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Telangiectasia macularis eruptiva perstans associated with a sicca complex

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Abstract

We present the case of woman in her 50s who developed numerous red-brown telangiectatic macules on her trunk and extremities, as well as persistent dry eyes and dry mouth. Skin biopsy was consistent with telangiectasia macularis eruptiva perstans (TMEP). Serum tryptase was elevated suggesting systemic involvement. Anti-Ro and La were negative. ANA was positive. Salivary gland biopsy revealed a focus score of 3 and immunostains revealed infiltrates of aberrant CD117 positive mast cells. This case suggests a mechanistic role of mastocytosis in salivary compromise.

Keywords: mastocytosis, telangiectasia macularis eruptiva perstans (TMEP), sicca, Sjögren syndrome

Introduction

Mastocytosis is the clonal proliferation of aberrant mast cells into one or more organ systems. Its clinical presentation is heterogeneous, ranging from cutaneous mastocytosis (CM), defined as skin-limited disease, to systemic mastocytosis (SM), characterized by progressive mast cell infiltration of internal organs accompanied by mast cell degranulation symptoms, organomegaly, organ impairment, or loss of function. Although SM frequently affects the lymph nodes, spleen, liver, gastrointestinal tract, bones, and bone marrow, mast cell infiltration of the exocrine glands is rare [1].

Case Synopsis

A 54-year-old woman with osteoporosis and gastroesophageal reflux disease developed a

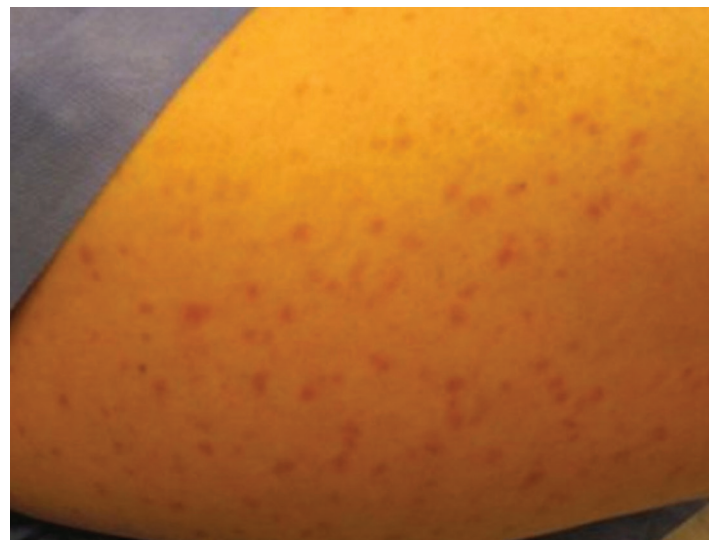


Figure 1. *Telangiectasia macularis eruptiva perstans.* Patient on the day of presentation with 3-4mm red-brown macules on the thighs.

persistent non-pruritic rash. Lesions emerged on the trunk and then spread rapidly to the bilateral upper and lower extremities. She subsequently developed dry eyes and dry mouth. On review of systems, the patient endorsed fatigue. Physical exam revealed hundreds of red-brown telangiectatic macules. Darier sign (urtication with stroking of the skin) was negative (**Figure 1**). There was no palpable hepatosplenomegaly.

Skin punch biopsy revealed superficial perivascular and interstitial spindle-shaped mast cells (**Figure 2, top**) highlighted by CD117 immunostain (**Figure 2, bottom left**). These findings were consistent with cutaneous mastocytosis (CM) – specifically TMEP. Cutaneous mast cells were positive for CD25 (**Figure 2, bottom right**). Serum tryptase level was elevated at 28 ng/mL (reference range: <11.5 ng/mL).

