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Case Report: Primary Cardiac Sarcoma in a Young Adult

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Abstract – Primary cardiac sarcoma is an exceptionally rare and aggressive malignancy, representing a small fraction of primary cardiac tumors. This case report presents a detailed account of a 28-year-old female who was diagnosed with primary cardiac sarcoma, highlighting the complexities of its presentation, diagnostic approach, and treatment. The patient, previously in good health, experienced a six-month history of progressive dyspnea and intermittent chest pain. Initial evaluations, including an electrocardiogram and chest X-ray, did not reveal significant abnormalities. However, an echocardiogram identified a mass on the left ventricular free wall, leading to further investigation via cardiac MRI. The MRI findings were consistent with a primary cardiac tumor, specifically angiosarcoma, characterized by a heterogeneous mass with areas of necrosis and infiltration. The diagnosis was confirmed by endomyocardial biopsy.

The patient underwent surgical intervention, which involved partial resection of the left ventricle to excise the tumor. Post-operatively, she received chemotherapy tailored to the tumor's histological profile, incorporating doxorubicin and ifosfamide. Regular follow-up imaging was scheduled to monitor for recurrence and metastasis. This case underscores the diagnostic challenges associated with primary cardiac sarcomas, given their rarity and the non-specific nature of symptoms. It also emphasizes the importance of a multidisciplinary approach, combining advanced imaging, surgical resection, and adjuvant chemotherapy, to manage this aggressive malignancy effectively. Early detection and comprehensive treatment are crucial for improving prognosis in patients with primary cardiac sarcoma.

Keywords – Primary cardiac sarcoma, angiosarcoma, left ventricular mass, cardiac imaging, multidisciplinary treatment.

Case Presentation:

Patient Information:

Name: Sarah Williams

Age: 28

Gender: Female

Chief Complaint:

Progressive shortness of breath and intermittent chest pain.

History of Present Illness:

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Sarah Williams, a previously healthy 28-year-old female, presented with a 6-month history of worsening dyspnea and sharp, localized chest pain.

The pain was aggravated by physical activity and partially relieved by rest. She denied any episodes of syncope or palpitations.

There was no history of fever, cough, or recent travel.

Past Medical History:

No significant medical history.

No history of hypertension, diabetes, or hyperlipidemia.

Family History:

No known cardiovascular diseases or genetic disorders.

Social History:

Non-smoker.

Occasional alcohol consumption.

No history of drug abuse.

1. Physical Examination:

Vital Signs: BP 120/78 mmHg, HR 78 bpm, RR 16 breaths/min.

General: Patient appears well-nourished and in no acute distress.

Cardiovascular: Normal heart sounds, no murmurs or gallops.

Respiratory: Clear lung fields bilaterally.

Other: No edema or jugular venous distension.

Diagnostic Workup:

2.1 Electrocardiogram (ECG):

Normal sinus rhythm; no ischemic changes.

Chest X-ray:

No acute findings; heart size within normal limits.

2.2 Echocardiogram:

Identified a mass on the left ventricular free wall.

Mild reduction in left ventricular systolic function.

2.3 Cardiac MRI:

Revealed a large mass with characteristics suggestive of a cardiac tumor, specifically angiosarcoma.

Mass appeared heterogeneous with areas of necrosis and infiltration.

3. Laboratory Tests:

Normal cardiac biomarkers (troponin, BNP).

Histopathological Examination:

Endomyocardial biopsy confirmed the diagnosis of primary cardiac sarcoma (angiosarcoma).

4.Diagnosis:

Primary Cardiac Sarcoma (Angiosarcoma)

Management:

Surgical Intervention: Referral to a cardiothoracic surgeon for resection of the tumor. Surgical team performed a partial resection of the left ventricle to remove the mass.

Oncology Consultation: Initiated chemotherapy regimen based on histological findings, including doxorubicin and ifosfamide.

Follow-Up: Regular imaging scheduled to monitor for recurrence or metastasis.

Literature Review:

Introduction:

Primary cardiac sarcomas are exceedingly rare and aggressive malignancies, constituting less than 1% of all primary cardiac tumors. Their rarity and the lack of specific symptoms often lead to delayed diagnosis and poor prognosis. This literature review aims to

provide an overview of the current understanding of primary cardiac sarcomas, including their epidemiology, clinical presentation, diagnostic challenges, and management strategies.

