Mastocytosis With Associated Essential Thrombocythemia☆

Mastocitosis asociada a trombocitemia esencial

To the Editor:

Maculopapular cutaneous mastocytosis, previously known as urticaria pigmentosa, is the most common cutaneous expression of indolent systemic mastocytosis (SM) in adults. This condition usually does not require specific treatment.[1] However, SM can be associated with other hematologic malignancies.[2] We present the case of a patient with indolent SM who developed essential thrombocythemia during clinical follow-up.

A 70-year-old woman with no relevant past history except dyslipidemia was referred to our dermatology department in 2005 for assessment of brownish papules measuring 3-5 mm in diameter on the limbs and upper chest that had appeared gradually over the previous 5 years (Fig. 1). On examination, the Darier sign was observed. The patient reported neither pruritus nor symptoms associated with mast cell degranulation. Skin biopsy revealed mast cell proliferation in the upper dermis. Serial bone radiography and abdominal ultrasound were normal. Complete blood count and serum biochemistry were normal except for tryptase levels (124 μg/L; normal < 11.4 μg/L). The patient was referred to the hematology department and underwent bone marrow aspiration, which revealed the presence of atypical mast cells consistent with a diagnosis of SM (flow cytometry detected 0.2% mast cells, 100% with pathologic phenotype CD2+/CD25+). Polymerase chain reaction detected the presence of the c-kit D816V mutation. With a diagnosis of indolent SM, the patient was once again referred to the dermatology department without treatment. The platelet count increased gradually from 2010 to 2013 until it reached 741×10^9/L, at which point the patient was once again referred to the hematology department for assessment of thrombocytosis. The patient was diagnosed with mastocytosis associated with essential thrombocythemia–type myeloproliferative neoplasm. Treatment was initiated with oral hydroxyurea (500 mg, 3 d/wk) and oral clopidogrel (75 mg, every 48 h). The patient has not developed any symptoms associated with mast cell degranulation. The lesions on the arms and upper chest have gradually resolved and only a few lesions persist on the thighs. Platelet count has remained below 700×10^9/L and serum tryptase levels remain at around 100 μg/L.

SM is classified as indolent SM, smoldering SM, aggressive SM, SM with associated clonal hematologic non-mast-cell lineage disease, mast cell leukemia, and mast cell sarcoma.[3] Indolent SM is the most common form, but it does not usually cause symptoms derived from mast cell infiltration and does not require cytoreductive therapy.[1] The second most common form is SM associated with hematologic disease, which accounts for between 21% and 44% of SM cases.[2] The hematologic disorders most frequently associated with SM are myeloproliferative syndromes (45% of cases), chronic myelomonocytic leukemia (29%), myelodysplastic syndromes (23%), and acute leukemia (3%).[2] Essential thrombocythemia is classified as a myeloproliferative syndrome. The association of SM with essential thrombocythemia is very rare. In a series of 123 patients with SM associated with hematologic disease, essential thrombocythemia accounted for only 6 cases.[2] Besides these 6 patients, the reviewed literature only contains a few isolated case reports describing the association between maculopapular mastocytosis and essential thrombocythemia.[4–10]

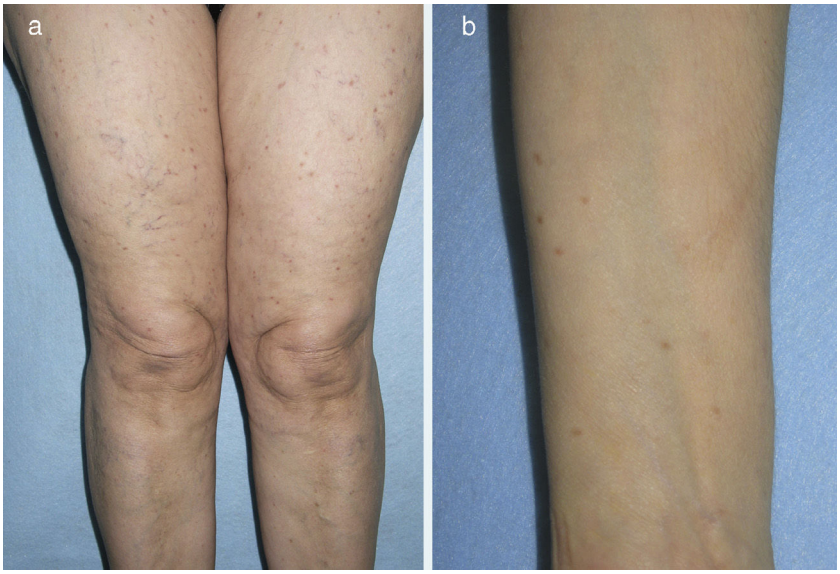

Figure 1 Clinical appearance of maculopapular mastocytosis lesions on the thighs (A) and forearm (B).

☆ Please cite this article as: Marcoval J. Mastocitosis asociada a trombocitemia esencial. Actas Dermosifiliogr. 2019;110:63–64.

