Early diagnosis of cutaneous mastocytosis in an infant: the importance of a clinical sign

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DESCRIPTION

A 3-month-old female infant presented with pruritic reddish-orange papules involving her face, trunk and limbs (figure 1A). Her parents had first noticed some papules on her back when she was 1 month old, with a progressive increase in number and extension and further involvement of the abdomen and then of the limbs. The parents described the skin lesions as becoming more evident and erythematous immediately after bathing. She had no other symptoms, was previously healthy and had no relevant known familiar history of skin conditions. On physical examination, an exacerbation of the reddish colour of the papules and swelling over the lesions was seen after rubbing or scratching—Darier’s sign (figure 1B). The parents’ description of this phenomenon after bathing can already be considered as a form of Darier’s sign. Owing to this clinical tip, the infant was promptly referred to a dermatologist with the clinical suspicion of cutaneous mastocytosis (CM). The diagnosis was supported by the dermatologist and confirmed by skin biopsy with histopathological staining with H&E showing a dense infiltrate of mast cells in the dermis forming agglomerations, particularly around capillary blood vessels (figure 2). The presented skin manifestation of mastocytosis can be classified as maculopapular CM or urticaria pigmentosa. In this case, the lesions had different sizes and shapes, consistent with the polymorphic subtype.1 The absence of systemic signs or symptoms (hypotension, syncope, flushing, nausea, vomiting, abdominal pain, lymphadenopathy or hepatosplenomegaly), the normal laboratory tests (complete blood count, kidney and liver function tests) and the normal level of serum total tryptase (5.2 µg/L) excluded systemic involvement (systemic mastocytosis), which enabled a wait-and-see approach.1 She initiated antihistamine therapy with dimetindene three times a day orally and practical measures, such as bathing with tepid water and air conditioning during hot weather, were carried out. During the following months, a progressive increase in the number and infiltration of the lesions was observed (figure 1C), with some new lesions on the palms and soles (figure 1D), with no other new-onset symptoms. At the age of 18 months, stabilisation in the number of lesions was seen and until the time of submission of this paper, no regression or worsening was reported.

Although the presentation and evolution of this case can be considered as an exuberant manifestation of CM in a child, the prognosis is, nevertheless,
Images in...

Figure 2  (A–C) Skin biopsy histopathology (H&E staining) showing a dense infiltrate of mast cells in the dermis forming agglomerations, particularly around capillary blood vessels.

Patient’s perspective

Mother—‘We noticed early that the lesions did not disappear and became more vivid after bathing, which led us to visit her paediatrician and we think that helped an early achievement of the diagnosis. I had never heard about this disease or about mast cells before. We got used to her appearance, but people frequently ask about it, because everyone can see the skin lesions, and sometimes it is hard to always have to explain.’

Learning points

► Early recognition of the Darier’s sign (a simple clinical sign to perform), which is pathognomonic for the presence of mast cells within the lesion, can enable an early diagnosis of mastocytosis, avoiding unnecessary and possibly harmful stress, examinations and treatments. This sign can also be recognised through inquiry to the parents about the exacerbation of the lesions after bathing or after scratching. A skin biopsy may be helpful when the diagnosis is in doubt, but it is not essential and biopsy findings alone are not sufficient for diagnosis, since mast cells can accumulate in the skin in other disorders.1

► Although mastocytosis (either in its systemic or cutaneous form) is a rare condition in children and adults, testing for the presence of Darier’s sign should be performed in persistent maculopapular skin lesions. On the other hand, lesions consistent with mastocytomas should not be rubbed or scratched, as this may lead to significant mast cell mediator release that can result in hypotension and flushing.1 In this case, the carer’s report of symptoms or photographs of the flares when the lesion is disturbed may be enough to raise clinical suspicion.

► Thorough clinical examination (search for lymphadenopathy, hepatosplenomegaly, flushing, gastrointestinal signs and symptoms) and laboratory testing (complete blood count, liver function tests and serum total tryptase) are crucial to evaluate possible systemic involvement and differentiate between cutaneous and systemic mastocytosis.1 3

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REFERENCES
