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A Case Report on Rare Presentation of Sporadic Disease: Dermatomyositis Sine Dermatitis—Diagnosis and Management

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Abstract

Dermatomyositis sine dermatitis (DMSD) is one of the rare idiopathic inflammatory myopathies. Based on predominant symptoms faced by patients, it is classified into 3 types: (1) classic dermatomyositis (DM), where patients have both muscle and skin symptoms; and (2) amyopathic DM, when only skin symptoms present with no muscle involvement. Whereas (3) DMSD has mainly muscle symptoms with muscle antibodies but no skin rashes. There have been only nearly 10 published articles about DMSD proving this disease's scarcity. At the same time, it shows the importance of discussing the unusual presentation of such a rare disease. Here we present, a 28-year-old woman with worsening proximal muscle weakness. The decreased muscle strength on physical examination and elevated creatinine kinase required more work up for autoimmune disease. Interestingly, on muscle biopsy, anti-melanoma differentiation-associated gene 5 (anti-MDA5) antibody returned positive, and the patient responded well to 3 days course of steroids. The lack of skin involvement, the predominance of muscle symptoms, and positive anti-MDA5 antibody indispensably diagnosed patients with DMSD. The previously published articles have proved the association between anti-NXP-2 antibody and DMSD, which was not seen in our case. The systemic involvement of DMSD can lead to interstitial lung disease, where due to diffuse alveolar damage and pulmonary fibrosis, patients end up requiring intubation and may be associated with higher-level mortality. In our case, chest X-rays and computed tomography (CT) scans were unremarkable for lung involvement, so as no paraneoplastic syndromes were present, which has also been reported in DMSD patients previously.

Keywords

rheumatology, dermatology, diagnostic testing

Introduction

Dermatomyositis (DM) is one of the rare idiopathic inflammatory myopathies. The patient presents with symptoms of skin rash, muscle weakness, and extra muscular involvement, such as dysphagia, interstitial lung disease (ILD), and myocarditis.1 Based on clinical presentation, DM is classified into 3 categories: (1) classic DM with skin and muscle symptoms, where patients usually have proximal muscle weakness associated with skin rashes such as Gottron's papules and heliotrope rash; (2) amyopathic DM, where the patient presents with skin symptoms without any muscular involvement or weakness.[1] In contrast, (3) dermatomyositis sine dermatitis (DMSD) has myopathy symptoms with muscle antibodies but no skin rashes.[1] However, DMSD usually has anti-melanoma differentiation-associated gene 5 (anti-MDA5) antibody-positive, ILD, and also accounts for poor prognosis.[1] Here, the patient presented with symptoms of DMSD but without any lung involvement and showed an excellent response to steroids. Around 8% of DM accounts for DMSD confirmed with muscle biopsy.[2]

Case Report

A 28-year-old African American woman with no significant past medical history presented with interval worsening proximal muscle weakness for the last 1 year, making her activities of daily living (ADLs) such as combing hair, sitting up and sitting down on a chair, and taking staircase difficult

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associated with dysphagia for the same period making her choke on food relieved on drinking water. On physical examination, decreased muscle strengths 4/5 in all 4 extremities were noted, along with the rest of the normal neurological and systemic examination. The elevated creatinine kinase levels required more evaluation of autoimmune disease. A detailed rheumatologic workup including antinuclear antibody (ANA), Anti-Smith, antinuclear ribnucleoprotein (anti-RNP), angiotensin converting enzyme (ACE) levels, anticardiolipin, anti-centromere, anti–Sjögren's-syndrome-related antigen A autoantibodies (anti-SSA), anti-Sjogren's Syndrome b (anti-SSB), antitopoisomerase I (anti-SCL-70), Anti-Jo1, Anti-dsDNA, mayo clinic myositis panel, anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase (anti-HMGCOA), and anti-single-nucleotid polymorphism (anti-SNP) was done. Also, a muscle biopsy was performed, followed by 3 days of intravenous (IV) Solu-Medrol 500 mg twice a day, and a noticeable improvement in muscle weakness was seen. Out of all, the ANA with titers of 1:5120, anti-Smith, and anti-RNP returned positive. The muscle biopsy showed atrophy, necrosis, and regeneration, with associated patchy endomysial fibrosis and chronic inflammation; also positive anti-MDA5 antibody, highly specific for DM. A computed tomography (CT) chest with contrast showed axillary, hilar, and submental lymphadenopathy but no signs of ILD. The XR barium swallow showed no abnormality in the esophagus, requiring no further intervention for dysphagia. Positive anti-Smith but negative anti-dsDNA ruled out SLE, and positive anti-RNP with no other associated symptoms rules out mixed connective tissue disease. Based on symptoms and autoimmune workup, the patient was diagnosed with DMSD. The patient was discharged with prednisone 80 mg everyday with gastrointestinal (GI) prophylaxis and outpatient rheumatology follow-up. Also, on her primary care physician follow-up visit after discharge, she endorsed improved ADLs with steroid treatment.