Although the pathophysiologic relationship between mast cell proliferation and associated hematologic malignancies is not well established, some lineage distribution studies of the c-kit D816 V mutation suggest the existence of a common pluripotent hematopoietic stem cell in most patients.[2] In some cases, however, it is possible that the 2 clonal hematologic disorders may develop coincidentally in the same patient.[2]

When we talk about SM, we tend to think of asymptomatic mast cell infiltration in bone marrow or the rare aggressive forms of mastocytosis. However, we must also be aware of the possibility of association with other hematologic malignancies, as the case of essential thrombocythemia in our patient illustrates. The prognosis of indolent mastocytosis associated with clonal hematologic non-mast-cell lineage disease depends on the type of associated hematologic disorder. Cutaneous manifestations of mastocytosis can therefore be considered possible markers of a more severe associated hematologic disorder that may require specific treatment.

All adult patients with mastocytosis should undergo a bone marrow study that includes mutational analysis and mast cell immunophenotyping. These patients require clinical follow-up not only because of the risk of developing aggressive forms of SM but also because of the risk of associated hematologic malignancies.

Conflicts of Interest

The authors declare that they have no conflicts of interest.

References

1. Azaña JM, Torrelo A, Matito A. Actualización en mastocitosis. Parte 2: categorías, pronóstico y tratamiento. Actas Dermosifiliogr. 2016;107:15–22.
2. Pardanani A, Lim KH, Lasho TL, Finke C, McClure RF, Li CY, et al. Prognostically relevant breakdown of 123 patients with systemic mastocytosis associated with other myeloid malignancies. Blood. 2009;114:3769–72.
3. Arber DA, Orazi A, Hasserjian R, Thiele J, Borowitz MJ, Le Beau MM, et al. The 2016 revision to the World Health Organization classification of myeloid neoplasms and acute leukemia. Blood. 2016;127:2391–405.
4. Dobrea C, Ciochinaru M, Găman A, Dănăilă E, Coriu D. Systemic mastocytosis associated with essential thrombocythemia. Rom J Morphol Embryol. 2012;53:197–202.
5. Spivacow FR, Sarli M, Nakutny R. Systemic mastocytosis: Bone impact. Medicina (B Aires). 2012;72:201–6.
6. Martin LK, Romanelli P, Ahn YS, Kirsner RS. Telangiectasia macularis eruptiva perstans with an associated myeloproliferative disorder. Int J Dermatol. 2004;43:922–4.
7. Lappe U, Aumann V, Mittler U, Gollnick H. Familial urticaria pigmentosa associated with thrombocytosis as the initial symptom of systemic mastocytosis and Down's syndrome. J Eur Acad Dermatol Venereol. 2003;17:718–22.
8. Le Tourneau A, Gaulard P, D'Agay MF, Vainchencker W, Cadiou M, Devidas A, et al. Primary thrombocythaemia associated with systemic mastocytosis: A report of five cases. Br J Haematol. 1991;79:84–9.
9. Krsnik I, Ricard MP, Escribano LM, Calero MA, Perez Rus G, Garcia Suarez J, et al. Systemic mastocytosis and primary thrombocythaemia. Br J Haematol. 1991;77:437.
10. Jackson A, Burton IE. A case of POEMS syndrome associated with essential thrombocythaemia and dermal mastocytosis. Postgrad Med J. 1990;66:761–7.

J. Marcoval

Servei de Dermatologia, Hospital Universitari de Bellvitge, Hospitalet de Llobregat, Barcelona, Spain

E-mail address: jmarcoval@bellvitgehospital.cat

1578-2190/

Dermatomyositis-like Eruption in a Woman Treated With Hydroxyurea☆

Erupción dermatomiositis-*like* en una paciente tratada con hidroxiurea

To the Editor:

Dermatomyositis is an idiopathic inflammatory myopathy that usually progresses with inflammation of the skin and skeletal muscle. However, there are hypomyopathic and amyopathic forms that progress without laboratory abnormalities and/or muscle weakness, respectively.[1,2] While the cause is not generally known, some forms can be induced or exacerbated by drugs.[3] We report a case of dermatomyositis-like eruption with no muscle involvement associated with hydroxyurea and review specific immunological, clinical, and epidemiological findings.

The patient was a 63-year-old woman diagnosed with essential thrombocytopenia who had been receiving treatment with hydroxyurea since 2010. She was evaluated at the dermatology clinic for a 3-year history of erythematous, scaly lesions on the dorsum of the interphalangeal and metacarpophalangeal joints of the hands, dorsum of the feet, elbows, knees, and presternal area (Fig. 1). No muscular weakness or other remarkable cutaneous or mucosal manifestations were observed.

The histopathology findings are shown in Figure 2. The laboratory workup revealed normal results for inflammatory parameters and muscle enzymes. Negative results were recorded for myositis-specific antibodies (anti-Mi2, anti-MDA5, anti–SAE, anti-TIF, anti–NXP-2, anti-t-RNA-synthetase, anti–PMS, anti–SSA/Ro, anti-U1RNP, anti-Pm-Scl, and anti-Ku) and for antinuclear antibodies.

☆ Please cite this article as: Moreno-Artero E, Paricio JJ, Antoñanzas J, España A. Erupción dermatomiositis-*like* en una paciente tratada con hidroxiurea. Actas Dermosifiliogr. 2019;110:64–67.