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Generalized Blistering in Infancy: A Rare Case Report on Bullous Mastocytosis

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Abstract

Mastocytosis is a rare myeloid neoplasm characterized by abnormal proliferation of mast cells, associated with *c-KIT* mutations. The disease is most frequently observed in childhood, with clinical presentations ranging from solitary mastocytoma to diffuse cutaneous involvement. Bullous mastocytosis is an uncommon variant of diffuse cutaneous mastocytosis, characterized by dermoepidermal separation due to mast cell degranulation, and may clinically resemble other vesiculobullous disorders.

We report the case of a 12-month-old male presenting with generalized pruriginous tense vesicles and bullae, evolving over three months with intermittent exacerbations. Histopathology revealed subepidermal blistering with a dense mast cell infiltrate, confirmed by CD117 and tryptase positivity on immunohistochemistry, establishing the diagnosis of bullous mastocytosis. The child was successfully managed with oral corticosteroids, antihistamines, topical corticosteroids, and supportive therapy, with resolution of lesions within two weeks.

This case underscores the importance of considering bullous mastocytosis in the differential diagnosis of infantile vesiculobullous disorders, as timely diagnosis and appropriate management can prevent morbidity.

Key words: Cutaneous mastocytosis, c-KIT mutation, Darier's sign, Mast cell disorders, Tryptase.

Introduction

Mastocytosis represents a group of disorders characterized by an abnormal accumulation of mast cells in one or more organs including the skin, bone marrow, liver, spleen, lymph nodes and gastrointestinal tract. More than 90% cases occur within the first 2 years of life. Bullous mastocytosis is a rare variant of diffuse cutaneous mastocytosis.[1] Here we report a case of a 1-year-old child with Bullous mastocytosis.

Case Report

A 1-year-old male child, born out of a non-consanguineous marriage, presented with multiple fluid filled lesions all over the body for 1 day. These ruptured on manipulation to form painful raw areas. Parents gave history of similar lesions in the past with exacerbations and remissions and preceding redness of skin over the past 3 months. These were associated with mild to moderate grade fever and irritability. No variation in incidence with rubbing or vigorous handling, symptoms of loose stools, flushing, difficulty in breathing, vomiting, syncope or failure to thrive were noted. His pre, post-natal, feeding, immunization history and developmental milestones were normal. Family history was unremarkable.

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On examination, the child was irritable but alert, general physical and systemic examination did not reveal any abnormalities. On cutaneous examination, multiple erythematous plaques with overlying coalescent, tense vesicles to bullae ranging from 3 mm to 4 cm involving scalp, forehead, malar regions, retroauricular area, neck, chest, abdomen, back, both arms, thighs, legs; containing serous and in few, hemorrhagic fluid, interspersed with wet, crusted erosions (figure 1a-d). Rest cutaneous examination was normal. Scarring, post inflammatory hyper or hypopigmentation, alopecia, milia formation were not present. Darier's sign could not be elicited due to extensive vesiculation. Differentials of Bullous mastocytosis, linear IgA bullous dermatosis, childhood bullous pemphigoid and epidermolysis bullosa simplex were considered.

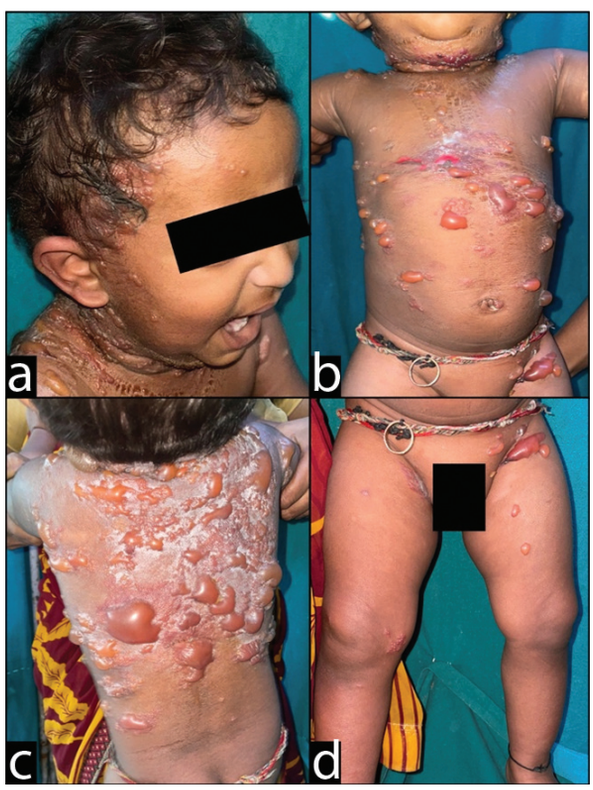

Figure 1a-d: Erythematous plaques with overlying tense vesicles to bullae on scalp, forehead, neck, chest, abdomen, back, both upper and lower limbs, with wet, crusted erosions at places (a-d).

