Case Report

International Journal of Clinical Rheumatology

A Rare Presentation of Mixed Connective Tissue Disease in a Young Female Patient

Abdi Barre*

Department of Physiology, University of Haiti, Haiti

***Author for Correspondence:**
Abdi89@gmail.com

Received: 01-Aug-2024, Manuscript No. fmijcr-24-150686; **Editor assigned:** 03-Aug-2024, Pre-QC No. fmijcr-24-150686 (PQ); **Reviewed:** 16-Aug-2024, QC No. fmijcr-24-150686; **Revised:** 22-Aug-2024, Manuscript No. fmijcr-24-150686 (R); **Published:** 29-Aug-2024, **DOI:** 10.37532/1758-4272.2024.19(8).212-214

Abstract

Mixed connective tissue disease (MCTD) is an autoimmune disorder characterized by features of multiple connective tissue diseases, primarily systemic lupus erythematosus, scleroderma, and polymyositis. This case report presents a unique presentation of MCTD in a 25-year-old female patient who exhibited atypical symptoms and highlights the challenges in diagnosis and management.

Keywords: Mixed Connective Tissue Disease (MCTD) • Autoimmune Disorder • Antinuclear Antibodies (ANA) • Anti-U1 Ribonucleoprotein (RNP) • Raynaud's Phenomenon

Introduction

Mixed connective tissue disease (MCTD) is a rare autoimmune disorder characterized by overlapping features of multiple connective tissue diseases, including systemic lupus erythematosus (SLE), scleroderma, and polymyositis. It was first described by Sharp et al. in the 1970s, and it primarily affects young women, often presenting with a combination of symptoms that can lead to diagnostic challenges. The clinical manifestations of MCTD are diverse, with patients frequently exhibiting Raynaud's phenomenon, arthritis, skin rashes, and muscle weakness. Early recognition and diagnosis are crucial, as they significantly influence management strategies and long-term outcomes. MCTD is associated with specific autoantibodies, notably anti-U1 ribonucleoprotein (RNP), which serve as important diagnostic markers. The treatment typically involves immunosuppressive therapy, including corticosteroids and disease-modifying antirheumatic drugs (DMARDs), tailored to the individual patient's symptoms and disease activity [1-3]. This case report presents a unique and atypical presentation of MCTD in a 25-year-old female patient, highlighting the complexities involved in diagnosing and managing this multifaceted disease. By discussing this case, we aim to enhance awareness of MCTD among healthcare providers and emphasize the importance of a thorough clinical evaluation and a multidisciplinary approach to treatment [4].

Case Presentation

A 25-year-old female presented to the outpatient rheumatology clinic with complaints of fatigue, joint pain, and skin changes over the past six months. Initially, she noted intermittent swelling in her fingers and wrists, accompanied by morning stiffness lasting more than an hour. These symptoms were gradually worsening, prompting her to seek medical attention [5].

The patient also reported a significant rash on her face, resembling a butterfly pattern, along

with Raynaud's phenomenon characterized by color changes in her fingers in response to cold exposure. Additionally, she experienced muscle weakness, particularly in her proximal muscles, affecting her ability to climb stairs and lift objects.

Her medical history was unremarkable, with no prior autoimmune diseases, and she had no family history of connective tissue disorders. The patient was a non-smoker, and her vital signs were within normal limits at presentation [6-8].

Laboratory and Imaging Studies

Laboratory tests revealed a positive antinuclear antibody (ANA) test with a titer of 1:640, predominantly nucleolar staining. Further serological tests showed elevated anti-U1 ribonucleoprotein (RNP) antibodies,

supporting the diagnosis of MCTD. Complement levels (C3 and C4) were within normal ranges, and inflammatory markers (ESR and CRP) were mildly elevated.

A complete blood count showed mild leukopenia and anemia, while renal function tests were normal. Imaging studies, including X-rays of the hands, did not reveal any erosive changes. An echocardiogram was performed to assess any potential pulmonary hypertension, which was normal [9].

Diagnosis

Based on the clinical presentation and laboratory findings, the patient met the diagnostic criteria for MCTD, characterized by the presence of clinical features from lupus, scleroderma, and polymyositis. The combination of Raynaud's phenomenon, arthritis, skin rash, and muscle weakness were consistent with this diagnosis.

Management and Treatment

The patient was started on a treatment regimen consisting of low-dose corticosteroids (prednisone 10 mg daily) to manage inflammation and alleviate her symptoms. In addition, hydroxychloroquine was initiated to provide symptomatic relief and to help manage the autoimmune process. The patient was advised on lifestyle modifications, including sun protection measures for her skin and the importance of avoiding cold exposure to manage Raynaud's phenomenon [10].

After three months of treatment, the patient reported significant improvement in her joint pain and muscle strength. The skin rash also improved with the corticosteroid therapy. However, she continued to experience occasional episodes of Raynaud's phenomenon, prompting the addition of a calcium channel blocker (nifedipine) to help manage these symptoms.

Follow-Up and Outcomes

At the six-month follow-up appointment, the patient reported continued improvement in her overall condition. Joint symptoms were minimal, and muscle strength had significantly improved. Laboratory tests showed stable anti-U1 RNP levels, while inflammatory markers returned to normal. The skin rash had resolved, and the patient was adhering well to her treatment plan.

Patient education played a crucial role in her management, as she was instructed on the importance of regular follow-ups and monitoring for potential complications such as pulmonary hypertension or renal involvement.

Discussion

Mixed connective tissue disease can present with a variety of symptoms, often leading to diagnostic challenges. This case illustrates how atypical presentations, such as muscle weakness and skin involvement, can complicate the diagnosis. The presence of anti-U1 RNP antibodies is a hallmark of MCTD and can help differentiate it from other autoimmune diseases.

Management of MCTD typically involves immunosuppressive therapies to control inflammation and prevent organ damage. Corticosteroids and hydroxychloroquine are commonly used, while additional medications may be required to address specific symptoms, such as Raynaud's phenomenon.

The long-term prognosis for patients with MCTD varies; early recognition and appropriate treatment can lead to improved outcomes. This case emphasizes the importance of a multidisciplinary approach, involving rheumatologists, dermatologists, and primary care providers, to ensure comprehensive care.

Conclusion

This case report highlights a rare presentation of mixed connective tissue disease in a young female patient. It underscores the need for clinicians to consider MCTD in patients with overlapping features of autoimmune disorders. Early diagnosis and individualized treatment are essential for optimizing patient outcomes and enhancing quality of life. Ongoing research into the pathophysiology and treatment of MCTD will further improve our understanding and management of this complex condition.

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