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Case Report

Heart's secret encounter - extra skeletal Ewings sarcoma with cardiac involvement in adult: a case report

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ABSTRACT

Ewing sarcoma is a rare, aggressive malignant tumour predominantly arising from bone or soft tissue in adolescents and young adults. Primary cardiac involvement is exceedingly uncommon, usually occurring as a result of metastatic spread. Here, we present a case of 22-year-old male with an atypical primary cardiac presentation of Ewings sarcoma, posing significant diagnostic challenges. A detailed evaluation revealed disseminated malignancy, including cardiac and systemic involvement. This report underscores the importance of rare malignancies in atypical cardiac presentations and highlights the value of a multidisciplinary approach in diagnosis and treatment.

Keywords: Ewing's sarcoma, Infiltrative cardiac mass, Cardiac metastasis

INTRODUCTION

Ewing's sarcoma is a rare tumour characterized by chromosomal translocations, most commonly t (11;22) (q24;12) translocation, resulting in EWSR1-FL1 fusion gene mainly affecting bones and soft tissues in adolescents and young adults.[1] The primary tumour usually occurs in bones or soft tissue; however, cardiac involvement is an extremely rare phenomenon, which usually occurs secondary to metastatic spread.[2] This atypical presentation does pose dilemmas in diagnosis and treatment, with symptoms sometimes characterized by chest pain, arrhythmias, and shortness of breath. Although cardiac involvement is rarely encountered, along with the complexity in treatment, the management of such cases must be ensured by a multi-disciplinary approach. Early identification and management would improve the outcome, even though the prognosis remains poor in metastatic cases.[3] This case helps to explore the clinical features, diagnostic strategies, and treatment options for Ewing's sarcoma with cardiac involvement.

CASE REPORT

A 22-year-old male patient, presented with complaints of fever, cough, and breathlessness for 2 weeks. Fever was high grade associated with chills and rigor. Cough was productive with expectoration of whitish sputum. He later developed headache more on frontal area. No history of any chest pain, abdominal pain, vomiting, loose stools, dysuria. No history of any travel outside state, weight loss. No relevant past surgical, or family history.

Physical examination

On general examination, the patient was moderately built and nourished. He was conscious and oriented. Clinical findings included pallor, bilateral pedal pitting edema,

tachycardia, tachypnea, and febrile status (100°F). Jugular venous pressure was raised. Cardiac auscultation revealed muffled heart sounds and a systolic murmur over the mitral area. Breath sounds were reduced bilaterally in the infra scapular region.

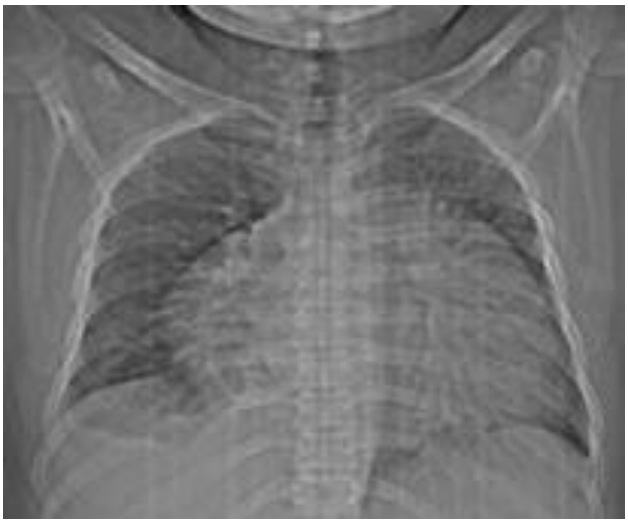

Figure 1: Chest X ray of massive cardiomegaly.

Investigations

Blood investigations revealed neutrophilic leucocytosis with elevated inflammatory markers. A chest X-ray showed massive cardiomegaly with possibility of pericardial effusion (Figure 1). Echocardiography revealed a large pericardial effusion with an infiltrative mass lesion predominantly involving the left atrium (6.4×8.4×8.5 cm), adjacent left ventricle, and pericardium (Figure 2). Pericardiocentesis was performed for diagnostic and therapeutic purposes, draining 600 ml of haemorrhagic fluid, whose cytology and cell block showed no malignant cells.

