** Vimentin Positive

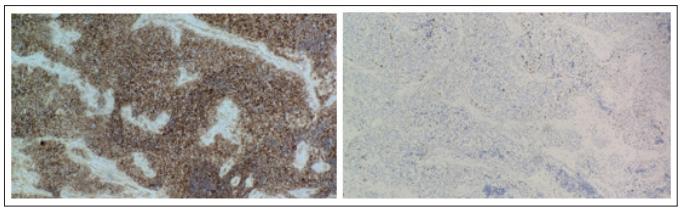

Figure 6: CD99 Positive **Figure 7:** Focal Positive for Desmin

Based on the clinical, radiological, and histopathological findings, the final diagnosis was mediastinal mature teratoma with somatic-type malignant transformation to rhabdomyosarcoma. This rare and aggressive tumour arises from a mature teratoma, where a somatic-type malignancy, in this case, rhabdomyosarcoma, develops. The patient was referred to the oncology team for further management. Chemotherapy was initiated for the patient.

Discussion

Mediastinal teratomas are rare tumors, typically benign, but they can undergo malignant transformation, as seen in this case. Rhabdomyosarcoma, though more common in children, can arise in adults, particularly in the setting of a preexisting teratoma [4]. The malignant transformation of a teratoma to rhabdomyosarcoma is an uncommon but well-documented phenomenon, often characterized by the presence of mixed tissue components, as seen in imaging studies [5]. The most common site for such tumors is the mediastinum, which can complicate diagnosis due to the proximity to vital structures, such as the trachea and major blood vessels [6].

There are only a limited number of cases reported in the literature. This rarity presents several challenges in both the preoperative and postoperative diagnostic processes. The main challenge is that clinical symptoms, such as chest pain, cough, or breathing difficulty, are vague and not specific to this condition. Blood tests are also unhelpful, as no tumor markers reliably indicate SMT. Imaging with CT or MRI often shows a heterogeneous mass, but it is hard to distinguish between benign teratoma tissue and areas of malignant transformation. While PET scans and MRI with diffusion sequences can sometimes detect suspicious areas, these are not always conclusive.

Biopsy can be limited because teratomas are heterogeneous tumors, and small samples may miss malignant areas. In most cases, somatic-type malignant transformation (SMT) is only confirmed after examining the entire tumor following surgical resection. Identifying rhabdomyosarcoma requires specific immunohistochemical stains like desmin, MyoD1, and myogenin, which may not be routinely done unless transformation is suspected like in our case [7].

To improve diagnostic accuracy, a multimodal approach is recommended. Enhanced imaging techniques, including diffusion-weighted MRI and fluorodeoxyglucose positron emission tomography (FDG-PET), may help identify hypermetabolic, potentially malignant areas within a teratoma preoperatively. Targeted image-guided biopsies of suspicious solid areas, rather than random sampling, could increase the likelihood of detecting SMT.

Moreover, integrating preoperative multidisciplinary team discussions involving radiologists, pathologists, thoracic surgeons, and oncologists can facilitate early suspicion and appropriate

diagnostic planning. Comprehensive histopathological evaluation, including immunohistochemistry for all resected mediastinal teratomas with solid areas, is crucial. In the future, techniques like liquid biopsy and greater awareness through case reporting may help detect malignant transformations earlier.

Conclusion

In summary, there are limited case reports on rhabdomyosarcoma in the literature. To the best of our knowledge, this is the first documented case of adult in Malaysia. This case highlights the importance of considering rare malignancies, such as rhabdomyosarcoma, in the differential diagnosis of a persistent mediastinal mass, particularly when a mature teratoma is involved. A heightened clinical awareness, multidisciplinary collaboration, multimodal diagnostic strategy, and proactive pathological assessment are essential for timely diagnosis and management of this rare but aggressive entity for managing rhabdomyosarcoma.

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