CASE REPORT

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Cutaneous Mucinosis and Anti-Jo-1 Dermatomyositis: An Atypical Case

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Abstract

Dermatomyositis (DM) is a type of idiopathic inflammatory myopathy (IIM) with an incidence of 9.63/million, characterised by muscle weakness, typical cutaneous features, and myositis-specific and myositis-associated antibodies. Cutaneous mucinosis is a heterogeneous group of excess mucin deposition in the dermis leading to the waxy appearing papules, plaques, or nodules, ranging from self-healing mucinosis to severe forms like scleromyxoedema. Significant mucinosis is a well-known entity in systemic lupus erythematosus, but is much less reported in dermatomyositis. We are reporting an interesting atypical case of significant mucinosis in a patient with dermatomyositis.

Key words: Dermatomyositis, scleromyxoedema, mucinosis, anti-Jo1-antibody.

Introduction

Dermatomyositis (DM) is a rare autoimmune disease characterised by proximal muscle weakness, distinctive cutaneous manifestations, and the presence of myositis-specific and myositis-associated autoantibodies leading to distinctive phenotypic pattern, prognosis, and organ involvement[1]. The classic dermatological features include Gottron's papules, V-like sign, shawl sign, and heliotrope rash. However, atypical presentations can make the diagnosis difficult. Cutaneous mucinosis is characterised by excess mucin deposition within the dermis, leading to waxy appearing papules, plaques, or nodules[2]. But it is rarely reported in dermatomyositis. This article highlights an atypical case of significant cutaneous mucinosis in a patient with anti-Jo-1 dermatomyositis, underscoring the diagnostic complexities and the importance of integrating clinical, histopathological, and immunological findings for accurate disease characterisation and management.

Case Discussion

A 69-year-old woman presented with complaints of a hyperpigmented rash on both legs, arms, and abdomen, weakness of proximal muscles of lower limbs and upper limbs, generalised malaise, and low-grade fever from 8 - 10 months. Examination showed diffuse erythematous scaly plaques on the abdomen, both arms, and legs. Her ANA by immunofluorescence assay showed Nuclear speckled pattern with an end titre of 1:320, and the ENA reflex showed positive anti SS-B antibody. Anti-Jo-1 Myositis-specific antibody came out to be positive. Magnetic resonance imaging of bilateral thighs showed patchy areas of oedema in bilateral gluteus maximus, proximal thigh, long head of biceps femoris, vastus lateralis, and right vastus medialis and intermedius. Electromyography showed an abnormal myopathic pattern. Muscle biopsy showed mild endomysial inflammation. Skin biopsy showed perivascular lymphocytic infiltrate, interface dermatitis, and mucin deposition. The erythrocyte sedimentation rate (ESR) was 28 mm/hr, and C-Reactive Protein (CRP) level was 12 mg/L. Computed tomography of showed a few soft tissue density nodules and no evidence of interstitial lung disease or malignancy. Thyroid profile, creatine kinase, bilirubin, transaminases, serum proteins, protein electrophoresis, and renal function tests were normal. Rheumatoid factor, anti-CCP antibody, and viral markers were negative. A workup for paraproteinaemia and malignancy was negative. 2017 EULAR/ACR classification criteria score for IIM was 10.1 s/o Definite IIM. According to the clinical profile, histopathological, and radiological imaging, a diagnosis of dermatomyositis with cutaneous mucinosis seemed most likely. She was given pulse steroid therapy followed by high-dose oral steroids and methotrexate along with supportive care and showed improvement in cutaneous and myopathic features.

