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Topical Sirolimus for Refractory Cutis Marmorata Telangiectatica Congenita in Adulthood: A Case Report

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ABSTRACT

Cutis marmorata telangiectatica congenita (CMTC) is a rare congenital vascular disorder characterized by persistent, violaceous, reticulated skin changes that may be complicated by painful ulcerations. Although localized disease often improves with age, generalized CMTC can persist into adulthood and may be associated with limb asymmetry, ocular abnormalities, and neurologic sequelae. Diagnosis can be challenging in atypical adult presentations despite established major and minor criteria. We report a case of a 50-year-old woman with congenital livedo reticularis and Raynaud syndrome who presented with lifelong unilateral left lower-extremity hypoplasia and fixed lacy violaceous patches. Over the preceding decade, she developed recurrent, spontaneous, painful ulcerations exacerbated by cold exposure. Examination revealed a hypoplastic left leg with reticulated violaceous patches and tender, crusted erosions without venectasia; biopsy findings ruled out vasculitis, and the overall clinicopathologic picture met all 3 major and multiple minor Kienast-Hoeger criteria for CMTC. Multiple therapies targeting vasospasm and microvascular flow (including sildenafil, pentoxifylline, diosmiplex, nifedipine, and aspirin) failed to improve symptoms. Initiation of once-daily topical sirolimus (1 mg/mL) resulted in marked pain reduction within 4 weeks and cessation of new ulcerations, with visible improvement and healed erosions by 2 months. This case supports topical sirolimus as a promising off-label option for adult CMTC with chronic ulcerative disease.

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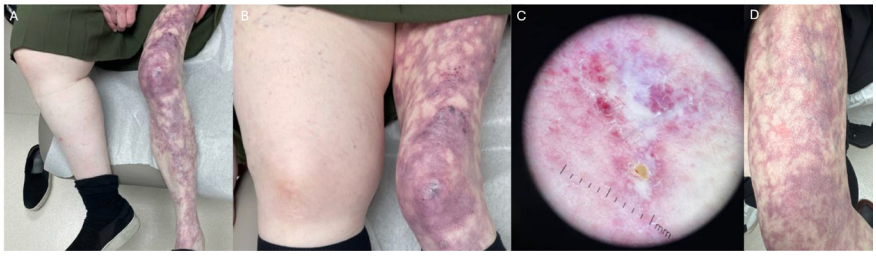
INTRODUCTION

Cutis marmorata telangiectatica congenita (CMTC) is a rare congenital vascular disorder characterized by violaceous, reticulated skin that may be complicated by painful ulcerations. Limited presentations often improve but rarely fully resolve with age, whereas generalized CMTC may persist into adulthood and is associated with limb asymmetry, glaucoma, and neurological deficits.[1] Kienast and Hoeger proposed widely accepted major and minor diagnostic criteria, yet diagnosis remains challenging with untraditional presentations and when symptoms progress beyond childhood.[2] Treatment is largely conservative, focused on avoiding cold exposure and trauma; therapies for extensive disease are limited to vasodilators, aspirin, and laser therapies. We present an unusual case of unilateral limb hypoplasia with vascular malformation, Raynaud syndrome, and livedo reticularis in an adult, ultimately diagnosed as CMTC and successfully treated with topical sirolimus.

A 50-year-old female with a history of congenital livedo reticularis and Raynaud syndrome presented with painful, violaceous, lacy patches on her left leg with associated limb hypoplasia present since birth. The erythema did not fade with warming, and she reported a 10-year history of spontaneous painful ulcerations of the affected leg that flared during cold weather. She denied a family history of similar presentations. She previously tried topical lidocaine for pain relief, but ultimately developed new erosions at application sites. Physical examination revealed a hypoplastic left leg from the upper thigh to the foot with lace-like violaceous patches and tender crusted erosions, without venectasia (Figure 1A-C). A skin biopsy was obtained from a crusted lesion on the thigh.

FIGURE 1. (A & B) Hypoplastic left leg with lace-like violaceous patches on initial evaluation. (C) Evaluation of crusted reticular lesion with dermoscopy. (D) Left leg after 2 months of treatment with topical sirolimus.



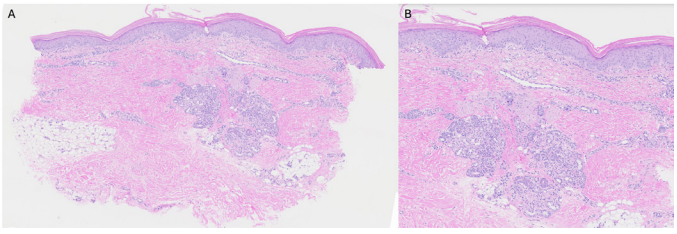
Histology showed variable epidermal acanthosis with dense orthokeratosis and focal parakeratosis. Within the dermis, extravasated erythrocytes, hemosiderin-laden macrophages, lobular superficial dermal neovascularization with dermal fibrosis, and a perivascular lymphohistiocytic infiltrate were noted (Figure 2). These clinical and histopathologic findings fulfilled all three major and multiple minor diagnostic criteria of Kienast and Hoeger, confirming generalized CMTC persisting into adulthood.[2]

Given the patient's history of Raynaud syndrome, sildenafil 20 mg daily was initiated, but quickly discontinued due to leg swelling and lack of pain relief. She was subsequently treated with pentoxifylline 400 mg daily to improve microvascular flow by reducing blood viscosity and increasing erythrocyte flexibility, and diosmiplex 630 mg daily for its anti-inflammatory, venotonic effects in chronic venous disease. She was concurrently treated with nifedipine for presumed vasospasm and aspirin for thrombosis prophylaxis, and anticoagulation was considered given the underlying vascular malformation.[3] Despite these targeted interventions, her pain and disease progression remained unchanged.

She then initiated therapy with once-daily topical 1 mg/mL sirolimus solution. Initial dryness was relieved with emollients. After four weeks, her pain had markedly improved, and the development of spontaneous ulcerations ceased. At her two-month follow-up visit, she reported substantial improvement in her quality of life. The affected leg appeared less violaceous, and previously present sores had healed with no new erosions (Figure 1D).

Sirolimus, an mTOR pathway inhibitor that slows cellular proliferation and abnormal vascular growth, has shown benefit in pediatric patients with vascular anomalies, including Klippel-Trenaunay syndrome[4] Limited case reports suggest that sirolimus, administered orally or topically, can reduce lymphatic leakage secondary to skin barrier disruption.[5] However, its use in CMTC, particularly in adults with chronic ulcerative disease, remains limited. In our patient, topical sirolimus was the only intervention associated with meaningful clinical improvement. This case highlights topical sirolimus as a promising off-label treatment for CMTC, particularly for reducing ulceration frequency and alleviating pain in patients who have not responded to traditional therapies. Effective topical therapy may allow clinicians to avoid systemic sirolimus with its greater adverse-effect burden and supports broader use of sirolimus-based approaches for vascular malformations beyond the pediatric population.

FIGURE 2. Hematoxylin and eosin staining of skin tissue biopsy demonstrating thick-walled capillary proliferation in the superficial dermis with foci of intercellular edema and parakeratosis at (A) 20x magnification and (B) 40x magnification.



DISCLOSURES

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