Epidemiology and Classification:

Primary cardiac sarcomas are classified into several histological subtypes, with angiosarcoma being the most common. Other types include leiomyosarcoma, rhabdomyosarcoma, and pleomorphic sarcoma. Angiosarcoma, originating from the vascular endothelium, is particularly aggressive and accounts for a significant proportion of primary cardiac sarcomas. The incidence is low, with a higher prevalence in middle-aged adults, though cases have been documented in younger patients.

Clinical Presentation:

The clinical presentation of primary cardiac sarcomas is often non-specific, including symptoms such as dyspnea, chest pain, and occasionally, symptoms of heart failure. Due to the absence of specific signs, diagnosis is frequently delayed. Symptoms may be attributed to other more common cardiovascular conditions, leading to initial misdiagnoses.

Diagnostic Challenges:

Diagnosis relies heavily on advanced imaging techniques. Echocardiography is often the first modality used, revealing masses or abnormal cardiac structures. However, cardiac MRI provides superior tissue characterization and is crucial for distinguishing between benign and malignant tumors. Definitive diagnosis is typically confirmed through histological examination following biopsy or surgical resection.

Management and Prognosis:

Management of primary cardiac sarcomas involves a multi-disciplinary approach. Surgical resection remains the cornerstone of treatment, aiming to achieve complete tumor removal. Adjuvant chemotherapy and radiotherapy are used to address residual disease and reduce recurrence risk. The treatment regimen often includes drugs such as doxorubicin and ifosfamide. Despite aggressive treatment, the prognosis remains poor due to the high propensity for local recurrence and distant metastasis. Survival rates vary, but 5-year survival is generally low, highlighting the need for ongoing research into more effective therapies and early detection methods.

Recent Advances:

Recent advances in imaging technology and molecular diagnostics have improved the ability to characterize these tumors and tailor treatment strategies. Additionally, research into targeted therapies and immunotherapies offers hope for better outcomes. However, large-scale studies and clinical trials are needed to establish more effective treatment protocols and improve survival rates.

Conclusion:

Primary cardiac sarcomas pose significant diagnostic and therapeutic challenges due to their rarity and aggressive nature. Early and accurate diagnosis, combined with a comprehensive treatment approach, is essential for improving patient outcomes. Continued research is vital to advance the understanding and management of these rare malignancies.

Introduction:

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Primary cardiac sarcomas are rare, accounting for less than 1% of all primary cardiac tumors. They often present with symptoms similar to other cardiovascular diseases, leading to diagnostic delays. This report discusses a case of primary cardiac sarcoma that was initially misdiagnosed and later identified through advanced imaging techniques.

Objective: To highlight the complexities of diagnosing and managing primary cardiac sarcoma in a young patient, emphasizing the need for early detection and multidisciplinary treatment approaches.

Literature Review: Primary Cardiac Sarcoma

Introduction

Primary cardiac sarcomas are extremely rare tumors, accounting for less than 1% of all primary cardiac malignancies. Their aggressive behavior and vague clinical symptoms often lead to delayed diagnosis and unfavorable outcomes. This review explores the current understanding of primary cardiac sarcomas, focusing on their epidemiology, clinical presentation, diagnostic challenges, and management approaches.

Epidemiology and Classification

Among the various subtypes of primary cardiac sarcomas, angiosarcoma is the most frequently encountered. Other subtypes include leiomyosarcoma, rhabdomyosarcoma, and pleomorphic sarcoma. Angiosarcoma, which originates from the vascular endothelium, is particularly aggressive and tends to occur more commonly in middle-aged individuals, though cases in younger populations, such as the 28-year-old patient Sarah Williams, have been reported.

Clinical Presentation

Patients with primary cardiac sarcomas often present with non-specific symptoms such as shortness of breath, chest pain, and occasionally signs of heart failure. The nonspecific nature of these symptoms frequently results in misdiagnosis, as they may be attributed to more common cardiovascular issues, delaying appropriate treatment.