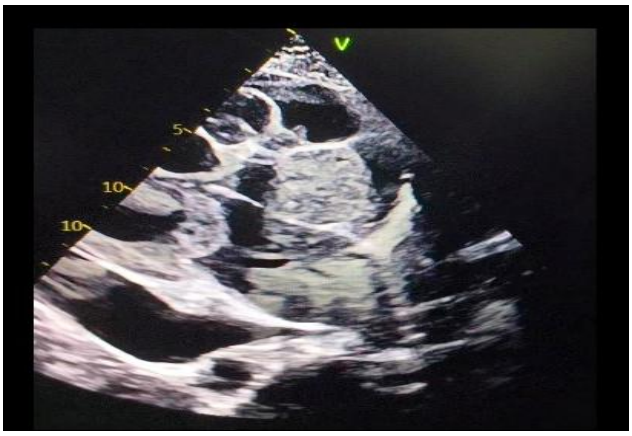

Figure 2: Echocardiography of infiltrative mass lesion.

Contrast-enhanced computed tomography (CECT) of the thorax and abdomen revealed a heterogeneously enhancing infiltrative lesion at the left atrioventricular groove (Figure 3), with infiltration of the left atrium and left ventricle. Additionally, a lobulated homogeneous mass lesion in the mid-jejunum (Figure 4) and a moderately enhancing single peripheral pulmonary nodule in the superior lingular segment of the left upper lobe were noted.

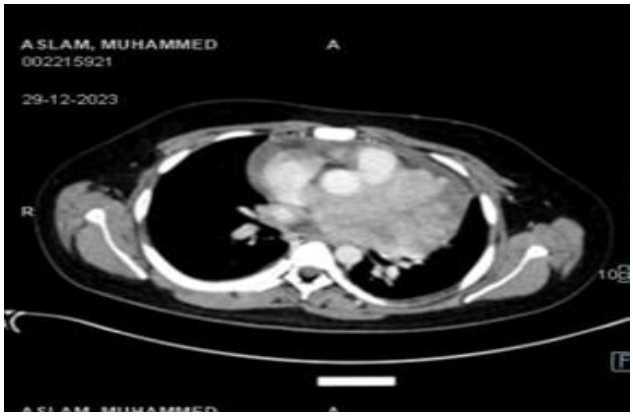

Figure 3: CT thorax of mass lesion.

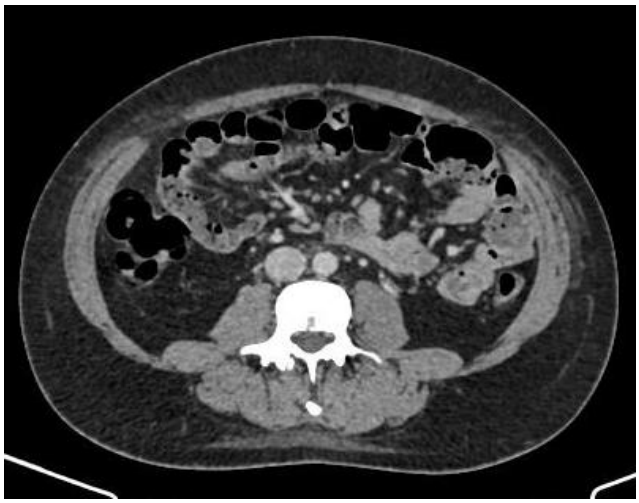

Figure 4: CT abdomen of enhancing homogenous, hyperdense lesion in jejunum.

A whole-body 18-fluorodeoxyglucose positron emission tomography (PET-CT) scan showed a infiltrative mass of size approximately 6.5×8.4 cm in the left atrium, left ventricle with predominant uptake and disseminated metabolically active lesions involving the brain, spleen, small bowel, and multiple intramuscular deposits, particularly in the lower aspect of the right gastrocnemius muscle (Figure 5-7). MRI of the brain revealed a small focus of active diffusion involving the splenium of the corpus callosum, suggestive of acute to subacute infarcts. Ultrasonography guided biopsy of the right gastrocnemius muscle revealed features those of small round blue cell neoplasm. with differential possibilities of rhabdomyosarcoma, lymphoma, and Ewing's sarcoma (Figure 8). Immunohistochemistry confirmed the diagnosis of Ewing's sarcoma with positivity for markers NKX2.2, CD99, and FLI-1(Figure 9).

