

Case Report

A report of seronegative clinically amyopathic dermatomyositis

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ABSTRACT

Clinical amyopathic dermatomyositis is a rare idiopathic inflammatory disease within the spectrum of dermatomyositis. This disease entity presents with similar cutaneous findings as dermatomyositis, but the clinical manifestation of myositis is absent. Given the increased risk also associated with clinical amyopathic dermatomyositis, a high index of suspicion is imperative to minimize rheumatologic and dermatologic misdiagnoses. Here we report a case of seronegative CADM in a young female athlete.

Keywords: Dermatomyositis, Amyopathic Dermatomyositis, Autoimmune, Inflammation

INTRODUCTION

Clinical amyopathic dermatomyositis (CADM) is a rare idiopathic inflammatory disease within the spectrum of dermatomyositis.¹ The estimated incidence of CADM is two per one million persons and may represent up to 20% of all cases of dermatomyositis.² The current theory for the pathogenesis of dermatomyositis includes the involvement of autoantibodies directed against endomysial and skin microvasculature.³ The presence of antibody-antigen complex will trigger and activate the complement cascade resulting in lysis of capillaries and cause hypoperfusion, microinfarction and necrosis of the affected tissues.³ CADM typically presents with cutaneous lesions, including a Gottron's papules, a malar rash, a heliotropic (violaceous periorbital) rash, and a shawl rash.⁴ While similar to classic dermatomyositis, the clinical manifestation of myositis in CADM is absent which is characterized by the lack of proximal and girdle

muscle weakness, elevated muscle enzymes and abnormal electromyography.⁵ However, when compared to classical dermatomyositis, CADM presents in similar demographic of patients, has identical dermatological findings and shares an increased risk of developing interstitial lung disease and malignancies.¹ As a result, given the similarities in its presentation and associated comorbidities, a high index of suspicion is imperative to minimize dermatologic and rheumatologic misdiagnosis of CADM (Table 1).⁶ Diagnosis of CADM is typically achieved clinically with a thorough history and physical exam. In addition, serology and tissue biopsy can further be of assistance to guide and confirm the diagnosis. Here we report a case of seronegative CADM in a young athlete.

CASE REPORT

A 27-year-old healthy female student athlete presented to rheumatology clinic due to brown discoloration on the

large knuckles of both her hands, a pink, butterfly like rash on her face and disproportionate shortness of breath with physical activity. She had no obvious muscular weakness, dysphagia or difficulty getting in or out of a chair. Her cutaneous symptoms presented approximately four weeks after a competitive injury that resulted in a pelvic avulsion fracture. At the time of presentation, she was taking non-steroidal anti-inflammatory drugs (NSAIDS) for her injury. There was no history of any other medication use. Her past medical history was significant for Raynaud's syndrome. Physical examination of both her hands revealed popular erythematous chocolate brown lesions on the dorsal aspect of the metacarpophalangeal joint's characteristic for Gottron's papules (Figure 1). Examination of her face revealed a mild erythematous malar rash that was continuous with the nasal labial fold (Figure 2). Reviews of systems was significant for dry mouth, photosensitivity in her hands and disproportionate shortness of breath with physical activity. Radiographic series of the chest and hands were negative for any pathological findings. Cardiac evaluation including an echocardiogram was unremarkable. Serological analysis did not reveal elevated creatine kinase, aldolase or complement levels. The antinuclear antibody (ANA), rheumatoid factor, and myositis specific auto-antibodies including anti-JO-1, anti-PL7, anti-PL12, anti-EJ, anti-SRP, anti-MI-2, anti-MDA-5, anti-p155 and anti-NXP-2 were all negative. Only the C-reactive protein (CRP) was elevated at 15.5 mg/L (nl <10 mg/L). The patient refused a skin biopsy and was clinically diagnosed with CADM. She was treated with a course of Prednisone which helped to clear her cutaneous symptoms and she refused to be placed on methotrexate for steroid sparing therapy. She was able to successfully wean off the corticosteroids within one month of initiation without recurrence of symptoms.



Figure 1: 27 years old healthy female presents with chocolate brown discoloration on the metacarpal bones of her hands.



Figure 2: A pink, butterfly like erythematous rash across the cheeks and bridge of the nose.

DISCUSSION

CADM remains a difficult etiology to diagnose early given its various cutaneous presentations, absence of muscle involvement and plethora of autoantibodies that may be involved in the pathogenesis. Therefore, an astute physician should have CADM high in their differential diagnoses in order to minimize dermatologic and rheumatologic misdiagnosis of CADM mentioned in Table 1. This is particularly important given that the treatment options amongst these diseases can be vastly different and result in mismanagement of the patient. In this report, our patient only presented with cutaneous lesions, specifically, Gottron's papules and a malar rash. There was no clinical manifestation of myopathy highlighted by the absent of proximal muscle weakness and normal muscle enzymes.

