



Sarcoidosis: A Comprehensive Case Report and Discussion in a 42-Year-Old Male

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Abstract

Sarcoidosis is a systemic inflammatory disease characterized by the formation of granulomas in various organs, with pulmonary involvement being the most common. We present a case report of a 42-year-old male with sarcoidosis, emphasizing the clinical presentation, diagnostic approach, and management strategies. The patient presented with persistent cough, dyspnea, and fatigue. Imaging studies revealed bilateral hilar lymphadenopathy and diffuse reticulonodular opacities on chest X-ray, consistent with sarcoidosis. Biopsy confirmed noncaseating granulomas. Treatment with corticosteroids resulted in symptomatic improvement and resolution of lesions. This case underscores the importance of considering sarcoidosis in patients with compatible symptoms and highlights the challenges in its diagnosis and management. Early recognition and intervention are crucial to prevent irreversible organ damage and optimize outcomes. Further research is needed to elucidate the underlying pathogenesis of sarcoidosis and develop targeted therapies for improved patient care. This case report contributes to the existing literature by providing insights into the clinical course and management of sarcoidosis in a young adult male,

...serving as a reference for clinicians managing similar cases.

Keywords: Sarcoidosis, Granulomas, Pulmonary, Cutaneous, Corticosteroids

Introduction

Sarcoidosis is a complex systemic inflammatory disorder characterized by the formation of noncaseating granulomas in various organs, particularly the lungs, lymph nodes, skin, and eyes. First described over a century ago, sarcoidosis remains an enigmatic disease with elusive etiology, diverse clinical manifestations, and unpredictable course. It poses diagnostic challenges for clinicians due to its nonspecific symptoms and mimicry of other diseases, necessitating a comprehensive approach to evaluation and management. The exact cause of sarcoidosis remains unknown, although it is postulated to result from a dysregulated immune response triggered by environmental exposures in genetically susceptible individuals. Genetic predisposition plays a significant role, with studies implicating several genes involved in immune regulation, antigen presentation, and inflammatory pathways. Variations in human leukocyte antigen (HLA) alleles, particularly HLA-DRB1 and HLA-DQB1, have been associated with increased susceptibility to sarcoidosis, suggesting a genetic

component to disease pathogenesis. Environmental factors such as infections, occupational exposures, and air pollution have also been implicated, although no specific agent has been consistently identified as a causative trigger.

Sarcoidosis exhibits a diverse clinical spectrum, ranging from asymptomatic disease detected incidentally on imaging studies to fulminant multisystem involvement with significant morbidity and mortality. Pulmonary involvement is the most common presentation, observed in up to 90% of cases, and may manifest as cough, dyspnea, chest pain, or constitutional symptoms such as fatigue and weight loss. Cutaneous manifestations are also frequent, with erythema nodosum, lupus pernio, and maculopapular eruptions being the most common presentations. Extrathoracic involvement can affect virtually any organ system, including the heart, liver, spleen, central nervous system, and musculoskeletal system, leading to a myriad of clinical manifestations depending on the affected site. The diagnosis of sarcoidosis relies on a combination of clinical, radiological, and histopathological findings. Chest radiography typically demonstrates bilateral hilar lymphadenopathy, often accompanied by reticulonodular opacities or ground-glass infiltrates in the lung parenchyma. High-resolution computed tomography (HRCT) provides detailed assessment of pulmonary involvement, facilitating the detection of micronodules, consolidation, or fibrotic changes. Definitive diagnosis requires histological evidence of noncaseating granulomas on tissue biopsy, obtained from accessible sites such as lung, skin, lymph nodes, or liver. Ancillary tests including serum angiotensin-converting enzyme (ACE) levels, gallium-67 scintigraphy, and positron emission tomography (PET) scanning may aid in disease staging and monitoring but lack specificity for diagnosis.

Management of sarcoidosis aims to alleviate symptoms, prevent organ damage, and induce remission while minimizing treatment-related complications. Systemic corticosteroids, such as prednisone, are the mainstay of therapy for moderate to severe disease. Initiation of corticosteroids is warranted in symptomatic patients with progressive pulmonary involvement, extrapulmonary disease, or organ dysfunction. However, the optimal duration and dosage of corticosteroids remain controversial, with individualized regimens tailored based on disease severity and response to therapy. Adjunctive immunosuppressive agents, such as methotrexate, azathioprine, or tumor necrosis factor (TNF) inhibitors, may be considered in refractory cases or to minimize corticosteroid-related adverse effects. In addition to pharmacological interventions, lifestyle modifications and supportive measures play a crucial role in the management of sarcoidosis. Patients are advised to avoid exposure to environmental triggers such as dust, smoke, and occupational hazards, which may exacerbate inflammation and trigger disease flares. Smoking cessation is particularly important, as tobacco smoke has been implicated in the pathogenesis of sarcoidosis and is associated with disease progression and poorer outcomes. Regular follow-up with healthcare providers is essential to monitor disease activity, assess treatment response, and detect potential complications.

