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INTERNATIONAL JOURNAL OF ADVANCED RESEARCH (IJAR)

Article DOI: 10.21474/IJAR01/22415
DOI URL: http://dx.doi.org/10.21474/IJAR01/22415

RESEARCH ARTICLE

DILATED CARDIOMYOPATHY AS THE INITIAL PRESENTATION OF CARDIAC SARCOIDOSIS IN A YOUNG PATIENT

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Manuscript Info

Manuscript History
Received: 10 October 2025
Final Accepted: 12 November 2025
Published: December 2025

Key words:-
cardiac sarcoidosis, dilated cardiomyopathy, cardiac MRI, immunosuppressive therapy, case report

Abstract

Sarcoidosis is a systemic granulomatous disease that most commonly affects the lungs and lymphatic system. Cardiac involvement is uncom mon but potentially life-threatening and may present with heart failure, arrhythmias or conduction disorders. Early diagnosis remains challengi ng due to the lack of specific clinical and biological markers.We report the case of a 21-year-old woman with no significant medical history who presented with dyspnea, palpitations and lower limb edema. Clinical examination revealed signs of right sided heart failure. Electro cardiography showed regular tachycardia and left atrial enlargement. Transthoracic echocardiography demonstrated a severe non-ischemic dilated cardiomyopathy with biventricular dilation and a left ventricular ejection fraction of 20%. Thoracoabdominal computed tomography revealed pulmonary nodules and micronodules suggestive of sarcoidosi s. Cardiac magnetic resonance imaging showed transmural and subepic ardial late gadolinium enhancement at the right and left ventricular insertion points, supporting the diagnosis of cardiac sarcoidosis. The patient was treated with high-dose corticosteroids and monthly cyclophosphamide pulses, followed by azathioprine maintenance therapy, resulting in significant improvement of left ventricular function.This case highlights the importance of cardiac imaging in the diagnosis and management of cardiac sarcoidosis.

Introduction:-

Sarcoidosis is a systemic granulomatous disease of unknown etiology, characterized by the formation of non-caseatingepithelioid granulomas[1]. It predominantly affects the mediastinal and pulmonary systems in the majority of patients. Other extra-pulmonary manifestations may occur, including ocular, cutaneous, lymph node and articular involvement.Cardiac involvement in sarcoidosis is uncommon but potentially life-threatening, occurring in approximately 5–20% of patients, often in a subclinical form[1,2]. It may present with conduction abnormalities, arrhythmias, heart failure, or, more rarely, with dilated cardiomyopathy, leading to significant morbidity and mortality[3,4].

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We report the case of a patient with cardiac sarcoidosis revealed by dilated cardiomyopathy. Through this observation, we emphasize the crucial role of cardiac imaging in suggesting the diagnosis and assessing the extent of cardiac involvement in sarcoidosis.

Case Presentation:-

A 21-year-old woman with no significant past medical history presented to the cardiology department with progressive dyspnea, palpitations and bilateral lower limb edema evolving over several weeks. She denied chest pain, syncope, fever, anorexia, excessive night sweating, or skin rashes. There was no family history of heart disease or sarcoidosis. Physical examination revealed signs of right-sided heart failure, including jugular venous distension and peripheral edema. Blood pressure was stable, and there were no murmurs or pulmonary crackles. Electrocardiography showed regular tachycardia and left atrial enlargement, without conduction block or ventricular arrhythmia. Transthoracic echocardiography revealed a severe non-ischemic dilated cardiomyopathy, with biatrial dilation and a dilated, hypokinetic left ventricle. The left ventricular ejection fraction was estimated at 20%. Thoracoabdominal computed tomography demonstrated multiple pulmonary nodules and micronodules, raising suspicion for sarcoidosis. Laboratory investigations showed normal lymphocyte count, normal inflammatory markers, normal serum and urinary calcium levels, and normal angiotensin-converting enzyme levels. Serologic testing for HIV and syphilis was negative, and autoimmune screening was unremarkable. Minor salivary gland biopsy revealed granulomatous lesions.