Hematological parameters were within normal limits. Radiological investigations including chest X ray and abdominal ultrasound did not reveal any abnormalities. Tzanck smear demonstrated mast cells. Histopathological examination revealed tense, sub-epidermal blister with festooning of papillary dermis, containing RBCs, plasma cells, neutrophils, eosinophils and mast cells; papillary and upper reticular dermis revealed moderately dense band of mast cells, contributing to the diagnosis of bullous mastocytosis (figure 2a-b). Direct immunofluorescence was negative for IgG, IgA, and C3. Immunohistochemistry demonstrated CD117 and Tryptase positivity in subepithelial sheets of mononuclear cells (figure 2c-d). Bone marrow aspiration studies couldn't be performed owing to the reluctance of parents.

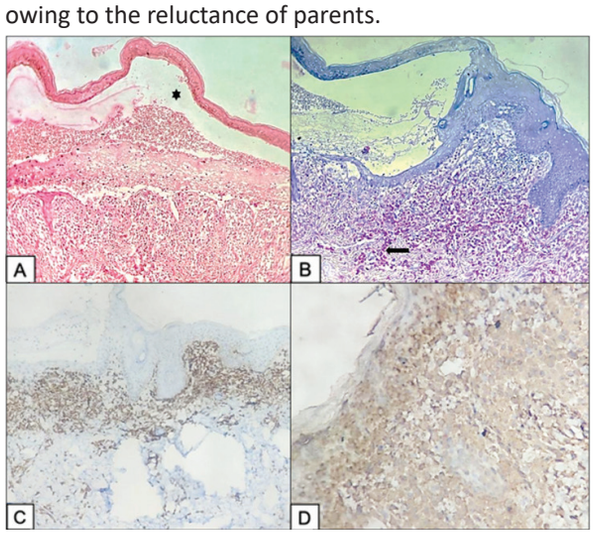

Figure 2a-d: Sub-epidermal blister (star), diffuse band like infiltration of dermis by mast cells (H&E 100x,a). Metachromatic purple red cytoplasmic granules in mast cells (arrow) (Giemsa 100x,b). Diffuse strong membranous and cytoplasmic positive in mast cells (IHC CD117 100x,c). Diffuse cytoplasmic positive in mast cells (IHC Tryptase 400x,d).

The patient was treated with syrup Deflazacort (6mg/5ml) 5ml with weekly tapering, syrup Cetirizine 2ml, systemic antibiotics and topical Desonide (0.05%) lotion. Parents were further counselled to avoid triggers. Resolution of lesions was observed within 2 weeks (figure 3a-c).

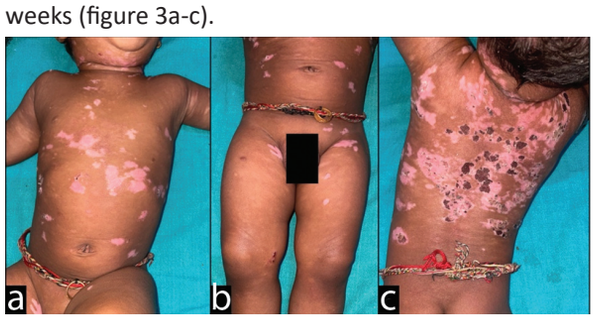

Figure 3a-c: Subsiding lesions after 2 weeks of treatment (a-c).

