CLINICAL VIGNETTE

A Rare Patient with Amyotrophic Dermatomyositis

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A 48-year-old male with no significant past medical history presented to dermatology after the gradual appearance of an erythematous rash over his elbow and knees, which progressed to his abdomen, back and thighs, over years. He was initially diagnosed with psoriasis and given topical corticosteroids which he intermittently used for a year, without significant improvement. The dermatologist took biopsies from his elbows, which showed nonspecific dermatitis. His rash progressed to include his eyelids, extensive erythema over his elbows, triceps region, and finally his hands. He noted onset of small white papules over his knuckles. He did not have the "shawl sign" or evidence of a photosensitivity rash. He obtained a second opinion from a dermatologist at an academic medical center, who also biopsied his rash and diagnosed him with dermatomyositis. By the time he presented for a dermatology second opinion, he developed an extensor rash and Gottron's papules that are pathognomonic for dermatomyositis. He had Raynauds syndrome, but no joint swelling/tenderness, or muscle weakness/myalgia.

Labs including CBC, CMP, and UA were normal. His CK was mildly elevated at 277(ULN 204), units/L with normal aldolase at 7.3 (ULN 7.6) units/L. His ESR was 1 ml/hr. Jo1, MI2 antibodies, quantitative immunoglobulins, and SPEP/UPEP were normal.

He subsequently saw a rheumatologist who scheduled extensive testing for malignancy. This included CT chest, abdomen and pelvis, colonoscopy, octreoscan, and a full body PET scan without any positive findings. The rheumatologist started methotrexate and hydroxychloroquine, with improvement in his symptoms. Annual chest CT's and PFT's were followed, but never identified any airway or interstitial lung disease. An MRI of his face and orbits as well as a transthoracic echocardiogram were also normal. His creatinine kinase was in the high normal range, and MRI of his extremities failed to identify any inflammatory changes. He never developed myalgias or muscle weakness, but his rash returned when he briefly stopped methotrexate due to financial reasons.

Dermatomyositis is a rare condition affecting 9.6 per million population. Approximately 20% present as clinically amyotrophic dermatomyositis (ADM) present for six months or more, without proximal muscle weakness. However, some patients may have mildly elevated muscle enzymes.[1] Complications include a 5-fold higher risk of malignancy in the lung, pancreas, and gynecological organs. Patients with amyotrophic dermatomyositis are also prone to developing interstitial lung disease.[2-4] One 2006 multinational study reported a 13% risk of ILD and 14% risk of malignancy in ADM patients. Another 2009 US based study reported higher risk of malignancy risk of 28%.[1] Patients with ADM can be viewed as being on the spectrum of the same disease as patients with classic dermatomyositis, with similar skin pathology and autoantibodies, and risk for similar complications. Fortunately, our patient was followed for more than 10 years without complication.

Our patient was initially misdiagnosed and ADM frequently presents diagnostic challenges. Skin biopsies may demonstrate nonspecific dermatitis, and patients may lack the muscle symptoms seen in classic dermatomyositis. Clinically amyotrophic dermatomyositis can be separated into true amyotrophic dermatomyositis, (without clinical, lab, electrophysiologic, or imaging evidence of myopathy); hypomyopathic dermatomyositis (HDM),[5] with no clinical symptoms but with EMG or imaging findings of myopathy. A subset of patients may later develop myopathy after intervening periods of months to years. It is helpful to view all of these as a spectrum of the same disease[6] which generally presents with the pathognomonic signs of Gottron's papules, Gottron's sign (erythematous and violaceous papules over extensor surfaces) and characteristic heliotrope rash, shawl sign, and periungal telangiectasias. Both ADM and HDM often have lab or imaging evidence of subclinical myositis and a minority can later manifest clinical myositis even years later while on systemic treatment.[7] Recognition of ADM is important as patients may benefit from screening for malignancy[7,8] and associated lung diseases.

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