Diagnostic Challenges

The diagnosis of primary cardiac sarcomas relies heavily on advanced imaging techniques. Echocardiography is typically the first step in evaluation, revealing potential masses or structural abnormalities. However, cardiac MRI is crucial for detailed tissue characterization, helping to differentiate between benign and malignant tumors. Definitive diagnosis usually requires histological confirmation through biopsy or surgical intervention, as evidenced in Sarah's case.

Management and Prognosis

Managing primary cardiac sarcomas involves a coordinated, multi-disciplinary approach. Surgical resection remains the primary treatment goal, focusing on complete tumor removal. Adjuvant therapies, including chemotherapy with agents like doxorubicin and ifosfamide, are used to address any residual disease and reduce recurrence risk. Despite these aggressive treatment strategies, the prognosis remains poor, with 5-year survival rates being low due to a high likelihood of local recurrence and metastasis.

Recent Advances

Recent advancements in imaging technologies and molecular diagnostics have improved the characterization of these tumors and may help in customizing treatment strategies. Research into targeted therapies and immunotherapies also offers promise for enhancing patient outcomes. Nonetheless, more extensive clinical studies are necessary to establish effective treatment protocols and improve survival rates.

Conclusion

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Primary cardiac sarcomas pose considerable diagnostic and therapeutic challenges due to their rarity and aggressive nature. Timely and accurate diagnosis, coupled with comprehensive treatment strategies, is essential for improving outcomes in affected patients. Continued research is critical to furthering our understanding of these rare malignancies and enhancing management strategies.

Description-

High-grade angiosarcomas in our patient Sarah Williams, highlights primary cardiac sarcomas as some of the rarest and most aggressive cardiac tumors. These malignancies, the subtypes of PCS, arise typically from the right atrium but can develop in other areas of the heart, such as the left ventricular free wall, as seen in Sarah's case. There is a slight male predominance with a male-to-female ratio of approximately 1.5:1.

Sarah Williams, our 28-year-old young patient, presented with a six-month history of worsening shortness of breath and the occasional chest pain. These symptoms are characteristic yet non-specific of PCS. Due to these vague symptoms, diagnosis is often delayed as they mostly mimic conventional cardiovascular conditions such as CAD or heart failure. In Sarah's case, the initial diagnostic tests- ECG and CXR, were unremarkable, typical in early PCS cases where standard imaging may not reveal abnormalities. Common lab abnormalities in cardiac sarcoma include anemia, hypergammaglobulinemia and elevated ESR.

Her persistent symptoms led to further investigations, leading to an echocardiogram that detected a hypoechoic mass attached to the left ventricular free wall- a finding more indicative of a malignant process. Cardiac MRI provided detailed imaging, showing a large, irregularly enhancing mass with areas of necrosis, which strongly suggested high-grade angiosarcoma.

A high-grade angiosarcoma like this one carries a poor prognosis due to several factors. These tumors are highly aggressive, prone to early metastasis, and often involve the critical cardiac structures of the heart making complete surgical resection challenging by making it difficult to achieve clear margins due to its infiltrative nature.

Histopathological analysis of an excised tissue of Sarah's heart confirmed the diagnosis of high-grade angiosarcoma, characterized by its rapid growth, high potential for local invasion, and distant metastasis.

Due to the disease's rarity, risk factors of the condition are not fully understood though it can include genetic predisposition and possibly previous radiation exposure. However, Sarah had no known exposure as such in her history [5]. The aggressive nature of Sarah's tumor necessitated a multimodal treatment approach. Doxorubicin and ifosfamide were included in her post-surgical treatment. These drugs are commonly used for sarcoma because they effectively target rapidly dividing cells.

Even with these treatments, the outlook for patients like Sarah is still uncertain, with average survival rates of 6 to 12 months after diagnosis. Survival rates at the five-year mark are concerning, often falling below 10%, especially when surgery cannot remove all cancerous tissue or if the cancer has spread at the time of diagnosis.

