SKIN

BRIEF ARTICLE

Seeing Double: Two Cases of Dermatomyositis Misidentified as Erythema Dyschromicum Perstans

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ABSTRACT

Without pathognomonic skin lesions or muscle weakness, early DM may be misdiagnosed as a disorder of hyperpigmentation. A 38-year-old male presented with a one-year history of progressive hyperpigmented patches on the face and neck. Outside biopsy was thought to be consistent with EDP, with which the patient's identical twin brother had recently been diagnosed. Later skin exam was notable for progression to erythematous and hyperpigmented slightly scaly macules and patches of the face, faint pink thin plaques on bilateral extensor forearms and bilateral retroauricular creases, and proximal nailfold capillary dilation and dropout. These findings raised concern for dermatomyositis. Repeat biopsies were suggestive of CTD. ANA was weakly positive. Extended myositis panel was negative. Re-evaluation of his twin brother's biopsy showed similar findings. The twin's ANA was negative, and extended myositis panel is pending. Ultimately, both were diagnosed with DM. Work-up for muscle and systemic involvement is underway. Our patient was started on MMF 500mg BID, which was increased to 1000mg BID at follow up. After little improvement on MTX, the patient's twin brother was switched to MMF 1000mg BID. Diagnosis of DM is particularly challenging in patients lacking classic dermatologic findings or myopathy. Recognition of subtle signs of DM, histopathologic analysis, and presence of MSAs may aid in correct diagnosis.

INTRODUCTION

Dermatomyositis (DM) is an idiopathic inflammatory myopathy that is classically identified by characteristic violaceous macules, papules, plaques on the eyelids (heliotrope rash), extremities (Gottron sign), and hands (Gottron papules), scalp involvement and proximal nail fold changes. However, DM is associated with a wide range of cutaneous findings that may vary as the disease progresses.[1] Amyopathic DM is also common. In the absence of pathognomonic skin lesions or muscle weakness, early DM may be misdiagnosed as a disorder of hyperpigmentation.[2]

CASE REPORT

A 38-year-old male with history of asthma and atrial fibrillation status-post ablation presented with a one-year history of

progressive hyperpigmented patches on the face and neck. Outside biopsy of the preauricular cheek and postauricular area revealed mild degeneration of the basal layer and superficial perivascular inflammation with lymphocytes, histiocytes, and melanophages. These findings were thought to be consistent with erythema dyschromicum perstans (EDP), with which the patient's identical twin brother had recently been diagnosed. He presented to our office with rash progression on the extensor arms, posterior auricular region, and scalp (**Figure 1**). His twin brother's facial skin lesions had also recently enlarged, and he was prescribed methotrexate (MTX) (**Figure 2a/b**). Our patient was otherwise healthy. He reported that the skin lesions were occasionally itchy and endorsed back pain for the past month, but denied fever, chills, or lymphadenopathy.

Skin exam was notable for progression to erythematous and hyperpigmented slightly scaly macules and patches of the face, particularly on the forehead and beard area, and neck. Faint pink thin plaques were visible on the bilateral extensor forearms and bilateral retroauricular creases. The proximal nailfold was notable for capillary dilation and dropout. This clinical presentation and outside biopsy showing vacuolar interface dermatitis raised concern for dermatomyositis.

Repeat biopsies taken from the pre- and post-auricular areas showed interface dermatitis with perivascular and periadnexal inflammation suggestive of connective tissue disease (CTD) (**Figure 3a/b**). DCAS was negative for fungal organisms and *T. pallidum* staining was negative. ANA was weakly positive (1:80) with a negative ENA antibody panel. Extended myositis panel was negative.

Re-evaluation of his twin brother's skin biopsy showed similar findings. The twin's ANA was negative, and extended myositis panel is pending. Ultimately, both were diagnosed with DM. Work-up for muscle and systemic involvement, including lung disease, is underway. Our patient was prescribed topical mometasone 0.1% and started on mycophenolate mofetil (MMF) 500 mg BID, which was subsequently increased to 1000 mg BID at follow up. After experiencing little improvement on Methotrexate, the patient's twin brother was switched to mycophenolate mofetil (MMF) 1000 mg BID.

