

Diffuse Cutaneous Mastocytosis, Bullous Variant, Presenting in a Six-Month-Old Infant

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A 6-month-old boy was presented with a 1-month history of intermittent facial flushing and a 1-day history of erythematous wheal rash on his trunk. The wheals rapidly evolved into bullae. There was a medical history of eczema and IgE specific for wheat, peanut, and egg. Scores of discrete, tense, and flaccid bullae of varying sizes and shapes were present on the torso and extremities (Figure 1). Large areas of glistening skin, denuded of epidermis also were evident. There were no lesions in the diaper area, on the palms, or soles, or on the oral mucosa. One hour after a skin biopsy, the patient developed signs of anaphylaxis but recovered quickly and did not require epinephrine. Results of the child's complete blood cell count and differential as well as the lymphocyte phenotyping was normal. The serum total tryptase level was elevated, at 81 ng/mL. The normal serum tryptase level: median 10 ng/mL, range 2-23 ng/mL. A skin biopsy specimen showed numerous CD117 and tryptase positive cells in clusters and bulla formation in the epidermis (Figure 2). Results of a bone marrow biopsy showed normocellular marrow with no c-kit gene mutation. The final diagnosis was diffuse cutaneous mastocytosis (DCM), bullous type. Mastocytosis is mast cell hyperplasia within tissue, with an incidence of approximately 2 in 300,000 per year.¹ Bullous mastocytosis is a severe variant of diffuse cutaneous mastocytosis, with approximately 30% cases manifesting within 6 months of age. The overall incidence is <1% of all mastocytosis cases.^{2,3} The typical childhood bullous mastocytosis is linked to Glu-839 Lys c-kit mutation, which our patient did not have.

Clinical features of DCM include blistering and erythematous rash, which may be reddish-brown or orange-yellow ("peau d'orange"); the skin becomes swollen, itchy, and red after stroking (Darier sign); there is flushing, urticaria, and pruritus.³ The bullous subtype skin lesions are characterized by peau



FIGURE 1. Multiple, large bullous lesions that extend across the trunk. Bullae are surrounded by thickened skin with a reddish hue and edematous texture.

d'orange skin overlaid by widespread bullae formation. DCM-induced anaphylaxis is a serious complication. Risk factors include extensive cutaneous involvement, and, as can be seen in this case, a biopsy of such lesions or the handling and/or trauma during the biopsy process can lead to anaphylaxis.³ To our knowledge, this is the first case reported in the literature in which a routine procedure such as the skin biopsy led to anaphylaxis, and this points out the importance of having drugs for emergency treatment of an allergic reaction kept at bedside during such procedures.

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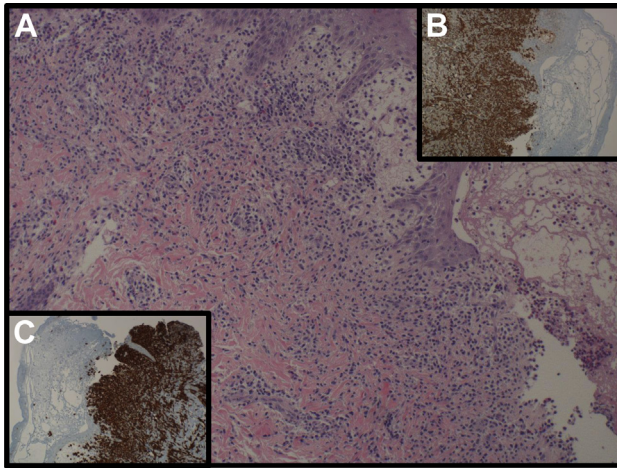


FIGURE 2. Composite histopathology image. **A**, Skin biopsy specimen, showing mast cell aggregation, supportive for mastocytosis (hematoxylin and eosin, original magnification $\times 10$). Skin staining for tryptase (**B**) and CD117 (**C**).

DCM prognosis seems to depend on the timeline of the presenting skin lesions. Some studies report that children who develop bullous eruptions as the first symptom have a higher

incidence of disease progress to systemic mastocytosis in comparison with patients who first present with infiltrative peau d'orange lesions.^{2,4,5} It is too early to make conclusions regarding this patient's disease progression.

Management for patients with mastocytosis includes H1 and/or H2 blockers for gastric protection and pruritus control as well as cromolyn sodium to prevent mast cell release of inflammatory mediators. Because the infant had significant skin involvement and the risk of inducing anaphylaxis by simply scratching the skin, topical cromolyn sodium also was prescribed to provide topical relief of pruritus and reduce possible allergic reactions. This treatment regimen was successful in that no further allergic reactions were observed.

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