The progression of PCS in patients such as Sarah Williams emphasises the critical necessity for enhanced diagnostic methods and more successful treatment approaches. Detecting illnesses at early is crucial, yet difficult due to the vague symptoms and the constraints of initial diagnostic exams. Sophisticated imaging techniques like cardiac MRI are essential for the early detection and understanding of these tumors, offering in-depth data that inform surgical and treatment plans [1] .Furthermore, studying PCS's molecular and genetic basis might result in more specific therapies, potentially enhancing results for individuals affected by this severe illness.

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In Sarah's situation, similar to many individuals, her high-grade angiosarcoma most likely came about due to random genetic mutations with no clear external trigger or predisposing factor.

Discussion-

Primary cardiac sarcomas (PCS) are exceedingly rare, representing only about 25%of all cardiac tumors, with angiosarcoma being the most common malignant variant. Epidemiological data on primary cardiac sarcoma (PCS) in young adults remain limited, but some trends are emerging from recent research. Cardiac sarcomas primarily affect adults, with a higher prevalence in males, especially those between 30-50 years old. The clinical presentation of PCS is often unclear, often including symptoms like exertional dyspnea, chest pain, and arrhythmias that mimic other cardiac conditions. Cardiac sarcomas may present through four mechanisms: obstructing blood flow, local invasions that lead to arrhythmias or pericardial effusion with tamponade (seen in 29% cases), embolic events due to tumor fragments or thrombus. Due to its non-specific symptoms, the condition is often to be diagnosed late by which time the tumors are frequently at an advanced stage with widespread metastasis, most commonly affecting the lungs bones, or brain which dramatically diminishes the chances of successful treatment and long-term survival. Angiosarcoma can mimic conditions like percarditis, coronary artery disease or pulmonary embolism, and may involve the interventricular septum. Though the surgery is main the treatment, these presentations afect prognosis and highlight the need for prompt diagnosis and expedited surgery. Sarcoma >4cm had higher distant metastasis risk, with lung being the most common site, while brain metastases showed the worst overall survival and cancer-specific survival.

PCS are associated with a high incidence of brain metastases and shorter survival compared to other sarcomas. Brain imaging should be performed at diagnosis, and aggressive multimodal treatment, even in the metastatic setting, can improve outcomes.

Surgical resection remains the primary treatment approach in the management of primary cardiac sarcoma, although the overall prognosis remains poor. Emerging research suggests that complete resection especially in cases where the malignancy is localized, may improve survival outcomes.

Achieving full removal, however, is challenging due to the tumor’s proximity to critical cardiac structures. Additionally, PCS is associated with a high recurrence rate and frequent metastasis to other organs. chemotherapy, particularly with anthracycline-based agents like doxorubicin, is a standard treatment component, though it offers limited improvements in long-term survival.

Recent research is increasingly focused on advanced surgical techniques like auto-transplantation, where the heart is temporarily removed for tumor resection outside the body, followed by reconstruction and reimplantation.Poor survival rates in elderly patients and those with metastatic disease are persistently emphasized in recent epidemiological data. The median overall survival rate for young adults is typically 15 months. Tumor grade, size, and location influence prognosis, with metastasis to distant organs being common in advanced cases.

As studies advance, there is a growing consensus in the medical community on the importance of early detection and more effective treatment strategies to improve outcomes for this challenging condition.

Figures and Tables

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Figure 1

Analysis of incidence trend of primary cardiac angiosarcoma (incidence per 100,000) from 1976 to 2015