Case Discussion

Dermatomyositis can have varied presentations ranging from amyopathic to severe muscle weakness, severe lung involvement, and cutaneous features. Myositis-specific antibodies are directed against antigens of protein synthesis pathways like Jo-1 and MDA-5. They have high specificity

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and a temporal link with malignancy, especially anti-TIF1-gamma (TIF1-γ) or anti-NXP2[1]. The typical cutaneous features of DM include Gottron's papule, Gottron's sign, heliotrope rash, shawl sign, holster sign, linear erythema, and mechanic hands, but all were absent in this patient. Mucinosis can be primary cutaneous or secondary, but the exact definitions and criteria are still lacking. Kauffman *et al*, 1998 reported similar atypical plaque-like mucinosis with dermatomyositis[3], Launay *et al* 2001 reported a case of scleromyxoedema associated with DM and treated with prednisone, azathioprine, and methotrexate. The muscular weakness and cutaneous features of DM improved, but not scleromyxoedema[4]. Perel-Winkler *et al* 2014 reported diffuse cutaneous mucinosis in a patient with DM and stated that secondary mucinosis is when mucin deposition is present in addition to some primary clinicopathological settings like connective tissue diseases, viral infections, and thyroid disorders[5]. Vysakha *et al* 2019 reported a case of a 38-year-old woman with myopathy and scleromyxoedema having characteristic mucin deposition and treated with intravenous immunoglobulin, oral prednisolone, and thalidomide[6]. The annular type of Lichen myxedematosus (LM) has also been reported in dermatomyositis[7]. However, in primary mucinosis, mucin deposition is the primary pathology. However, many extra-cutaneous features are well documented with primary mucinosis, like paraproteinaemia, multiple myeloma, dermato-neuro syndrome, dysphagia, cardiomyopathies, proximal myopathy, etc. So, it is difficult to ascertain the types, especially in the absence of standard classification or diagnostic criteria. The exact pathogenesis is not yet known, but it is postulated that cytokines like Interleukin-1, Interleukin-6, TNF-alpha, TGF-beta, and autoantibodies upregulate glucosaminoglycan synthesis from fibroblasts. Most of the earlier case reports did not specify myositis-specific antibodies, while recently, a case of Anti-MJ/NXP2 antibody-positive adult-onset dermatomyositis with LM and endometrial carcinoma which responded to resection of comorbid malignancy and prednisolone, was reported[8]. Although workup for malignancy was negative in our patient, but ovarian, gastric, endometrial, and nasopharyngeal carcinomas have been reported. Perel Winkler *et al* 2014 reported that around 30% (3 out of 12) DM with cutaneous mucinosis patients had associated

