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Case Report

Dermatomyositis with Normal CPK, Elevated Aldolase, and Borderline Anti-PL7 Antibody Positivity: A Case Report

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Abstract

Dermatomyositis is an idiopathic inflammatory myopathy typically characterized by proximal muscle weakness, elevated muscle enzymes, and characteristic skin manifestations. We report a case of a 64-year-old male who presented with classic clinical features but had normal serum creatine phosphokinase (CPK) levels and elevated aldolase, posing a diagnostic challenge. MRI findings, muscle biopsy and serologic test confirmed dermatomyositis with borderline positivity for Anti-PL7 antibodies. This case highlights the importance of a comprehensive diagnostic approach in inflammatory myopathies.

Keywords: Dermatomyositis; Aldolase; Anti-PL7 Antibody; Normal CPK; Muscle Biopsy; MRI

Introduction

Dermatomyositis (DM) is a systemic autoimmune condition primarily affecting skeletal muscles and skin. Diagnosis typically relies on clinical presentation, elevated serum muscle enzymes, electromyography (EMG), imaging, autoantibodies, and muscle biopsy findings. Although elevated CPK is a hallmark, atypical case with normal CPK but elevated aldolase has been described, often complicating the diagnostic process.

Aldolase can be more sensitive marker in cases with less muscle necrosis but ongoing muscle fibre injury [1].

Additionally, the presence of myositis -specific antibodies, such as anti -PL7, can guide diagnosis and management.

Case Presentation

A 64-year-old male presented with bilateral proximal muscle weakness of the upper limbs (shoulders) and lower limbs (thighs), more severe on the right side, along with associated myalgia. There was no history of rash, dysphagia, or respiratory symptoms. He is diabetic and hypertensive for 2 years and on oral medications.

Physical examination

- Bilateral weakness of hip flexors (Medical Research council grade [MRC] grade 3/5)
- Right knee jerk absent

Investigations

- Total Count: 32,910
- Differential count: N-70.5% L-20.1% E-3.9% M-5% B-0.5%
- Platelet count: 3.94L
- ESR: 98
- Serum CPK: 15
- CRP: 238
- Serum aldolase: Elevated
- Anti -PL7 antibody: Borderline positive
- Urine myoglobin: Negative
- MRI Thigh: Patchy T2/STIR hyperintense signal changes involving gluteus maximus, minimus, and anterior and medial compartments of right thigh.
- MRI (T2 -Weighted STIR) showing irregular high signal intensities scattered throughout the muscle tissues more on the right thigh.
- Muscle biopsy (right thigh): Perifascicular atrophy and inflammatory infiltrates consistent with dermatomyositis [3].
- Blood investigations ruled out infections, paraneoplastic etiologies, and other connective tissue disorders.

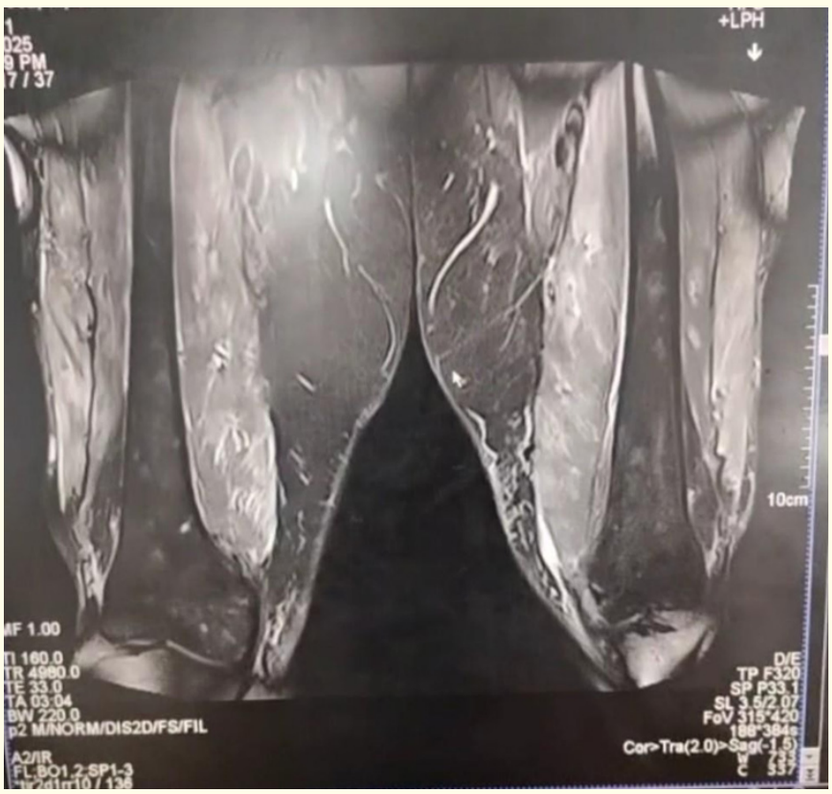

Figure 1

Treatment

- Patient was initiated on immunosuppressive therapy with IV corticosteroids (INJ. METHYL PREDNISOLONE 500MG IV OD) for 5days and then changed to oral steroids (TAB WYSOLONE 20MG OD) for 5 days.
- The patient responded well to steroids, which is consistent with standard treatment protocols for inflammatory myopathies [1].

Discussion

- Dermatomyositis often presents with elevated CPK and aldolase, however, isolated aldolase elevation may occur, particularly in cases of predominant muscle fibre injury without extensive necrosis. This can delay diagnosis if clinician rely solely on CPK levels.
- Anti-PL7 antibodies, part of the Antisynthetase syndrome profile. These autoantibodies are formed against aminoacyl transfer RNA synthatases (ARS). Eight types of anti-ARS have been identified: anti- histidyl (anti-Jo1), anti-threonyl (anti-PL7), alanyl (anti- PL12), glycyl (anti-EJ), isoleucyl (anti- OJ), asparaginyl (anti-KS), phenylalanyl (anti-Zo), tyrosyl (anti-Ha). They are usually associated with interstitial lung disease (ILD), arthritis, and myositis, Raynaud's phenomenon, fever, and/or mechanic's hand.
- Anti-PL7 positivity, even borderline, suggests a myositis subtypes, reinforcing the diagnosis in the absence of elevated CPK [2].
- However in this case lung involvement was absent at presentation. MRI is invaluable for detecting muscle edema, especially when enzyme levels are inconclusive.

Conclusion

- Dermatomyositis can present with atypical biochemical profiles, including normal CPK levels. Not excluding dermatomyositis in the presence of normal CPK alone [1].
- Utilizing aldolase, MRI, and muscle biopsy when laboratory findings are atypical [3].
- Recognizing anti-synthetase antibodies, even borderline, as important diagnostic clues [2].
- Early initiation of immunosuppressive therapy improves outcomes, particularly in atypical presentation.

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