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Case Report

From teratoma to sarcoma: unveiling a rare somatic transformation in a mediastinal germ cell tumor

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ABSTRACT

Germ cell tumors (GCTs) are neoplasms primarily arising from gonadal tissues, occasionally occurring in extra gonadal sites. Though generally responsive to chemotherapy, a rare complication is the development of somatic malignancy within the tumor, such as sarcomas or carcinomas, leading to poorer prognosis and complex management. They are driven by genetic/epigenetic changes and dedifferentiation of pluripotent cells. Here we present a rare case of 31-year-old male with malignant peripheral nerve sheath tumor (MPNST) subtype of somatic-type malignancy within a mediastinal GCT.

Keywords: Malignant peripheral nerve sheath tumor, Sarcoma, Teratoma, Somatic-type malignancy, Mediastinal germ cell tumor

INTRODUCTION

Germ cell tumors (GCTs) are a heterogeneous group of neoplasms primarily arising from gonadal tissues, though they can occur in extra gonadal sites. A rare but significant complication is the development of somatic malignancy within the tumor such as sarcomas or carcinomas which poses unique diagnostic and therapeutic challenges.[1] They are associated with poorer prognosis and require tailored management strategies.[2] This case report presents a rare instance of one particular aggressive form of somatic transformation, the development of MPNST within mediastinal GCTs.

CASE REPORT

A 31-year-old male patient presented with cough for 1 month. Initial chest radiograph revealed radio opacity in the left mid zone extending from mediastinum, obliterating the hilar shadow (Figure 1). A contrast enhanced computed tomography (CECT) scan of the thorax revealed a heterogeneously enhancing 9.5×6.4×9.8 cm necrotic mass in the prevsacular anterior mediastinum with irregular borders causing mild compressive atelectasis of adjacent left upper lobe of lung, suggestive of a mediastinal teratoma (Figure 2). A positron emission tomography (PET)-CT scan demonstrated increased fluorodeoxyglucose (FDG) uptake within the mass (standardized uptake value [SUV] max of 5.8), consistent with high metabolic activity (Figure 3). Magnetic resonance imaging (MRI) of the thorax further characterized the mass, showing a large heterogeneously enhancing solid mass in the left anterior mediastinum (Figure 4) with internal cystic areas, necrosis, internal hemorrhage and pericardial infiltration. Initial CT guided biopsy of mediastinal mass revealed poorly differentiated malignant tumour on histology.

Laboratory evaluation at presentation revealed elevated tumor markers consistent with a non-seminomatous germ cell tumor. There was elevated Serum alpha-fetoprotein (AFP) [807IU/mL (reference range: N <7.22 IU/mL)], and beta-human chorionic gonadotropin (β-hCG) [51.43 mIU/mL (: <5 mIU/mL)]. Lactate dehydrogenase (LDH)

was initially within normal limits, 225 U/L (reference range: 120-246 U/L).

The patient was started on neoadjuvant chemotherapy followed by mediastinal tumor excision and specimens were sent for histopathological examination (Figure 5). There were areas of stratified squamous epithelium with keratin material and glandular epithelium. Multiple sections showed tumor composed of spindle cells and focal rhabdoid cells with areas of necrosis. Immunohisto-chemical analysis showed the tumor cells were positive for Desmin, S100 and SOX10. CytoKeratin was positive in glandular elements. The tumor cells were negative for SALL4, glypican 3, CD30, AFP and NKX2.2. A final diagnosis of a mature teratoma with somatic-type malignancy (MPNST with Rhabdomyoblastic differentiation) was made.

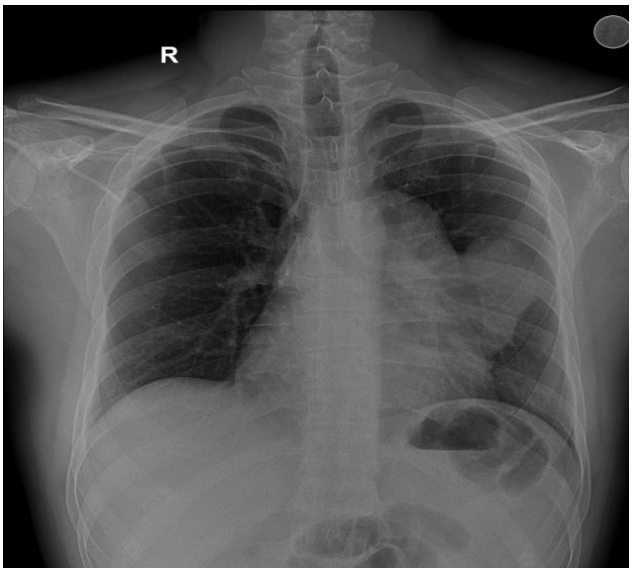

Figure 1: Chest radiograph showing radio opacity in the left mid zone extending from mediastinum, obliterating the left hilar shadow.

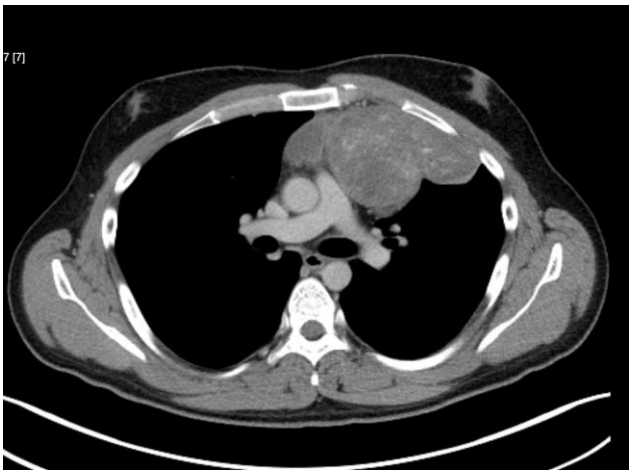

Figure 2: Axial CECT image showing heterogeneously enhancing necrotic mass in the prevsacular anterior mediastinum.