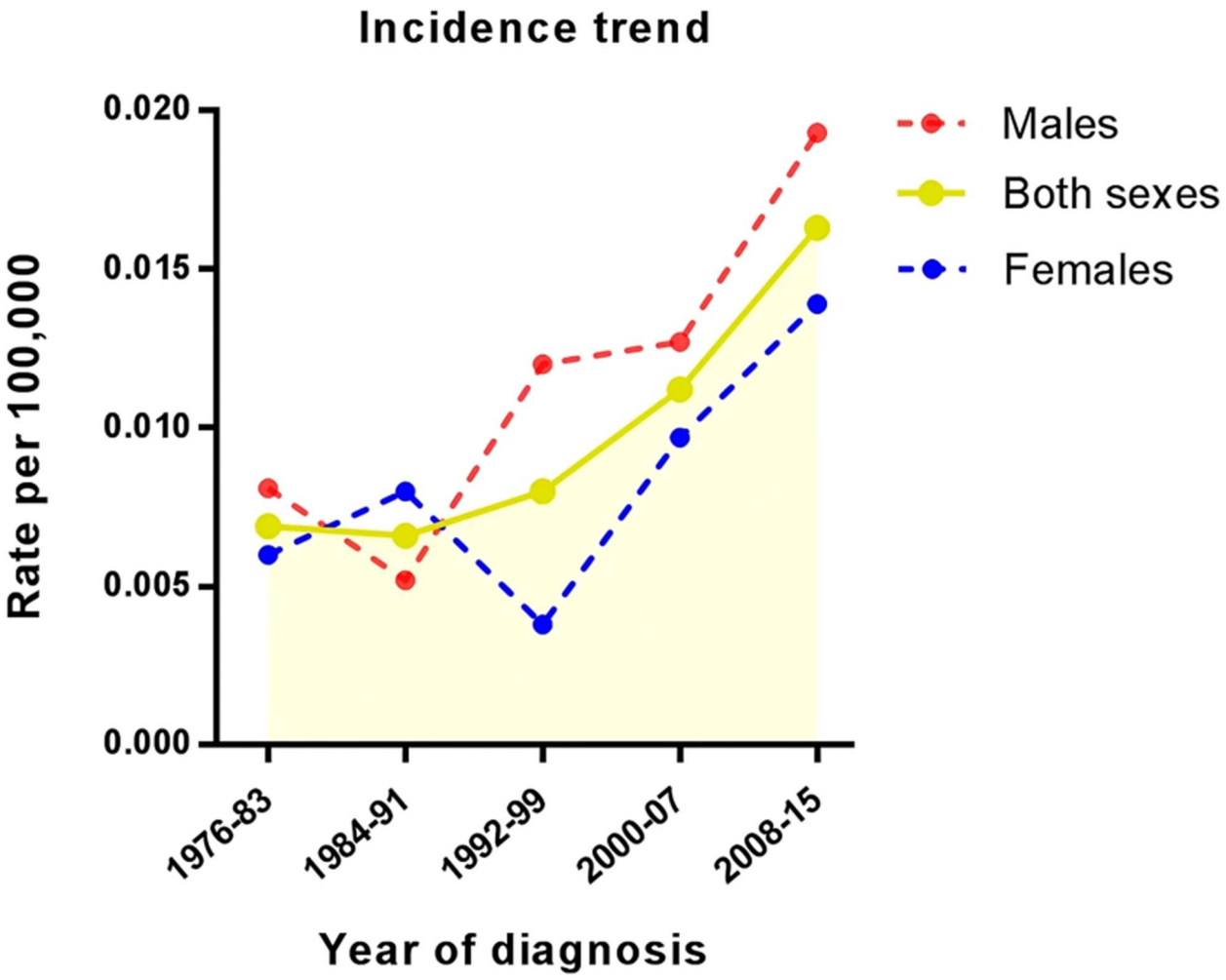

Figure 2

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Primary cardiac tumor histology according to location