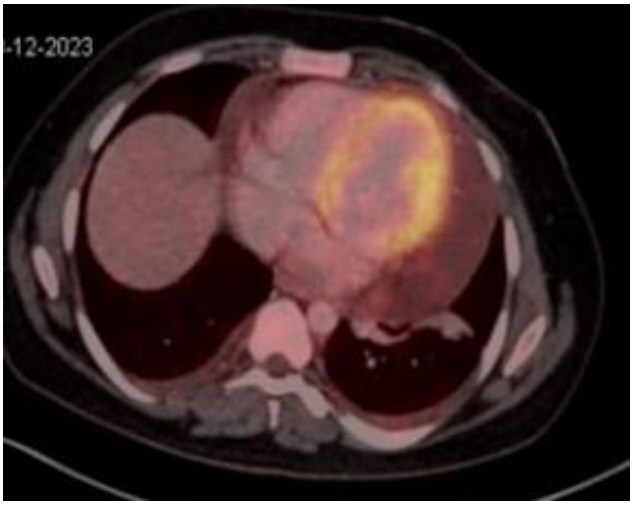

Figure 5: PET CT of increased uptake along left atrium and left ventricle.

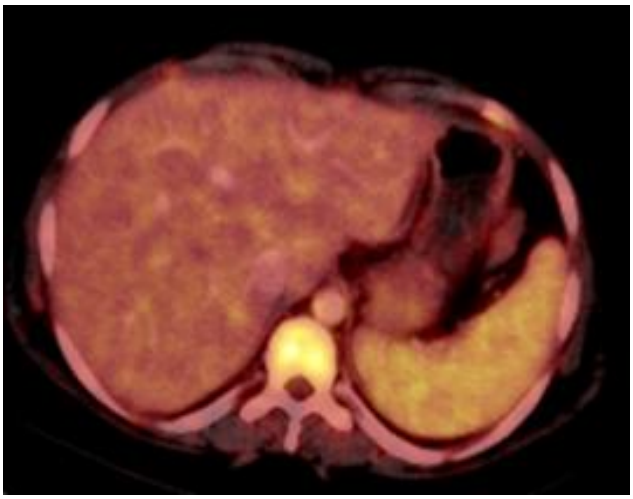

Figure 6: PET CT of abdomen showing increased uptake.

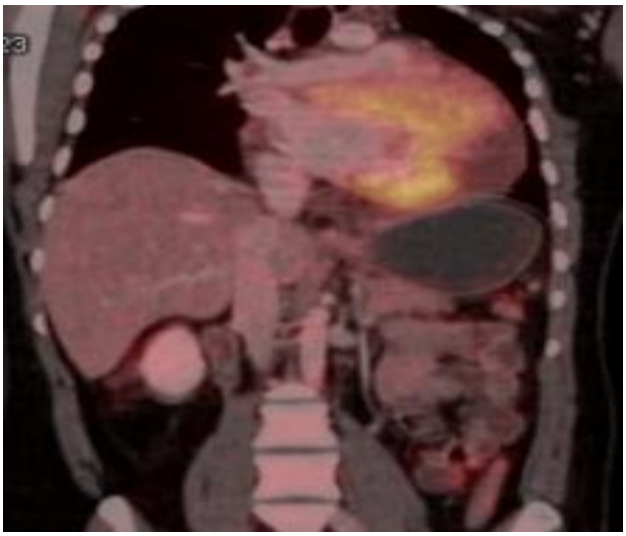

Figure 7: PET CT of increased uptake of left atrium and left ventricle.