Discussion

This section highlights how this patient's presentation and contradictory autoimmune marker make it an unusual yet interesting subtype of DM. Dermatomyositis has different types based on muscle and skin, and only muscle or skin involvement.[1] Dermatomyositis sine dermatitis is diagnosed when only muscle weakness is present without any skin symptoms, such as Gottron's papule and heliotrope rash, muscle biopsy changes, and muscle antibody suggestive of DMSD, which was seen in our patient. Anti-MDA5 antibody is one of the specific antibodies to DM and is more specific to amyopathic DM than DMSD; also, patients with DMSD who have anti-nuclear matrix protein 2 [NXP-2] positive show more specific antibody marker for DMSD than any other subtypes of DM.[2] The cohort study published by Inoue et al[2] showed the existence of DMSD in the DM group of patients, and also the significant association between anti-NXP-2 antibody and DMSD. Despite the previously proven association of the anti-NXP-2 antibody and DMSD, we could not find this association in our case, as muscle biopsy was negative for the anti-NXP-2 antibody.[3] The DMSD patients with anti-NXP-2 antibody-positive have noticed rashes after muscle biopsy was done; contradictory in our case, the patient did not have skin rash throughout the hospital course or even after.[2] As previously mentioned, the anti-MDA5 antibody correlated with amyopathic DM; however, in this case, the patient diagnosed with DMSD showed positive anti-MDA5 antibody.[2,3]

The DMSD also causes multi-system involvement apart from muscular symptoms such as myocarditis, dysphagia, neuromayositis, and interstitial lung disease (ILD).[1] The positive anti-MDA5 antibody usually has associated with ILD; negative CXR and CT chest for ILD did not prove this correlation in our case despite the positive anti -MDA5 antibody.[1,2] The anti-MDA5 antibody in DM patients correlates with the severity of lung disease in terms of ILD, respiratory muscle weakness with poor outcome, and an inadequate response to immunosuppressive therapy.[1,4]

In contrast, our patient has anti-MDA5 antibody-positive with no lung involvement and showed an excellent response to steroids making it a good prognostic disease.[3,4]

The paraneoplastic DMSD presentation with paraneoplastic cardiac involvement with classic DMSD symptoms elevated creatine kinase (CK) and cardiac enzymes have also been seen in a recently diagnosed patient with renal cell carcinoma.[5] On the contrary, a patient diagnosed with DMSD treated with prednisone presented a few days later with worsening muscle weakness, eventually diagnosed with a tumor of the renal excretion system; however, the symptoms finally improved after the nephrectomy.[6] These studies showed neoplastic and paraneoplastic correlation with DMSD, and it is important to rule out patients with all neoplastic and paraneoplastic sources after diagnosing with DMSD, which was unremarkable in our case.[5,6]

Unfortunately, the diagnosis was initially underlooked because of no lung involvement, which is typically seen with anti-MDA5 DM.[1,4] But the presence of this antibody on muscle biopsy (Figure 1) with muscle symptoms makes it specific to DMSD.[5] The excellent responsiveness to steroids and the lack of extensive disease involvement such as ILD or malignancy make it a good prognosis.[5,6]

Conclusion

Different types of DM have been noticed, but DMSD is yet a sporadic and challenging disease to diagnose. The patient's presentation with only muscle involvement and positive anti-MDA5 antibody makes it unique and also challenging to diagnose. The positive anti-MDA5 antibody with lack of lung involvement and sufficient response to the given treatment makes it one of the exclusive presentations of DMSD, which is different than the previously published articles. Fortunately, this patient's excellent response to steroids is a positive sign. It

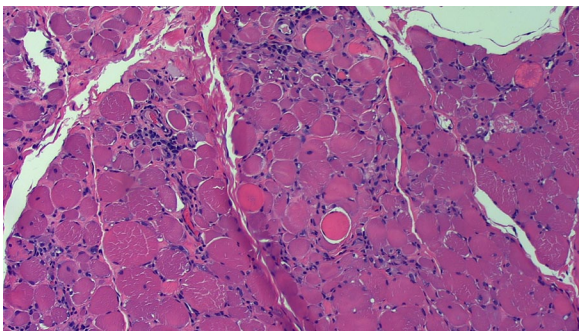

Figure 1. The muscle biopsy showed atrophy, necrosis, and regeneration with associated patchy endomysial fibrosis and chronic inflammation. The biopsy was also positive anti-MDA5, an antibody highly specific for dermatomyositis.

is important to note that this article shows a different presentation of DMSD and the management aspect of treating the disease. This article would be resourceful for patients with a similar presentation and provide information on diagnosing and treating the disease.

Authors' Note

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