In summary, sarcoidosis is a heterogeneous multisystem disorder characterized by granulomatous inflammation and diverse clinical manifestations. Early recognition and prompt intervention are crucial to prevent irreversible organ damage and optimize long-term outcomes. Clinicians should maintain a high index of suspicion for sarcoidosis in patients presenting with compatible symptoms, particularly those with pulmonary and cutaneous involvement. A comprehensive diagnostic

approach including clinical evaluation, imaging studies and tissue biopsy is essential for accurate diagnosis and appropriate management. Treatment strategies should be individualized based on disease severity, organ involvement, and patient preferences, with close monitoring for treatment response and adverse effects. Further research is warranted to elucidate the underlying pathophysiology of sarcoidosis and develop targeted therapies for improved patient care.

Case Report

A 42-year-old male presented to the outpatient clinic with a four-month history of persistent dry cough, exertional dyspnea, and generalized fatigue. He denied fever, weight loss, or night sweats. His medical history was unremarkable, with no significant past illnesses or recent travel history. He worked as a construction worker and denied exposure to environmental toxins or occupational hazards. Family history was noncontributory. On physical examination, the patient appeared fatigued but was in no acute distress. Vital signs were within normal limits, with oxygen saturation of 96% on room air. Examination of the respiratory system revealed bilateral inspiratory crackles on auscultation. No palpable lymphadenopathy or skin lesions were appreciated. Cardiovascular, abdominal, and neurological examinations were unremarkable. Given the persistent cough and dyspnea, further evaluation was pursued. Chest X-ray revealed bilateral hilar lymphadenopathy with diffuse reticulonodular opacities in the lung fields. Pulmonary function tests demonstrated a restrictive pattern with reduced forced vital capacity (FVC) and forced expiratory volume in one second (FEV1). High-resolution computed tomography (HRCT) of the chest confirmed bilateral hilar and mediastinal lymphadenopathy with multiple nodular opacities scattered throughout the lungs (Figure 1). Based on the

clinical presentation and imaging findings, sarcoidosis was suspected. Laboratory investigations including complete blood count, liver and renal function tests, serum calcium levels, and angiotensin-converting enzyme (ACE) levels were within normal limits. Tuberculin skin test and sputum acid-fast bacilli smear were negative. Flexible bronchoscopy with transbronchial lung biopsy was performed, revealing noncaseating granulomas consistent with sarcoidosis. The patient was initiated on oral prednisone at a dose of 40 mg daily. Follow-up evaluation after four weeks showed significant improvement in symptoms, with resolution of cough and dyspnea. Repeat chest X-ray demonstrated regression of hilar lymphadenopathy and pulmonary opacities. Prednisone was tapered over six months with close monitoring for disease recurrence or treatment-related complications. The patient remained asymptomatic at one-year follow-up, with stable pulmonary function tests and no evidence of disease progression.

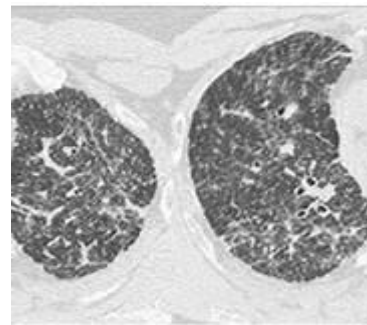


Fig. 1: HRCT

Discussion

Sarcoidosis is a complex disease with diverse clinical presentations, variable disease courses, and challenges in diagnosis and management. The discussion section delves into several key aspects of sarcoidosis, including its pathogenesis, clinical manifestations, diagnostic approach, treatment strategies, and prognosis.

Pathogenesis: The exact etiology of sarcoidosis remains elusive, although it is widely believed to result from a dysregulated immune response to environmental triggers in genetically susceptible individuals. The hallmark histopathological finding of noncaseating granulomas reflects an aberrant immune reaction characterized by the accumulation of activated macrophages and T lymphocytes at sites of inflammation. Multiple factors contribute to the development of sarcoidosis, including genetic predisposition, environmental exposures, infectious agents, and immune dysregulation. Genetic susceptibility plays a significant role in sarcoidosis pathogenesis, with studies identifying associations with specific human leukocyte antigen (HLA) alleles, cytokine polymorphisms, and immune regulatory genes. Variations in HLA-DRB1 and HLA-DQB1 genes have been implicated in sarcoidosis susceptibility, suggesting a role for antigen presentation in disease initiation. Polymorphisms in cytokine genes such as tumor necrosis factor-alpha (TNF- α), interleukin-2 (IL-2), and interferon-gamma (IFN- γ) have also been implicated in disease susceptibility and severity, highlighting the importance of cytokine-mediated immune responses in sarcoidosis pathogenesis. Environmental factors may trigger or exacerbate sarcoidosis in genetically susceptible individuals, although specific agents remain elusive. Occupational exposures to organic and inorganic dusts, such as beryllium, silica, and metal fumes, have been implicated in sarcoidosis development, particularly in high-risk occupations such as construction workers, firefighters, and healthcare workers. Infections, including mycobacteria, *Propionibacterium acnes*, and Epstein-Barr virus, have also been implicated as potential triggers of sarcoidosis, although conclusive evidence is lacking.