Given the unexplained cardiomyopathy and radiological pulmonary findings, cardiac magnetic resonance imaging was performed. It demonstrated transmural and subepicardial late gadolinium enhancement at the right and left ventricular insertion zones (figure 1), a pattern highly suggestive of cardiac sarcoidosis.Based on the clinical presentation, imaging and histological findings and exclusion of alternative diagnoses, a diagnosis of cardiac sarcoidosis was retained. Due to the severity of cardiac involvement, the patient received intravenous corticosteroid pulses for three days, followed by high-dose oral corticosteroids. Monthly cyclophosphamide pulses were administered for six months, and maintenance therapy with azathioprine was subsequently initiated. In addition to immunosuppressive therapy, the patient was started on optimized guideline directed medical therapy for heart failure, including furosemide (20 mg orally once daily), valsartan (80 mg orally once daily), bisoprolol (2.5 mg orally once daily), spironolactone (25 mg orally once daily), and dapagliflozin (10 mg orally once daily).The clinical course was favorable, with progressive improvement of symptoms. Follow-up echocardiography showed a significant recovery of left ventricular systolic function, with an ejection fraction improving to 45%.

Discussion:-

Cardiac sarcoidosis is a challenging diagnosis due to its heterogeneous presentation and the frequent absence of systemic sarcoidosis at the time of cardiac involvement[3]. Although autopsy studies suggest a higher prevalence of cardiac granulomas, clinically overt disease remains uncommon and is often underdiagnosed[1]. Dilated cardiomyopathy is a less frequent but severe manifestation of cardiac sarcoidosis, resulting from extensive myocardial inflammation and fibrosis[2,4]. In young patients presenting with unexplained non-ischemic cardiomyopathy, especially in the presence of extracardiac radiological findings suggestive of sarcoidosis, cardiac sarcoidosis should be considered.Biological markers such as angiotensin converting enzyme levels and calcium metabolism are neither sensitive nor specific[3], as illustrated in our patient. Endomyocardial biopsy has limited sensitivity due to the patchy distribution of granulomas and is not always feasible[2,5]. In this context, cardiac MRI has emerged as a pivotal diagnostic tool[6,7]. Late gadolinium enhancement with subepicardial, transmural or patchy distribution, particularly at ventricular insertion points, is highly suggestive of cardiac sarcoidosis and has prognostic value.

In addition to cardiac magnetic resonance imaging, positron emission tomography with fluorodeoxyglucose (18F-FDG PET/CT) plays an important complementary role in the evaluation of cardiac sarcoidosis[2,3,8]. PET imaging allows the detection of active myocardial inflammation and is particularly useful in specific clinical situations. It is recommended in patients with established extracardiacsarcoidosis who present abnormalities on screening tests for myocardial involvement, in patients younger than 60 years with unexplained de novo conduction disorders, and in patients presenting with idiopathic sustained ventricular tachycardia[1–3]. Furthermore, FDG-PET is a valuable tool for monitoring treatment response and disease activity during follow-up in patients with confirmed cardiac sarcoidosis. Although PET imaging was not performed in our patient, its use is increasingly recognized as a key modality for both diagnostic assessment and therapeutic monitoring in this condition.

Immunosuppressive therapy remains the cornerstone of treatment[3–5]. Corticosteroids are first-line therapy and may improve cardiac function and prevent disease progression. In severe or refractory cases, additional immunosuppressive agents such as cyclophosphamide, methotrexate or azathioprine are recommended[2–4]. In our patient, aggressive immunosuppressive therapy led to a marked improvement in left ventricular function, supporting the benefit of early and intensive treatment.

Conclusion:-

Cardiac sarcoidosis is a rare but serious manifestation of sarcoidosis that can present as severe dilated cardiomyopathy, particularly in young patients. Diagnosis requires a high index of suspicion and relies heavily on advanced cardiac imaging, especially cardiac MRI. Early initiation of immunosuppressive therapy may significantly improve cardiac function and prognosis. This case underscores the importance of considering cardiac sarcoidosis in unexplained cardiomyopathy and highlights the essential role of imaging in diagnosis and management.

Authors Contributions: All authors approved the final version.
Conflict of Interest: The authors declare no conflicts of interest.
Funding: No external funding was received.
Consent: Written informed consent was obtained from the patient for the publication of this case.