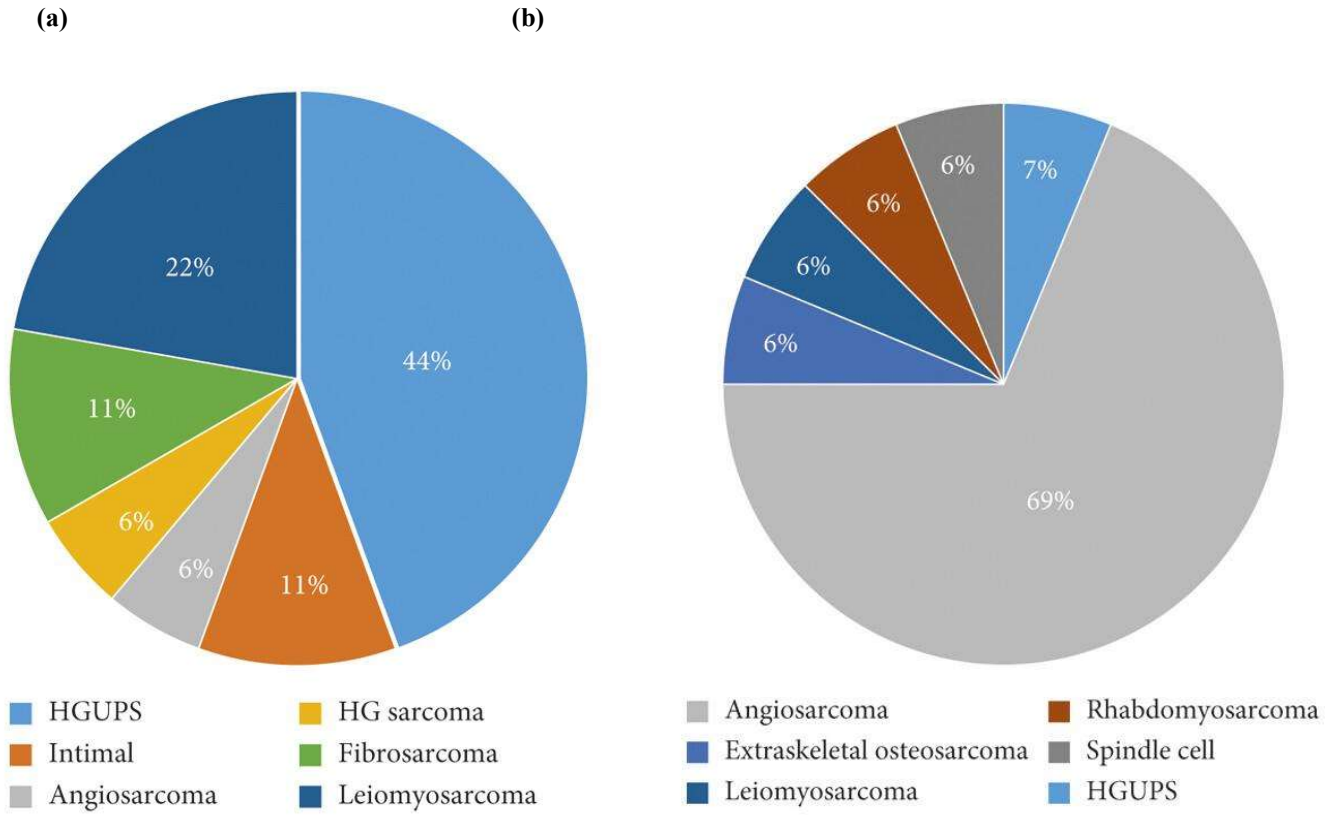

Figure 2 : Cardiac magnetic resonance image shows a right ventricular angiosarcoma (arrow)

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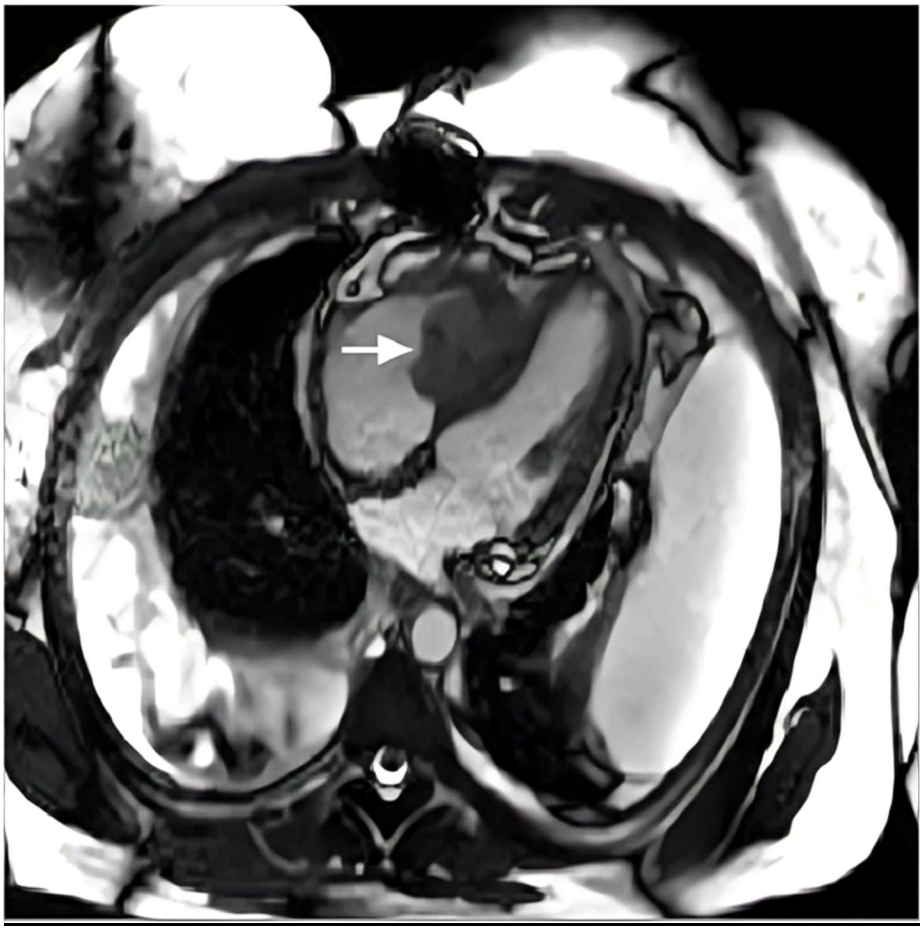