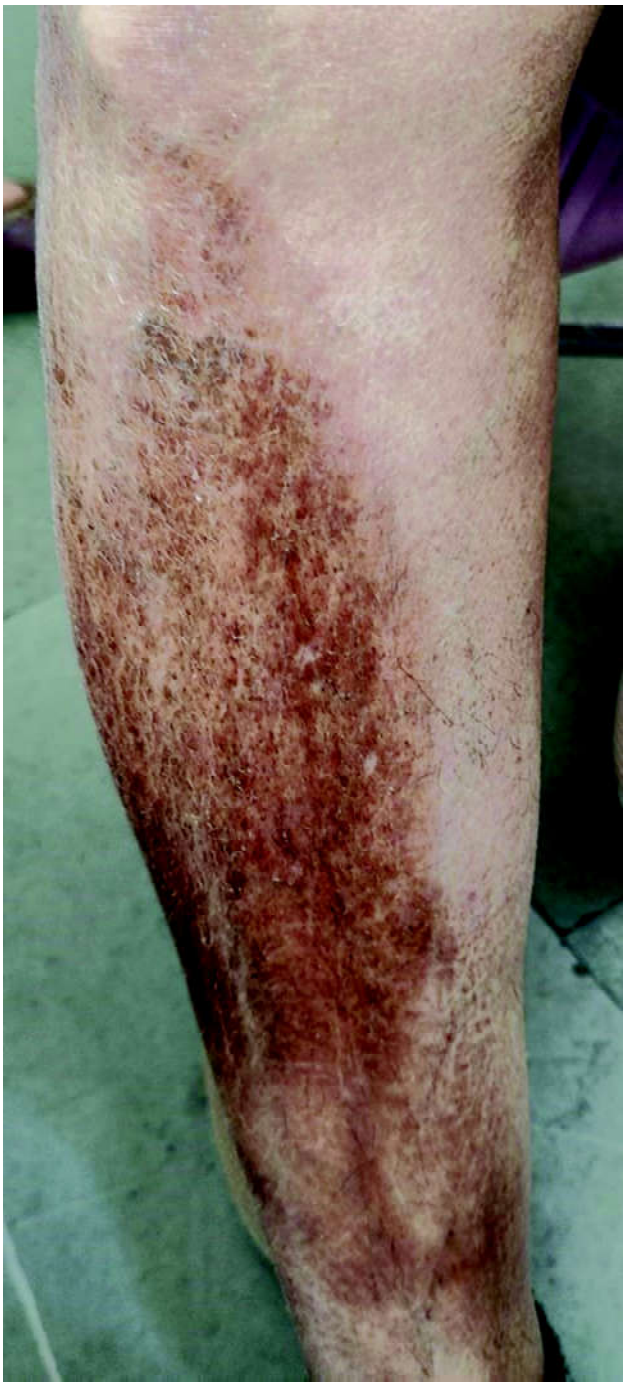

Fig. 1: *Scaly, waxy erythematous rash on the right leg.*

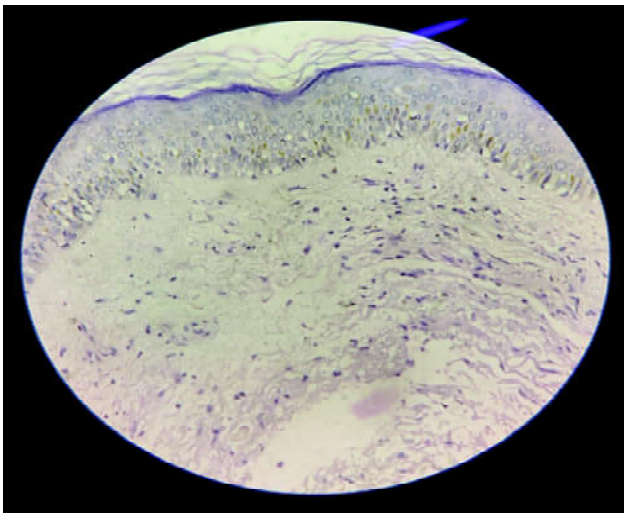

Fig. 2: *Perivascular lymphocytic infiltrate, interface dermatitis, and mucin deposition shown in skin biopsy.*

malignancy, which was similar to dermatomyositis alone[5]. LM is also reported in other connective tissue diseases like rheumatoid arthritis and mixed connective tissue disease[9]. Our patient responded well to high-dose steroid therapy along with a steroid-sparing agent; however, intravenous immunoglobulin is of some benefit in resistant cases. The presence of significant diffuse mucinosis in a setting of connective tissue disease can lead to atypical presentations and pose a diagnostic challenge. We need to evaluate the clinical relevance of mucinosis in connective tissue diseases, especially dermatomyositis. Primary and secondary mucinosis must be differentiated, as management, associations, and prognosis differ. Primary mucinosis can have systemic involvement and is also associated with paraproteinaemia and malignancy. A strict close follow-up is warranted because of the temporal association with malignancy. The systemic features do respond early with partial or complete resolution of cutaneous features. We need more studies and data to understand the spectrum of this disease and formulate standard nomenclature and classification criteria.

Conclusion

Dermatomyositis (DM) can present with atypical features of significant cutaneous mucinosis, making diagnosis and management challenging. A multidisciplinary approach is required to differentiate primary and secondary mucinosis because of distinct prognostic and therapeutic implications. Limited data is available for mucinosis and myositis-specific antibodies, like the Anti-Jo-1 antibody. Close follow-up is required to monitor disease progression and assess the risk of malignancy. Further studies are required to refine diagnostic criteria and management guidelines for mucinosis in connective tissue diseases.

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