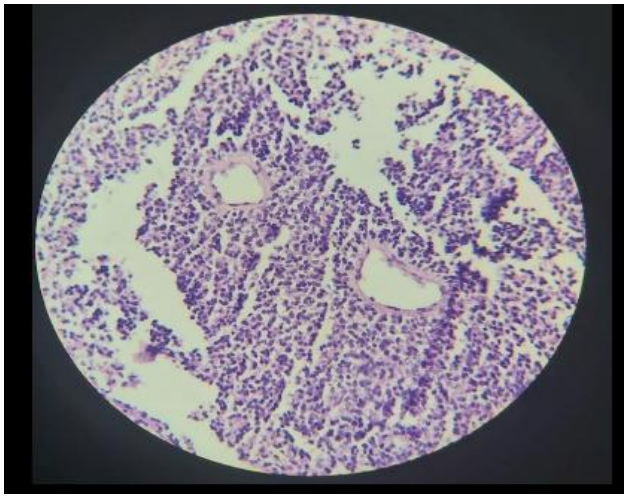

Figure 8: Biopsy of features those of small round blue cell neoplasm.

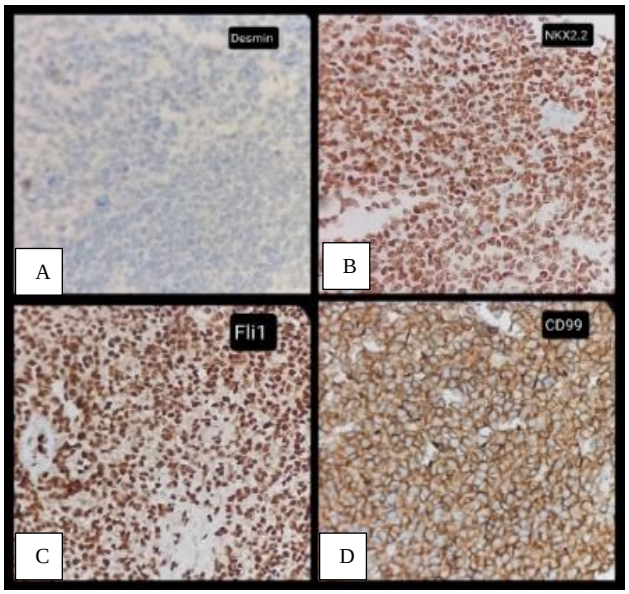

Figure 9 (A-D): Immunohistochemistry panel.

Diagnosis

Stage IV Ewings sarcoma with metastasis.

Treatment

Initial pericardial effusion was managed by pericardiocentesis. Later patient was initiated on palliative cytotoxic chemotherapy-VAC-IE regimen which includes vincristine sulfate, doxorubicin hydrochloride (Adriamycin), cyclophosphamide, ifosfamide and etoposide. He completed 1 year of treatment and is symptomatically doing well.

Follow up

A repeat PET CT scan taken after 1 year were suggestive of good partial response to therapy with significant

reduction in metabolic activity of the lesion (Figure 10-12).

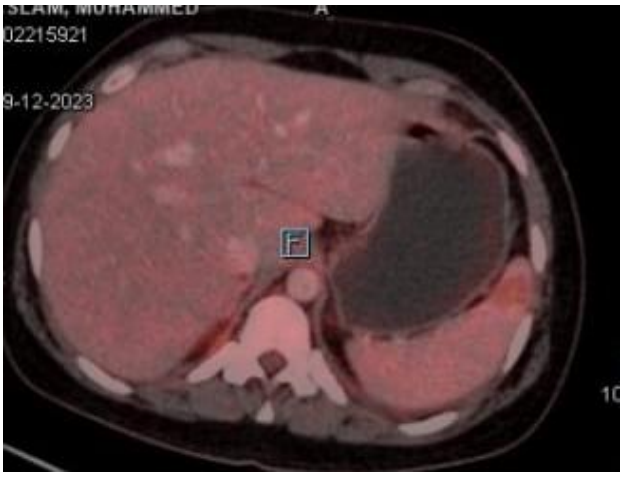

Figure 10: Repeat PET CT of abdomen.