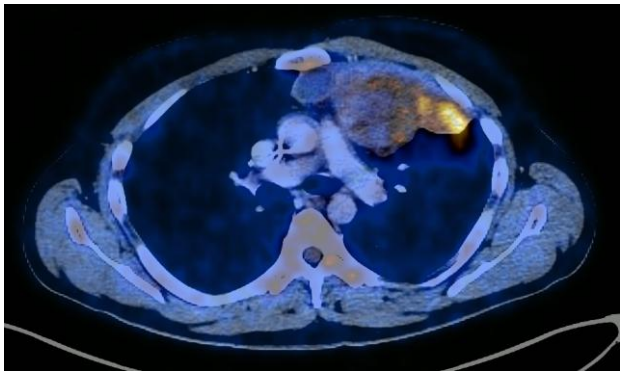

Figure 3: Axial 18F FDG PET CT image of increased metabolic uptake within anterior mediastinal mass.

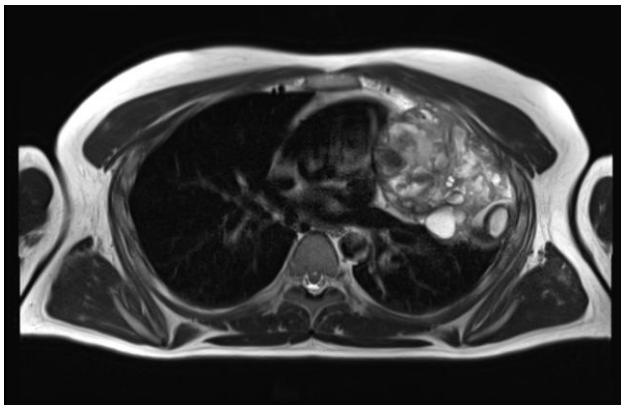

Figure 4: Axial T2 weighted MRI image of large heterogeneous solid mass in left anterior mediastinum with internal cystic areas, necrosis and hemorrhage.

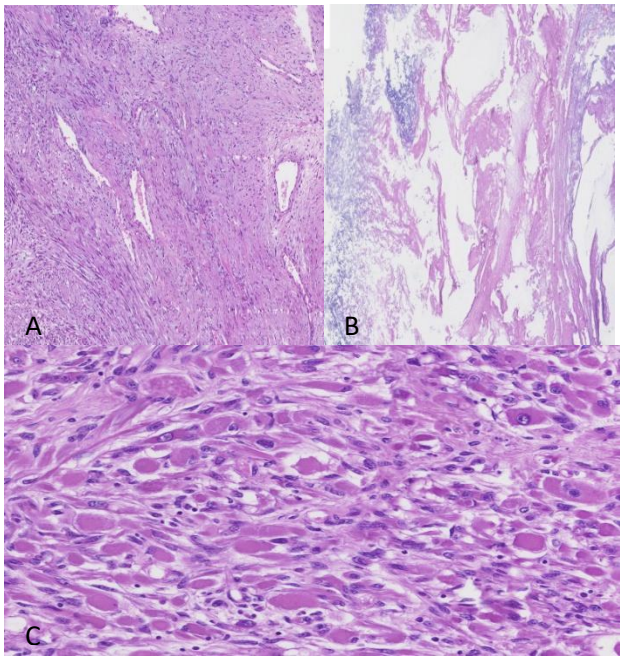

Figure 5 (A-C): Histopathological findings of mediastinal tumor. H and E-stained sections of mediastinal tumor demonstrating sarcoma (A), teratoma (B) and Rhabdoid areas (C).

DISCUSSION

Somatic malignancy arising within GCTs represents a rare and complex clinical entity that complicates the management and prognosis of these neoplasms.[3] Also known as teratomas with malignant transformation (TMT), they are characterized by the transformation of germ cell elements into non-germ cell histologies such as sarcomas, carcinomas, or other malignant lineages.[4] This phenomenon is most observed in non-seminomatous GCTs, particularly in the context of teratomas. They are driven by genetic/epigenetic changes and dedifferentiation or malignant transformation of pluripotent cells into malignant somatic tissues.

Even though others have observed somatic malignancy in mediastinal teratoma, ours is the first case reporting MPNST with rhabdomyoblastic differentiation in a mediastinal teratoma.[5-8] Wang and Kazmi have reported a case of testicular teratoma with metastatic TMT/embryonal rhabdomyosarcoma.[9]

This case illustrates a rare presentation of somatic malignancy within a GCT, characterized by sarcoma in background of teratoma. Diagnostic challenges were evident, as somatic malignancies may mimic the primary GCT on imaging necessitating comprehensive histopathological and immunohistochemical analysis to confirm the diagnosis.

The management of this case involved surgical resection with neoadjuvant and adjuvant chemotherapy, which aligns with current literature advocating for histology-specific treatment protocols. The prognosis for patients with somatic malignancy in GCTs remains guarded, with studies reporting lower overall survival rates compared to pure GCTs.[10]

CONCLUSION

This case report illustrates the rare occurrence of MPNST within a mediastinal GCT, emphasizing its diagnostic challenges. Early recognition and individualized therapy can optimize patient care. This case contributes to the limited literature on somatic malignancy in GCTs and reinforces the need for multidisciplinary collaboration and further studies to improve outcomes in this rare but aggressive subset of GCTs.

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