DISCUSSION

DM is a heterogenous disorder that can present with a variety of skin and systemic manifestations, making it difficult to diagnose at times. Diagnosis is particularly challenging in patients lacking classic dermatologic findings or myopathy.[1] DM has a strong genetic component and multiple environmental risk factors, such as UV radiation, viral infections, and medications, which may act as triggers in genetically susceptible patients.[1] Immune activation, debated to be either antibody-dependent or secondary to activation of the classical complement cascade, also plays a central role in pathogenesis.[3] DM has a bimodal onset: between 5-15 years, and between 40-60 years, with a female to male ratio of 2:1.[4] The potential for other systemic involvement, including pulmonary, cardiac, gastrointestinal, endocrine, and vascular, should not be overlooked. Importantly, the estimated prevalence of malignancy in adult patients with DM is approximately 20% and is highest in those who are either anti-transcription intermediary factor 1– or anti-nuclear matrix protein 2 –antibody positive.[2]

The amyopathic dermatomyositis (ADM) subtype (previously, *dermatomyositis sine myositis*) involves hallmark cutaneous findings of DM in the absence of clinical or laboratory evidence of muscle disease for ≥ 6 months, whereas the hypomyopathic subtype lacks clinical muscle weakness despite evidence of muscle disease upon laboratory investigation.[5] These subtypes account for at least 20% of all DM cases.[6] Diagnostic and classification criteria for these skin-predominant forms of DM are less well characterized, leading to increased risk for erroneous diagnosis. The only validated criteria for ADM in existence require two out of the three pathognomonic cutaneous DM features (Gottron's sign, Gottron's papules, and heliotrope rash) and absence of muscle involvement. However, the use of this criteria may result in as many as 25% of patients with ADM being overlooked.[5]

Dermatological mimickers of DM exist, with the most common being cutaneous lupus erythematosus (CLE). In fact, one retrospective study revealed that 37.2% of patients with DM were previously diagnosed with either CLE or SLE.[7] Hyperpigmented patches on the head and neck in evolving DM can also mimic EDP both clinically and histologically. EDP is a disorder of pigmentation that is characterized by gray ("ashen"), or blue-brown macules or patches distributed over the trunk, limbs, neck, and face. On biopsy, degeneration of the basal cell layer with dermal melanosis and perivascular infiltrate is observed.[8]

The varying cutaneous manifestations of DM are traditionally divided into "pathognomonic", "characteristic", "compatible", "less common" and "nonspecific" categories.[1,4] Without classic skin lesions, DM can be diagnosed using skin biopsy showing characteristic findings of epidermal atrophy, dermal edema, basement membrane thickening, vacuolar interface dermatitis, a lymphocytic perivascular infiltrate, and pigment incontinence.[1] Other characteristic features of dermatomyositis that may aid in differentiation from other pigmentary disorders include dilated capillary loops at the base of the fingernails, irregular and thickened cuticles, and cracked palmar fingertips ("mechanic's hands").[3]

Myositis-specific antibodies (MSAs) are antibodies associated exclusively with the idiopathic inflammatory myopathies. Anti-MDA5 and anti-ARS antibodies are associated with interstitial lung disease, while anti-TIF1-gamma and anti-NXP2 antibodies portend an elevated risk of malignancy. Severe cutaneous manifestations are observed in patients with anti-SAE antibodies.[9,10] In the absence of vasculopathic and calcinotic lesions, such as in our patient, the first-line systemic therapies are mycophenolate mofetil (MMF) and methotrexate (MTX).[2]

The clinical heterogeneity associated with DM may present a significant challenge to diagnosis. Varying diagnostic and classification criteria may not adequately account for those patients presenting without classic cutaneous findings and myopathy. Recognition of early subtle signs of DM, histopathologic analysis, and presence of MSAs may aid in the correct diagnosis.

Conflict of Interest Disclosures: None

Funding: None

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Figure 1. First twin's rash on posterior auricular region and scalp.

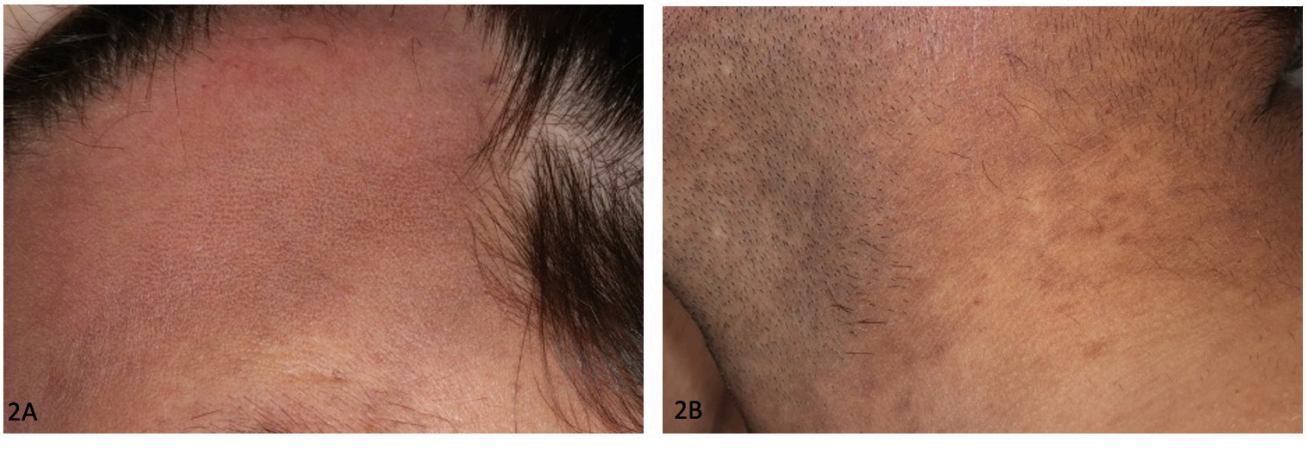

Figure 2A and 2B. Second twin's rash on face.