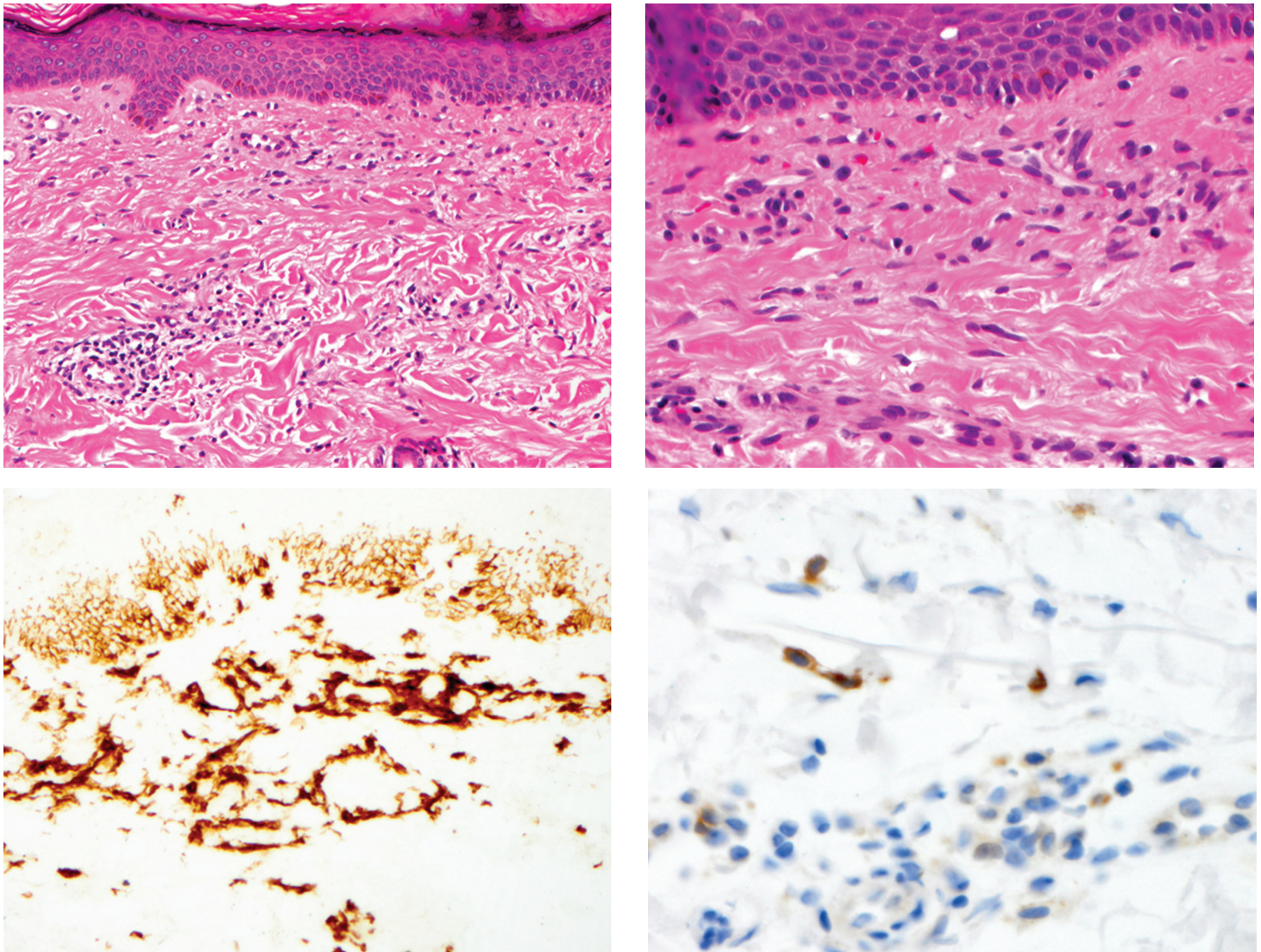


Figure 2. Skin punch biopsy specimen. Histopathology reveals a superficial perivascular and interstitial dermal spindle-cell infiltrate (H&E; A, 200%; B, 400%); highlighted by CD117 immunohistochemistry (C, 400%) consistent with cutaneous mastocytosis; a subset of mast cells was also immunoreactive for CD25 (D, 600%).

The patient endorsed daily persistent dry eyes and dry mouth for a period of greater than 3 months. Anti-SSA (Ro) and anti-SSB (La) were negative. Anti-nuclear antibody was positive with a titer of 1:2569. Rheumatoid factor and anti-double stranded DNA were negative. Complement C3 and C4 were within normal limits. Lower lip salivary gland (LSG) biopsy revealed a focus score of 3 (defined as a number of lymphocytic foci with an aggregate of ≥ 50 lymphocytes). Immunostains of the LSG biopsy revealed infiltrates of abnormal appearing CD117 positive mast cells (**Figure 3**).