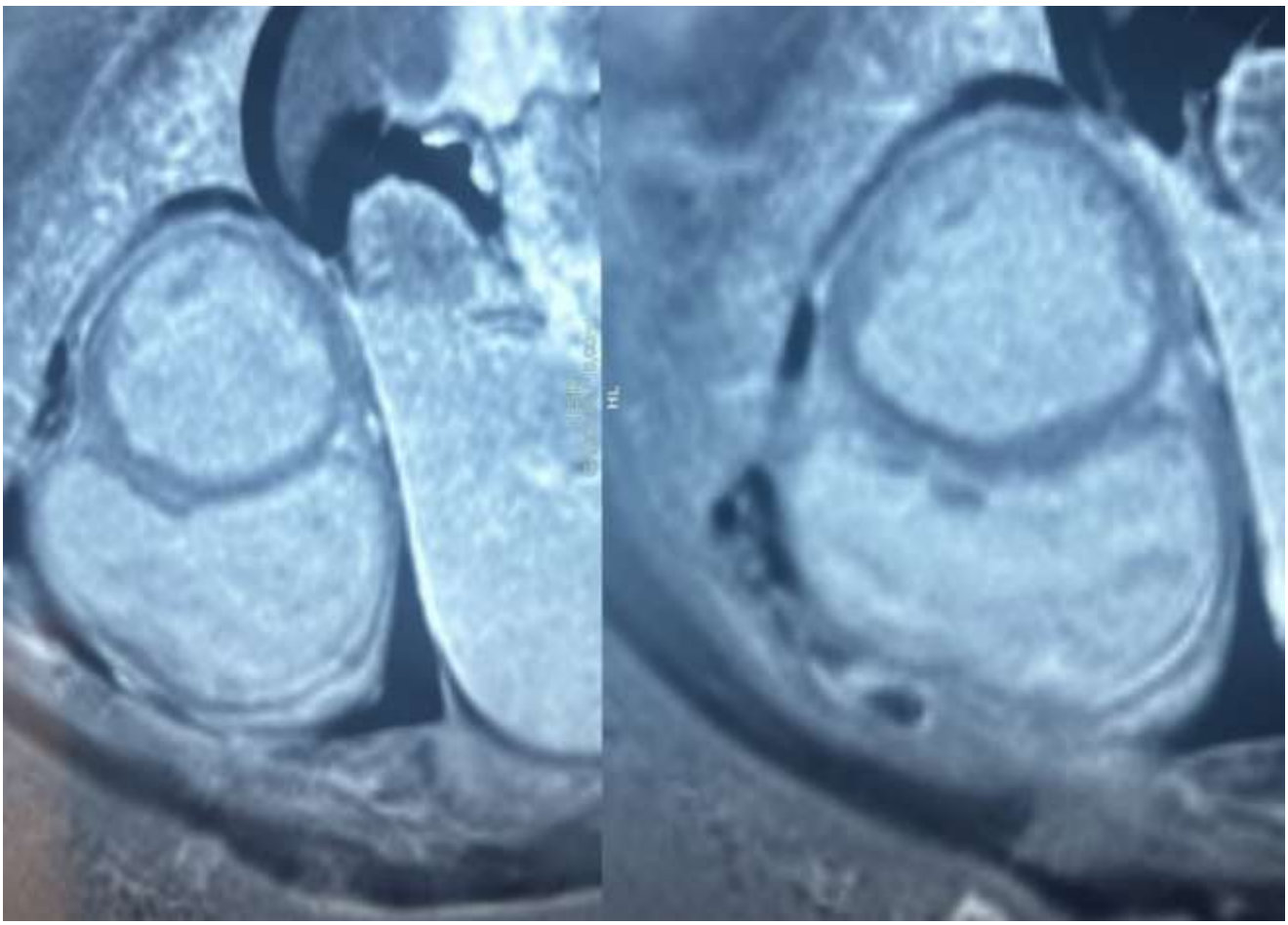

Figure 1: Cardiac magnetic resonance imaging showing transmural and subepicardial late gadolinium enhancement at the ventricular insertion points, consistent with cardiac sarcoidosis.

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