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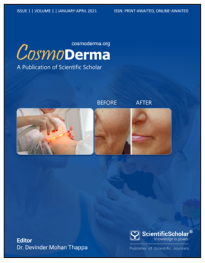

Letter to the Editor

Bullous mastocytosis: A rare but challenging diagnosis in infancy

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Dear Sir,

Mastocytosis is a rare disease characterized by an overabundance of mast cells in one or more organs.[1] Mastocytosis encompasses cutaneous mastocytosis and systemic mastocytosis. Cutaneous mastocytosis can present in different forms such as maculopapular, diffuse, or mastocytoma.[2] Bullous mastocytosis is a rare variant of diffuse cutaneous mastocytosis and is caused by mast cell degranulation, which releases proteases that separate the dermoepidermal layer. This results in bullous lesions that can mimic other skin conditions, such as epidermolysis bullosa simplex or staphylococcal scaling skin syndrome.

A 1-year-old boy presented with history of generalized fluid-filled bullae associated with itching and irritation since birth. The initial bulla appeared on his thigh the day of his delivery, followed by the development of new bullae on his face, neck, and trunk. These bullae contained yellow or hemorrhagic fluid, and ruptured easily on handling of the child. They healed with hyperpigmentation, without scarring. The lesions appeared in a cyclical pattern, with exacerbations and remissions, accompanied by recurrent vomiting and diarrhea. The baby was extremely irritable due to severe and generalized pruritus [Figure 1a]. Previously diagnosed as staphylococcal scalded skin syndrome and epidermolysis bullosa simplex, the child has received multiple intravenous, oral, and topical antibiotics. There was no relevant family history. Skin examination revealed post-inflammatory hyperpigmentation and a thickened, doughy texture of the skin. A tense hemorrhagic bulla was present on the scalp [Figure 1b]. Skin rubbing elicited a positive Dariers' sign. The systemic examination revealed no abnormalities. Tzanck smear and Gram staining showed only inflammatory cells, and all laboratory investigations were normal. Histopathology confirmed the diagnosis of bullous mastocytosis by showing a hyperkeratotic epidermis with acanthosis and focal subepidermal vesiculation with sheets of mast cells arranged in dermis [Figure 2a]. The upper dermis showed sheets of CD117-positive mast cells [Figure 2b]. X-rays of the head, spine, chest, extremities, and an ultrasound of the whole abdomen, were all normal. However, serum tryptase testing was not possible due to affordability issues. The parents were advised to avoid mast cell triggers, and the child was started on antihistamines, topical steroids, and topical antibiotics. Oral corticosteroids (betamethasone 0.1 mg/kg/day) were started for 3 months and then tapered. As a result, the doughy texture of the skin has improved, and there are no new bullous lesions and the child is under regular follow-up since past 1 year.

Bullous mastocytosis is a rare form of mastocytosis that primarily affects the skin, and it can be challenging to diagnose due to its resemblance to other dermatological conditions. Mutations have been found in the c-Kit (CD117) gene, which codes for the transmembrane receptor

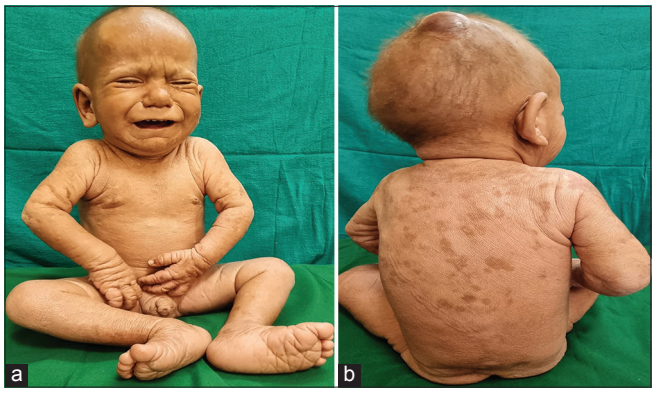

Figure 1: (a) The child looking irritable due to generalized pruritus; skin looking aged; and doughy (b) tense bulla on scalp filled with hemorrhagic fluid.

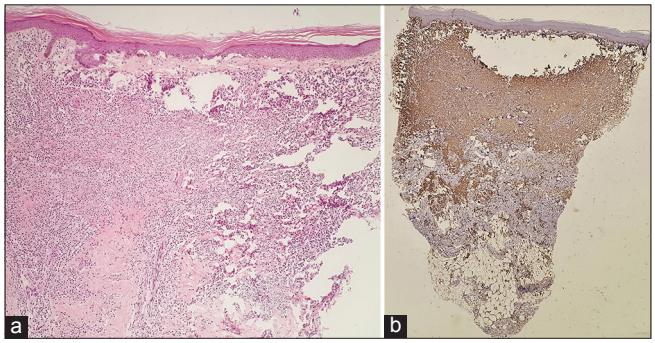

Figure 2: (a) Photomicrograph showing subepidermal vesiculation and dermal infiltrate of abundant mast cells (H and E; ×100) and (b) Mast cells with CD 117-positive antibody arranged in sheets in the dermis (CD 117 staining; ×40).

KIT.[2] Orkin *et al.* have identified two distinct presentations of bullous mastocytosis, namely, the neonatal form and the late-onset form.[3] The neonatal form is associated with severe complications and extracutaneous involvement, while the late-onset form usually presents with minimal extracutaneous involvement and a better prognosis. In our patient, the diagnosis of the neonatal form of bullous mastocytosis with no extracutaneous features was made. The clinical presentation of bullous mastocytosis can be easily mistaken for other skin conditions such as acrodermatitis enteropathica, epidermolysis bullosa simplex, and staphylococcal scalded skin syndrome making the diagnosis a challenge.[4] The appearance of diffuse, leathery skin may take several months to develop, and common features such as severe pruritus, diarrhea, vomiting, wheezing, and dermographism can aid in the diagnosis. Early diagnosis is essential to avoid unnecessary treatment and complications, including the risk of shock and sudden mortality triggered by massive mast cell degranulation which was possible in our patient. Typically, the first line of treatment includes oral antihistamines and corticosteroids. Other treatment options are phototherapy, imatinib mesylate, masitinib (a highly selective oral tyrosine kinase inhibitor), and omalizumab.[2] Studies have shown that cladribine may also be effective in treating cutaneous mastocytosis, with positive results observed in three patients in a study conducted by Barete *et al.*[5] In addition, miltefosine, a raft modulator, has shown promise as a topical treatment option for cutaneous mastocytosis.[6] Although bullous mastocytosis typically resolves by adolescence, the prognosis remains uncertain.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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