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Anti-MDA5 Positive Juvenile Dermatomyositis Presenting with Respiratory Manifestations: A Case Report

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Abstract

We report the case of a 16-year-old girl who presented with heliotrope rash, Gottron's papules, and proximal muscle weakness, consistent with juvenile dermatomyositis (JDM), a rare chronic inflammatory multisystem disease of childhood. Laboratory evaluation revealed elevated CA-19-9 and CA-15-3 tumor markers, as well as anti-MDA5 and anti-Ro52 antibodies. Anti-MDA5 is strongly associated with rapidly progressive interstitial lung disease (RP-ILD) and portends an unfavorable prognosis if not promptly managed. The patient was treated aggressively with systemic corticosteroids, methotrexate, and hydroxychloroquine, resulting in rapid resolution of symptoms and complete clearance of pulmonary abnormalities on follow-up imaging. This case underscores the importance of early recognition and treatment of anti-MDA5 positive JDM to prevent life-threatening complications.

Keywords: Anti-MDA5, Juvenile Dermatomyositis (JDM), Rapidly Progressive Interstitial Lung Disease (RP-ILD)

Introduction

Juvenile dermatomyositis (JDM) is a rare, complex, and potentially life-threatening autoimmune disease of childhood, primarily affecting proximal muscles and skin. The disease typically presents between the ages of 5 and 10 years, with a female predominance (2:1). Its annual incidence is estimated at two to four cases per million children. Although the exact etiology is unknown, both genetic and environmental factors contribute to its pathogenesis.

Hallmark clinical features include Gottron's papules and heliotrope rash, which are considered pathognomonic. Other features may include calcinosis, periorbital edema, and vasculopathic complications affecting the gastrointestinal tract, skin, or central nervous system.

Autoantibody testing plays an important role in disease classification and prognosis. Anti-MDA5 antibodies are associated with amyopathic dermatomyositis and RP-ILD, whereas anti-Ro52 antibodies are also linked to ILD, particularly organizing pneumonia. Tumor markers such as CA-15-3, CA-19-9, and CA-125 may be useful in malignancy screening in patients with dermatomyositis, especially in adults or in cases without ILD.

We present an unusual case of anti-MDA5 positive JDM with respiratory manifestations in an adolescent girl, emphasizing the significance of early recognition and aggressive immunosuppressive therapy.

Case Presentation

A 16-year-old female initially presented to a dermatology clinic with a rash on her hands, which was treated as atopic dermatitis. She was later referred to Suhar Hospital with new onset of proximal muscle weakness, small joint tenderness with swelling, and a characteristic heliotrope rash involving the face and chest ("shawl sign"). Additional findings included periorbital edema, Gottron's papules, erythematous lesions on extensor surfaces, palmar vascular lesions, and painful oral ulcers with dysphagia.

Laboratory investigations revealed mild thrombocytopenia, microcytic anemia, elevated AST, ALT, and creatine kinase. A myositis antibody panel showed positivity for anti-MDA5 and anti-Ro52 antibodies.

She was initially treated with prednisolone (30 mg daily) and methotrexate (15 mg weekly), which led to improvement in both symptoms and laboratory parameters. However, upon steroid tapering, she relapsed with recurrence of rash and weakness.

Comprehensive imaging was performed, including whole-body CT and tumor marker screening. CA-19-9 and CA-15-3 were elevated. Chest CT demonstrated bilateral small irregular lung nodules, scattered peripheral alveolar densities, and subpleural opacities in the basal lower lobes. Abdominal CT revealed a bulky pancreas with a small hypodense lesion in the body (9.5 × 7 mm). Bronchoscopy ruled out malignancy and tuberculosis.

She was restarted on prednisolone (1 mg/kg) with methotrexate (20 mg subcutaneous weekly) and hydroxychloroquine (200 mg daily). The possibility of escalation with intravenous immunoglobulin (IVIG) or rituximab was discussed but deferred due to good clinical response.

On follow-up, the patient demonstrated marked clinical improvement. Repeat CT chest showed complete resolution of pulmonary lesions with no pleural effusion or thickening. Abdominal CT showed persistence of a small stable pancreatic lesion. Physical examination revealed residual Gottron's papules, with marked improvement in muscle strength and skin disease.

Discussion

JDM is broadly classified into classical JDM and clinically amyopathic JDM. Immune dysregulation remains central to its pathogenesis. Our patient had hallmark skin manifestations along with proximal muscle weakness, fulfilling the criteria for classical JDM.

Anti-MDA5 antibodies are strongly associated with rapidly progressive ILD and poor prognosis. Serum levels correlate with disease activity and outcomes. Although anti-MDA5 is usually linked to amyopathic DM, our patient demonstrated clear muscular involvement, highlighting the heterogeneity of disease expression.

Early aggressive treatment was essential in preventing progression of ILD in this case. Combination therapy with corticosteroids, methotrexate, and hydroxychloroquine achieved complete radiological resolution of pulmonary disease. Although biologics such as IVIG or rituximab may be required in refractory cases, our patient responded well to conventional therapy.

The incidental pancreatic lesion and elevated tumor markers emphasize the need for careful long-term surveillance, as dermatomyositis can be a paraneoplastic phenomenon. However, pediatric cases are rarely associated with malignancy.

This case highlights the critical role of early recognition of anti-MDA5 positivity and pulmonary screening in JDM, as timely immunosuppressive therapy may dramatically alter the disease course and prevent respiratory failure.

Notes on Patient Consent

The authors certify that they obtained all appropriate patient consent. The patient's guardian provided verbal consent for the publication of clinical information and images. Identifying details have been withheld.

References

1. Juvenile Dermatomyositis. Arthritis Foundation. Accessed August 29, 2022. Available at: https://www.arthritis.org/diseases/juvenile-dermatomyositis
2. American College of Rheumatology. Diseases & Conditions: Juvenile Dermatomyositis. Accessed August 29, 2022. Available at: https://www.rheumatology.org/i-am-a/patient-caregiver/diseases-conditions
3. Gupta P, Shruti S, Chaudhary V, Khullar G, Siraj F. Juvenile dermatomyositis: A case report and review of literature. Cureus. 2019; 11(1):e3885. Doi: 10.7759/cureus.3885
4. Martin N, Li CK, Wedderburn LR. Juvenile dermatomyositis: New insights and new treatment strategies. Ther Adv Musculoskelet Dis. 2012; 4(3):139-147. Doi: 10.1177/1759720X11436242