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Facial erythromelalgia: A rare entity to consider in the differential diagnosis of connective tissue diseases

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To the Editor: A 53-year-old man presented to an outside dermatologist with an erythematous facial eruption of 4 years' duration. The eruption was associated with burning and swelling, which the patient reported worsened with sun exposure. These symptoms were debilitating, preventing the patient from working and maintaining daily activities. Initially, the eruption was thought to be rosacea and actinic damage; however, it was unresponsive to standard therapies. Given the refractory nature of the facial erythema and reported photosensitivity, a diagnosis of connective tissue disease (CTD) was considered. Antinuclear, anti-Ro/SSA, and anti-La/SSB antibodies were negative; however, skin biopsy revealed vacuolar interface alteration with perivascular and periadnexal lymphoid infiltration and mucin deposition (Fig 1). A diagnosis of CTD was made, and treatment with hydroxychloroquine and systemic corticosteroids was initiated. However, the patient had minimal clinical response, resulting in subsequent referral to our CTD clinic for management. On our initial examination, the patient had prominent full facial and auricular erythema and edema, as well as erosions of the bilateral helices and a violaceous appearance of his earlobes (Fig 2, *A*). Despite the biopsy findings, clinical exam did not support a diagnosis of CTD. Instead, facial erythromelalgia was suspected. Upon further questioning, the patient reported skin burning not only with sun exposure, but also with heat exposure. Furthermore, he reported attempting cooling measures, such as the frequent use of ice packs, to relieve his symptoms. With these additional details, a clinical diagnosis of facial and auricular erythromelalgia was established. Further workup excluded potential causes of secondary erythromelalgia. Given that erythromelalgia is thought to involve both small fiber neuropathy as well as vasculopathy, we initiated therapy with aspirin, pentoxifylline, and gabapentin.[1] Subsequently, nifedipine was added to this treatment regimen. At 5 months' follow-

up, the patient reported 80% improvement (Fig 2, *B*) and was able to return to work and enjoy normal daily activities.

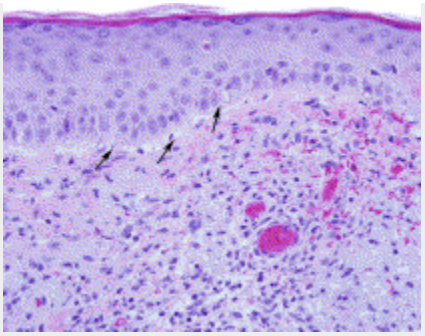

Fig 1

Facial erythromelalgia mimicking connective tissue disease. Biopsy showed vacuolar interface alteration (*arrows*), perivascular lymphocytic inflammation, and marked vascular congestion (*right*). Mucin deposition was also evident in the mid to deep dermis (not shown). (Hematoxylin and eosin; original magnification ×400.)

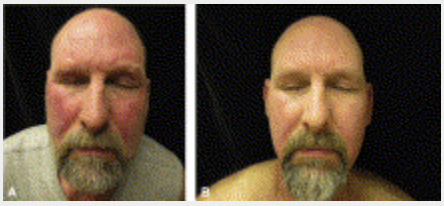

Fig 2

A, Facial erythromelalgia before therapy. There is prominent full facial erythema and edema, which may mimic the facial erythema seen in connective tissue diseases. **B,** Facial erythromelalgia after treatment. Improvement in erythema and edema 5 months following initiation of therapy with aspirin, pentoxifylline, gabapentin, and, subsequently, nifedipine.

Erythromelalgia is a rare condition characterized by episodic erythema, swelling, warmth, and burning, which can become persistent over time. Although it primarily occurs on the extremities, it may also manifest on the ears and face. Heat exposure triggers episodes, and cooling can alleviate symptoms.[2] Erythromelalgia can cause significant morbidity and mortality, with suicide being a notable cause of death, making timely recognition and initiation of therapy essential.[3] There is no diagnostic test for erythromelalgia; therefore, a high level of clinical suspicion, along with a detailed history and physical exam, is required to establish the diagnosis. Although biopsy findings are often nonspecific, there has been 1 reported case of histopathologic findings of CTD in a patient with erythromelalgia involving the hands.[4]

This case highlights a rare condition, which can be mistaken for CTD, particularly when the patient presents with facial erythema, reports photosensitivity, or has skin biopsy findings that support CTD. To our knowledge, we report the first case of facial erythromelalgia with biopsy findings demonstrating vacuolar interface dermatitis and mucin deposition. This case underscores the need for physicians who follow patients with CTD to be aware of the clinical presentation of facial erythromelalgia.

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