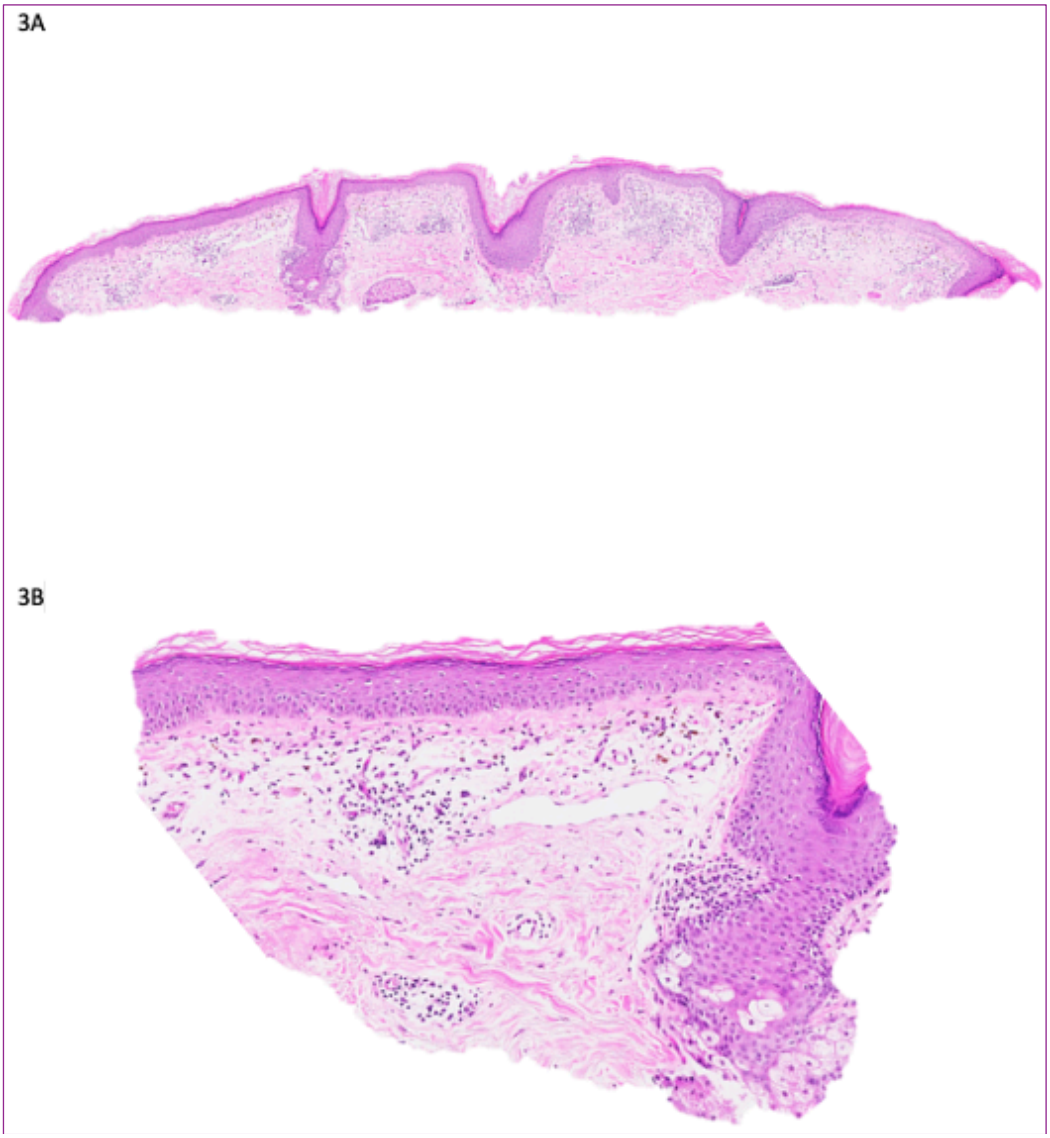

Figure 3A and 3B. Repeat biopsies taken from the pre- and post-auricular areas showed interface dermatitis with perivascular and periadnexal inflammation.