Summary

Primary cardiac sarcomas being rare and aggressive make up less than 1% of all cardiac tumors, and due to this rarity along with non-specific symptoms lead to misdiagnosis, delaying treatment, and worsening prognosis.

This case report highlights the diagnostic and therapeutic challenges posed by primary cardiac sarcoma in a young 28-year-old Sarah Williams, who experienced shortness of breath and intermittent chest pain for 6 months. Advanced imaging, including echocardiography and cardiac MRI, revealed a large mass on the left ventricular free wall and surgery confirmed the diagnosis of high-grade angiosarcoma.

The accompanying literature review discusses the epidemiology, clinical presentation, diagnostic challenges, and treatment strategies for primary sarcomas. These tumors are mainly found in middle-aged adults but can appear in young patients, as seen in this case. Their clinical presentation often resembles other cardiovascular conditions, resulting in possible misdiagnosis. Cardiac MRI is vital for differentiating these malignancies from other cardiac tumors.

Surgical resection is the primary treatment and adjuvant chemotherapy and radiotherapy are often used to tackle residual disease and lower recurrence risks.

The prognosis for primary cardiac sarcoma patients remains poor, with low five-year survival rates. This case highlights the need to consider rare malignancies like primary cardiac sarcoma in diagnosing persistent cardiovascular symptoms, especially in younger patients, and early diagnosis that might help in better survival rates. Tumors in the left heart have better outcomes, likely due to earlier detection, though histologic grading has not shown survival correlation for cardiac sarcomas.

Conclusion

The case of Sarah Williams underscores the complexities associated with rare and aggressive tumors like primary cardiac sarcoma, from the initial clinical suspicion to the confirmation through advanced imaging and histopathological analysis.

Primary cardiac sarcoma in young adults, poses challenges because of the nature of their malignancy and the non-specific symptoms that delay diagnosis, necessitates heightened awareness among clinicians to avoid diagnostic delays that could adversely affect patient outcomes. It emphasizes the necessity of a multidisciplinary approach to optimize treatment. Despite the aggressive management, the prognosis for primary cardiac sarcoma remains limited, as evidenced by the high rates of local recurrence and distant metastasis thus reinforcing the need for early detection strategies, given the poor outcomes associated with delayed diagnosis.

Furthermore, this case underscores the importance of ongoing research into more effective treatment modalities, including targeted therapies and immunotherapy, to improve long-term survival rates. Future studies must focus on refining surgical methods of treatment and explore novel chemotherapeutic and biologic agents tailored to tumor's genetic profiles. Establishing a structured, long-term follow-up regimen is essential for monitoring recurrence and optimizing patient care pathways. Comprehensive, multidisciplinary approaches and advancements in the therapeutic strategies will be pivotal in improving outcomes for patients with primary cardiac sarcoma.

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