Clinical Manifestations: Sarcoidosis can affect virtually any organ system, leading to a wide spectrum of clinical

manifestations ranging from asymptomatic disease to severe multisystem involvement. Pulmonary involvement is the most common presentation, observed in up to 90% of cases, and may manifest as cough, dyspnea, chest pain, or constitutional symptoms such as fatigue and weight loss. Cutaneous manifestations are also frequent, with erythema nodosum, lupus pernio, and maculopapular eruptions being the most common presentations. Extrapulmonary involvement can affect the eyes, heart, liver, spleen, central nervous system, and musculoskeletal system, leading to a myriad of clinical manifestations depending on the affected site. The clinical course of sarcoidosis varies widely among individuals, with some experiencing spontaneous remission and others developing chronic progressive disease with significant morbidity and mortality. The presence of extrapulmonary involvement, particularly cardiac, neurologic, or renal disease, is associated with a poorer prognosis and increased mortality. Conversely, isolated pulmonary disease with minimal symptoms and preserved lung function may have a more favorable prognosis, although progression to fibrotic lung disease can occur in a subset of patients.

Diagnostic Approach: The diagnosis of sarcoidosis requires a comprehensive approach integrating clinical, radiological, and histopathological findings. Chest radiography typically demonstrates bilateral hilar lymphadenopathy, often accompanied by reticulonodular opacities or ground-glass infiltrates in the lung parenchyma. High-resolution computed tomography (HRCT) provides detailed assessment of pulmonary involvement, facilitating the detection of micronodules, consolidation, or fibrotic changes. Definitive diagnosis requires histological evidence of noncaseating granulomas on tissue biopsy, obtained from accessible sites such as lung, skin, lymph nodes, or liver. Ancillary

tests including serum angiotensin-converting enzyme (ACE) levels, gallium-67 scintigraphy, and positron emission tomography (PET) scanning may aid in disease staging and monitoring but lack specificity for diagnosis. The differential diagnosis of sarcoidosis includes infectious, neoplastic, and autoimmune disorders, necessitating a thorough evaluation to exclude alternative etiologies.

Treatment Strategies: Management of sarcoidosis aims to alleviate symptoms, prevent organ damage, and induce remission while minimizing treatment-related complications. Systemic corticosteroids, such as prednisone, are the mainstay of therapy for moderate to severe disease. Initiation of corticosteroids is warranted in symptomatic patients with progressive pulmonary involvement, extrapulmonary disease, or organ dysfunction. However, the optimal duration and dosage of corticosteroids remain controversial, with individualized regimens tailored based on disease severity and response to therapy. Adjunctive immunosuppressive agents, such as methotrexate, azathioprine, or tumor necrosis factor (TNF) inhibitors, may be considered in refractory cases or to minimize corticosteroid-related adverse effects. Biologic agents targeting specific cytokines or immune pathways, such as TNF inhibitors or interleukin-2 receptor antagonists, hold promise as potential therapeutic options for sarcoidosis, although further research is needed to elucidate their efficacy and safety profile.

Prognosis: The prognosis of sarcoidosis varies widely depending on disease severity, extent of organ involvement, and response to therapy. The majority of patients with sarcoidosis have a favorable prognosis, with spontaneous remission or stable disease observed in up to two-thirds of cases. However, a subset of patients may experience chronic progressive disease with significant

morbidity and mortality, particularly those with extra pulmonary involvement or fibrotic lung disease. Long-term follow-up is essential to monitor disease activity, assess treatment response, and detect potential complications, including pulmonary fibrosis, pulmonary hypertension, and cardiac arrhythmias.

Conclusion

Sarcoidosis is a heterogeneous multisystem disorder characterized by granulomatous inflammation and can manifest with a wide spectrum of clinical features. Early recognition and prompt intervention are crucial to prevent irreversible organ damage and optimize long-term outcomes. Clinicians should maintain a high index of suspicion for sarcoidosis in patients presenting with compatible symptoms, particularly those with pulmonary and cutaneous manifestations. A comprehensive diagnostic approach including clinical evaluation, imaging studies, and tissue biopsy is essential for accurate diagnosis and appropriate management. Treatment strategies should be individualized based on disease severity, organ involvement, and patient preferences, with close monitoring for treatment response and adverse effects. Further research is warranted to elucidate the underlying pathophysiology of sarcoidosis and develop targeted therapies to improve outcomes and quality of life for affected individuals.

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