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Letter

Adult-onset cutaneous mastocytosis in monozygotic twins

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Adult-onset cutaneous mastocytosis in monozygotic twins

Pietro Quaglino, MDa, Corresponding author Federica Mola, MDa, Anna Peroni, MDa,b, Michela Ortoncelli, MDa, Cristiana Di Bello, PhDc, Paola Savoia, MDa, Maria Grazia Bernengo, MDa

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To the Editor: A 36-year-old white man was evaluated for multiple pruritic skin lesions of 15 years duration. He had a history of symptomatic extrasystole during physical or emotional stress, sporadic flushing, anaphylaxis after an insect bite reaction, and endoscopic diagnosis of chronic gastritis with a positive culture for *Helicobacter pylori*. His monozygotic twin brother, who had suffered from asthma since early childhood, had developed similar cutaneous lesions on his trunk in the past few years. They had no siblings and their parents were unrelated. There was no history of skin or hematologic disease in the parents or other family members.

Clinical examination of the patient revealed multiple, red-brown, sharply defined macules and papules in a symmetric distribution over the trunk and limbs; they varied in size up to 0.5 mm (Fig 1). There was no lymphadenopathy or hepatosplenomegaly. His brother had identical cutaneous lesions localized to the trunk. Darier sign was slightly positive in each case.



Fig 1. Cutaneous mastocytosis on chest: multiple, round, red-brown macules and papules, sharply defined and not confluent, with symmetric distribution.

Lesional skin biopsy specimens showed similar histopathological features. There was a diffuse inflammatory infiltrate in the superficial dermis composed of a moderate number of mast cells arranged around dilated vessels and in the interstitial spaces, lymphocytes, rare histiocytes, and sporadic eosinophils; there was mild hyperpigmentation of the basal layer of the epidermis. Mast cells showed the classic metachromatic reaction with a toluidine blue stain. Imaging studies (chest radiograph, bone scan, computed tomography of the abdomen), a bone marrow biopsy specimen, and endoscopy of the esophagus and stomach did not disclose evidence of systemic mastocytosis. Serum tryptase levels were 10 and 8 ng/mL in the two twins, respectively (reference value <20 ng/mL in healthy subjects). A diagnosis of urticaria pigmentosa (UP) was therefore established.¹ Polymerase chain reaction (PCR) performed on cutaneous biopsy specimens from both patients using primers designed for exons 9, 11, and 17 of the c-kit proto-oncogene did not reveal any mutation.

Urticaria pigmentosa is the most frequent form of cutaneous mastocytosis and typically appears as a sporadic disease in childhood. The occurrence of UP in twins has been rarely described, with a total of 20 cases reported in the literature.² and ³ In every case the disease occurred during infancy or before 6 years of age and was characterized by the absence of extracutaneous involvement either at the time of diagnosis or during follow-up. Our twins were unusual because of the onset of cutaneous lesions during adult life. One had systemic symptoms presumed to be related to the release of mast cell–derived mediators (histamine, prostaglandins), but neither had evidence of extracutaneous disease.

Age at onset influences the clinical behavior of mastocytosis. Pediatric mastocytosis often resolves spontaneously with time and is usually limited to the skin, with only about 10% of patients developing extracutaneous disease.⁴ Mastocytosis arising in adulthood (35% of cases) is often persistent and associated with involvement of other organs. It has been suggested that the divergent clinical behavior between pediatric and adult mastocytosis may be related to the presence or absence of genomic mutations, in particular involving the c-kit proto-oncogene.⁵ A significantly higher percentage of c-kit mutations (77%) was demonstrated in adult-onset mastocytosis compared with pediatric cases (42%).⁵ Therefore the absence of a mutation in our patients could possibly indicate a favorable prognosis comparable to that of childhood UP.

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Conflicts of interest: None declared.

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