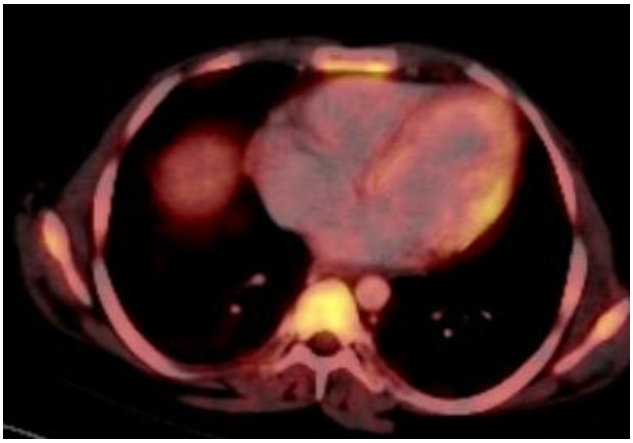

Figure 11: Repeat PET CT of thorax showing reduced uptake.

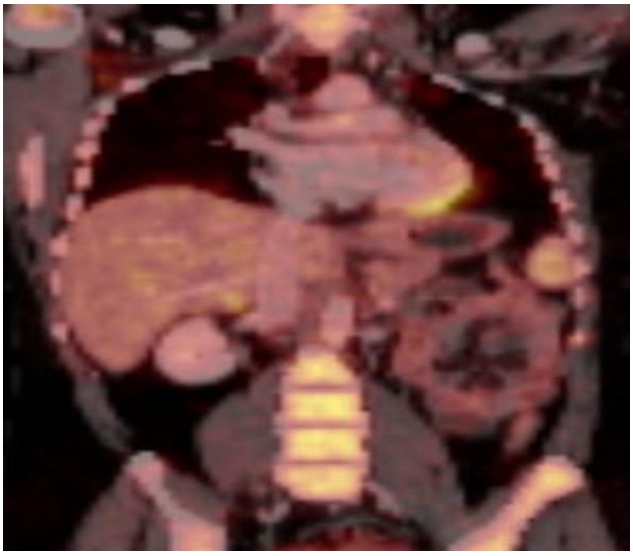

Figure 12: Repeat PET CT showing reduced uptake of left atrium and left ventricle.

DISCUSSION

Ewing sarcoma is a rare, aggressive malignant tumour most commonly arising from bone or soft tissue in adolescents and young adults.[4-6] There have been a few associations described with chromosomal translocations, most especially the t (11;22) (q24; q12) translocation resulting in the EWSR1-FLI1 fusion gene. Primary involvement of the heart is extremely rare; the majority are due to secondary spread.[7,8] This is a rare presentation of Ewing sarcoma involving the heart that may initially present with fever, cough, and breathlessness which mimics infectious conditions and cause diagnostic challenges. X ray revealed pericardial effusion and echocardiographic findings of an infiltrative mass lesion in the left atrium that raised suspicion for malignancy. Systemic involvement due to PET scans showing metabolically active lesions of the bowel, as well as intramuscular deposits, implicated a disseminated malignancy. The rarity of primary cardiac neoplasms further necessitated thorough diagnostic studies to establish nature and origin. The left atrial mass involved might be the cause of pericardial effusion by occlusion of venous drainage, direct invasion of the pericardium, or necrosis due to the tumour. Such involvement can lead to dyspnoea, chest pain, or even hemodynamic compromise. The management by pericardiocentesis, therefore becomes critical to prevent complications such as cardiac tamponade. Management is challenging and includes multi-agent chemotherapy, with options for surgical resection or radiation therapy depending on the stage of prognosis. The prognosis is poor due to advanced disease at presentation, although it puts into perspective the importance of considering rare malignancies in atypical cardiac presentations and adopting a multidisciplinary approach for diagnosis and treatment.

CONCLUSION

This case highlights an unusual presentation of Ewings sarcoma with primary cardiac involvement. The diagnostic process required careful evaluation, and multidisciplinary approach was instrumental in achieving a definitive diagnosis and effective management. Future research is needed on understanding the pathophysiology and optimizing treatment strategies for this rare entity.

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