When evaluating for specific autoantibodies, it is important to recognize the plethora of autoantibodies associated in the spectrum of dermatomyositis including but not limited to, anti-HMG- CoA, anti-JO-1, anti-PL7, anti-PL12, anti-EJ anti-MDA5, anti-Mi2, anti-NXP2, anti-P155, anti-Pm/Scl, anti-SAE, anti-SRP, and anti-synthetases.⁷ More specific autoantibodies with respect to CADM include anti-CADM-140 autoantibodies that target melanoma differentiation-associated gene 5 (MDA5) and anti-P155 autoantibodies that target transcription intermediary factor 1-c (TIF1-c) protein.⁷ However, autoimmune panel analysis was negative in this report which highlights the challenges of using serology to help assist in diagnosis of CADM. The only additional diagnostic clue was the presence of disproportionate episodes of shortness of breath with exercise which may highlight the involvement of the lungs typically seen in dermatomyositis.

Table 1: Differential diagnoses of clinically amyopathic dermatomyositis may include.⁶

S. No	Diseases
1.	Systemic lupus erythematosus
2.	Psoriasis
3.	Mixed connective tissue disease
4.	Contact dermatitis
5.	Atopic dermatitis
6.	Lichen planus
7.	Polymorphous light eruption
8.	Seborrheic dermatitis
9.	Trichinosis
10.	Acute HIV infection

Typically, most patients with CADM may develop mild, chronic, and non-progressive lung disease; however, small subset of these cohort of patients can progress to severe rapidly progressive interstitial lung disease.⁸ In a Japanese cohort of CADM patients with rapidly progressive ILD, an anti-MDA-5 autoantibody was identified in 19-35%, however anti-MDA-5 was not present in our patient.⁹ Nonetheless, appropriate surveillance with multiple assessments, not limited to pulmonary function tests, are essential to assess for disease progression and ensure that individuals are offered appropriate interventions. Lastly, it is estimated that patients with CADM have a 14-28% increased risk of developing malignancy.⁸ It has been suggested that anti-TIF1-gamma (TIF1- γ) or anti-NXP2 autoantibodies are associated with increased risk of malignancy.⁷ Therefore, in addition to age appropriate cancer screening protocols, all newly diagnosed CADM patients require comprehensive screening evaluation for malignancy. In men, associated cancers typically include nasopharyngeal, bladder, colorectal, lung, and prostate cancers whereas in women CADM is commonly associated with breast, ovary, cervix, and uterine cancers.¹⁰ As a result, initial malignancy evaluation should include a rectal exam, and recommendations for chest radiography, ultrasound or computed tomography (CT) of the abdomen and pelvis, complete blood count, comprehensive metabolic panel, stool guaiac, urinary analysis and cytology.¹⁰ Upon identification of abnormal findings, further and more aggressive diagnostic interventions may be necessary to discover the underlying etiology.

CONCLUSION

Clinical amyopathic dermatomyositis remains an elusive etiology to diagnose quickly and accurately. Given the

increased risk associated with the development of malignancy, a high index of suspicion and thorough comprehensive screening evaluation is imperative in the management of these patients.

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REFERENCES

1. Ghazi E, Sontheimer RD, Werth VP. The importance of including amyopathic dermatomyositis in the idiopathic inflammatory myositis spectrum. *Clin Exp Rheumatol.* 2013;31:128.
2. Reeder MJ, Wetter DA, Li X, Davis M. Incidence of dermatomyositis and clinically amyopathic dermatomyositis: a population-based study in Olmsted county, Minnesota. *Dermatol Clin.* 2010;146(1):26-30.
3. Dalakas MC. The molecular and cellular pathology of inflammatory muscle diseases. *Curr Opin Pharmacol.* 2001;1:300-6.
4. Callen JP. Cutaneous manifestations of dermatomyositis and their management. *Curr Rheumatol Rep.* 2010;12:192-7.
5. Sontheimer RD. Cutaneous features of classic dermatomyositis and amyopathic dermatomyositis. *Curr Opin Rheumatol.* 1999;11:475-82.
6. Koler RA, Montemarano A. Dermatomyositis. *American Family Physician.* 2001;64(9):1565.
7. Cassius C, Le Buanec H, Bouaziz JD, Amode R. Biomarkers in adult dermatomyositis: tools to help the diagnosis and predict the clinical outcome. *J Immunol res.* 2019;2019.
8. Udkoff J, Cohen PR. Amyopathic dermatomyositis: a concise review of clinical manifestations and associated malignancies. *Am J Clin Dermatol.* 2016;17(5):509-18.
9. Sato S, Hirakata M, Kuwana M, Suwa A, Inada S, Mimori T et al. Autoantibodies to a 140-kd polypeptide, CADM-140, in Japanese patients with clinically amyopathic dermatomyositis. *Arthritis Rheumatism.* 2005;52(5):1571-6.
10. Ungprasert P, Bethina NK, Jones CH. Malignancy and idiopathic inflammatory myopathies. *North Am J Med Sci.* 2013;5(10):569.

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