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Recurrent anaphylaxis in an infant with Cutaneous Mastocytosis

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Abstract

Introduction: Rashes during infancy are mostly secondary to infection or allergy but can be due to rare serious disorders with poor prognosis; hence, the importance of clinical differential diagnosis and appropriate laboratory evaluation for early diagnosis and appropriate treatment, as in this case.

Case Report: A 10-mo-old boy presented with a rash since 5 mo of age. He was the first-born to non-consanguineous healthy parents, a product of uneventful full-term pregnancy and normal delivery and was exclusively breastfed.

At 5 mo of age, he developed vesicular eruptions started in the right axilla then spread over 24 hours to the trunk, neck and back. It was associated with irritability, colic, and vomiting but no fever. Neither he nor his mother was taking any medications. The eruption changed to tense bullae and became confluent with sizes ranging from 10-50 mm, with serous content and indurated erythematous base. The lesions spread further to the diaper area and extremities, with increasing severity and some bullae ruptured. Darier sign was demonstrated on erythematous skin areas. There was no involvement of mucus membranes. He had frequent exacerbations and on some occasions associated with severe gastrointestinal upset and difficulty in breathing, rhinorrhea, conjunctivitis, symptoms compatible with anaphylaxis. He recovered on treatment with parenteral dexamethasone and H1 anti-histamines for few days. CBC was WNL.

Apart from the skin lesions, the physical examination in-between episodes, was essentially normal, including growth and development. The mother noticed some exacerbations were related to hot climate, sun exposure, or during bathing which made the mother suspects soap.

Because supplementation of breastfeeding with baby foods began around the time of onset of the rash and the mother suspected a relation to her eating spicy food, a pediatrician initially thought of food allergy. However, at 8 mo his serum total IgE level was moderately high being 76 mg/dl and specific IgE was negative to several foods consumed at that time. Also a trial of dietary elimination (cow milk, wheat, egg, fish and nuts) from the diet of the infant and his lactating mother was not helpful. Topical antibiotics and corticosteroids had a limited temporary effect.

At 10 mo of age, during hot climate, he had a severe exacerbation and was promptly hospitalized due to extensive hemorrhagic bullae with rupture of many (figure 1), conjunctival congestion, rhinorrhea, progressive respiratory distress, vomiting, and fainting. Despite aggressive supportive therapy,

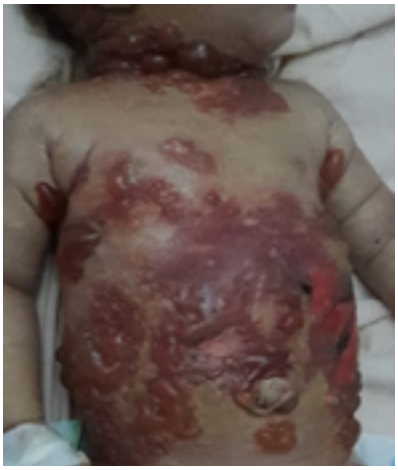

Figure 1: Skin lesions during the last severe exacerbation at 10 mo of age

he deteriorated with respiratory failure, undetectable blood pressure and developed multi-organ failure. He was transferred to the pediatric ICU with presumptive diagnosis of septic shock for assisted ventilation, adrenaline IV drip, pulse methylprednisolone (30 mg/kg/day), with improvement within 3 days followed by prednisolone 2 mg/kg/day.

Upon Allergy/Immunology consultation, systemic mastocytosis was suspected. He was found to have very high serum tryptase level at 120 ng/ml (Nl < 11). Skin biopsy revealed dermal and sub-epidermal vacuoles with extensive edema in the superficial layer of dermis and dense mast cells infiltrate, compatible with bullous mastocytosis and not other bullous disorders (figure 2). CBC, liver enzymes and kidney function gradually normalized. Bone marrow aspirate was normal without significant mast cells, supporting the diagnosis of diffuse cutaneous mastocytosis. C-Kit analysis did not reveal mutation.

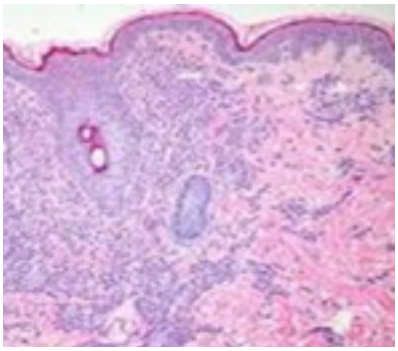

Figure 2: Skin biopsy showing abundance of mast cells.

Corticosteroids (2mg/kg/day) and tyrosine kinase inhibitor (imatinib mesylate 340mg/m^2/d) were administered for fear of systemic transformation with significant improvement. SCORMA (scoring mastocytosis) during the last exacerbation was very high at 116. In addition to an anaphylaxis action plan and avoiding common triggers (heat, sun, friction, maternal intake of spices), the management comprised skin care, tacrolimus ointment 0.03%, ketotifen (0.05mg/kg twice daily), sedating H1 and H2-antihistamine. With noticeable improvement, imatinib was discontinued after 1 mo and prednisolone was gradually withdrawn. Monthly follow up showed continued improvement of itching and skin lesions; reaching a remarkable degree by the age of 14 mo, with SCORMA down to just 5 (figure 3). Serum tryptase after 3 mo has markedly decreased to 55 ng/ml. Regular diet was gradually introduced without any reaction. Improvement was noticed in appetite, growth, and general wellbeing. He is currently 22 mo old and will be followed periodically.

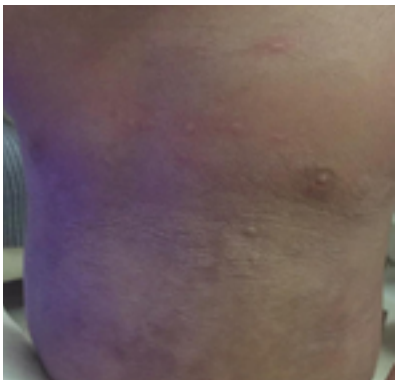

Figure 3: Skin marked improvement at age 14 months after pharmacologic therapy and avoidance of triggering factors.

Conclusion: The clinical and laboratory findings in our patient are compatible with diffuse cutaneous bullous mastocytosis. The onset was during early infancy and the course was rather complicated. It is the rarest variant of cutaneous mastocytosis and is usually seen from birth to the age of 2 yr, with recurrent anaphylaxis, and a guarded prognosis. The course and prognosis are influenced by the density and distribution of the mast cells. In addition to avoidance of triggers of mast cell activation and symptomatic therapies, specific therapeutic modalities depend on the severity of the disease, with imatinib reserved for severe cases.

References:

1. Da Costa ALF, Carvalho TCB, de Sousa AVL. Bullous mastocytosis in children: case report. An Bras Dermatol. 2005; 80(6): http://dx.doi.org/10.1590/S0365-05962005000700006.

2. Frieri M, Quershi M. Pediatric mastocytosis: a review of literature. Pediatr Aller Immunol Pulmonol. 2013; 26(4):175-80.

3. Ghosh A, Choudhury J, Dhar S. An infant with bullous mastocytosis: a rare form of bullous disorder. In J Paediatr Dermatol. 2017; 18(2): 122-4