Case Discussion

We describe an unusual case of patient with TMEP with evidence of indolent systemic involvement and a clinical sicca complex. TMEP is an unusual variant of

CM. It is a well-established entity in the dermatologic literature with descriptions dating back to 1930. Lesions are characterized by telangiectatic macules on a red to brown background. Darier sign is typically negative. The histology of TMEP demonstrates a perivascular infiltrate of mast cells on skin biopsy [2]. TMEP is rarely associated with systemic involvement.

The World Health Organization (WHO) has established diagnostic criteria for SM, requiring the presence of one major and one minor criterion or three minor criteria [1]. The major criterion of multifocal dense mast cell infiltrates in sections of bone marrow could not be evaluated in our patient as she declined the procedure. However, the persistently elevated serum tryptase levels (meeting one WHO minor criterion), history of osteoporosis and fatigue were suggestive

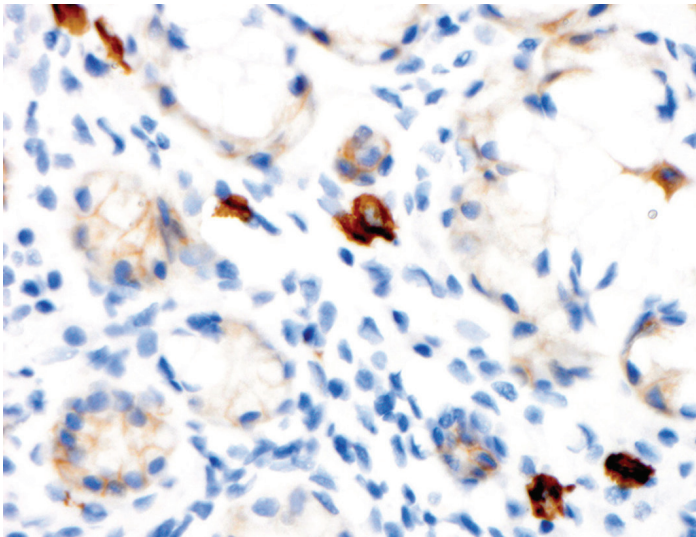


Figure 3. Lower lip salivary gland biopsy. Minor salivary gland tissue demonstrating CD117 positive stromal mast cells by immunohistochemistry, 600%.

of systemic involvement. In addition, skin biopsy revealed cutaneous mast cells positive for CD25 – a finding predictive of systemic mastocytosis (SM), [3]. Furthermore, recent studies demonstrate that adult-onset CM is highly suggestive of SM, especially in the setting of elevated tryptase levels [4]. Additional studies show that almost all patients with adult onset CM, in fact have SM with cutaneous involvement [5].

We considered a diagnosis of primary Sjögren Syndrome (pSS) in our patient. However anti-Ro and anti-La were negative. Despite LSG biopsy focus score of 3, there was higher clinical suspicion for a secondary cause of the patient's sicca symptoms. The aggregates of mast cells noted on LSG histopathology were unusual. Mast cells are typically sparse and sporadic in the histology of normal salivary glands and in pSS [6,7]. In the setting of biopsy-proven TMEP and elevated serum tryptase levels, this case suggests a mechanistic role of mastocytosis; it is plausible that aberrant mast cell infiltration of the salivary glands resulted in glandular compromise producing a clinical sicca complex. A similar mechanism has been reported only once in the literature. Bac and Kooy described the case of a patient presenting with urticaria pigmentosa, another form of cutaneous mastocytosis, with systemic involvement and sicca symptoms. Histopathologic findings on LSG biopsy were similar to the present case, revealing an abnormal mast cell infiltrate [7].

Alternatively the findings in our case may represent concomitant pSS and TMEP. This association has been reported once in the literature by Erbay et al. However their patient exhibited more conclusive evidence of pSS with positive Ro antibodies. LSG histology in that case revealed an increased number of tryptase positive mast cells similar to the findings we report [8]. It is thought that mast cell abnormalities and autoimmunity may be linked as has been well established in rheumatoid arthritis [9].

Conclusion

Differentiating between pSS and other potential etiologies of sicca symptoms is challenging and requires comprehensive investigation including clinical evaluation, laboratory studies, and histopathology [10]. Clinicians should pay close attention to any extra-glandular signs and symptoms, as these may guide additional diagnostic work up.

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