Discussion

Cutaneous mast cell diseases were first discovered by Nettleship and Tay in 1869.[2] In a series of 101 cases, cutaneous mastocytosis appeared by ages 6 months and 2 years in 73% and 97% of children, respectively without any sex predilection.[3]

Childhood disease is most commonly observed and frequently resolves by adolescence. Familial mastocytosis, transmitted in an autosomal dominant manner, is seldom encountered. Few cases don't show c-kit mutation, implying the role of other genes.[4] It almost always affects the skin with three main clinical presentations: (1) solitary or few (≤3) lesions, referred

to as "mastocytomas"– 15–50% of patients; (2) multiple (<10 to >100) lesions, referred to as "urticaria pigmentosa" (UP) or "maculopapular" mastocytosis– 45–75% of patients; and (3) diffuse cutaneous involvement– <5–10% of patients.[3] Diffuse cutaneous mastocytosis presents within initial months of life with pachydermia and variable hyperpigmentation in a generalized distribution. Blistering following urtication, due to serine protease produced by mast cells, is common in infants and young children but rarely extensive bullae and erosions may occur, as seen in our case. Release of heparin in some cases may contribute to hemorrhagic blisters.[5] Less common presentations in children include ill-defined tan or telangiectatic patches and xanthelasmoid mastocytosis.[6]

Children with cutaneous mastocytosis persisting till adolescence develop systemic mastocytosis in 15-30% cases. Although mastocytosis affects both children and adults, cutaneous variant is common in children while systemic mastocytosis presents in adults predominantly. Few patients develop systemic symptoms like pruritis, flushing, gastrointestinal disturbance, and even anaphylaxis due to the release of mast cell mediators.[7]

Diagnosis of cutaneous mastocytosis requires one major criterion [typical skin lesions], plus one or two minor criteria [(1) monomorphic mast cell infiltrate (large aggregates of tryptase-positive mast cells with >15 cells/cluster) or scattered mast cells exceeding 20 cells per microscopic high power field(40x); and (2) detection of an activating KIT mutation in lesional skin].[8] Our patient satisfied all criteria.

Systemic mastocytosis is suspected in cases with severe systemic symptoms, extensive blistering, large surface area of involvement, organomegaly, high serum total tryptase and lack of response to symptomatic treatment. Systemic variants are associated with cytopenias, ascites, gastrointestinal disturbances, malabsorption, osteolysis, hemodynamic instability and malignant (leukemic) transformation, rarely associated with cutaneous mastocytosis.[9]

Mainstay of management involves avoiding triggers precipitating mast cell degranulation and use of systemic antihistamines. Acetyl salicylic acid, ketotifen, oral cromolyn sodium (400–800 mg/day)and topical cromolyn are also implemented. Omalizumab benefits in symptomatic adult-onset mastocytosis recalcitrant to antihistamine therapy. Psoralen plus UVA or narrowband ultraviolet B therapy, potent topical corticosteroids, topical calcineurin inhibitors, topical miltefosine and intralesional steroids may control pruritus and whealing.[10]

Systemic therapies for aggressive mastocytosis include Interferon-α-2b, Cladribine, Imatinib mesylate, Midostaurin, Dasatinib, Nilotinib. Our patient responded well to oral steroids and antihistamines along with topical steroids. Regular follow-up every six to 12 months is essential until resolution of skin lesions as few cases may progress to systemic mastocytosis.[10] Only a handful of cases have been reported globally, a literature review of diffuse cutaneous bullous mastocytosis revealed varied clinical presentations and treatment approaches. Asati et al. reported a 3-month-old male with a 10-day history who was managed with antihistamines and topical mid-potent corticosteroids.[11] Verma et al. described a 7-month-old female with a 3-month history who received oral betamethasone (0.1 mg/kg/day), antihistamines, and topical mid-potent corticosteroids.[12] Achehboune et al. documented a 16-month-old female with a 10-month duration who was treated with antihistamines and topical mid-potent corticosteroids.7 Ridlo et al. reported a 3-month-old male with a 2-month history who was managed with topical mid-potent corticosteroids, antibiotics, and saline compresses.[13] Almheiri et al described a 6-month-old male infant with blistering since 1 month, treated with antihistamines and topical mid-potent corticosteroids on the affected areas.[14] Dev et al reported a 1-year-old male child with a history since birth managed with antihistamines, oral corticosteroids (betamethasone 0.1 mg/kg/day) in tapering doses and topical steroids and antibiotics.[15] These cases highlighted the consistent use of antihistamines and topical mid-potent corticosteroids, with systemic steroids and antibiotics added in select cases based on severity.

Conclusion

This case underscores the importance of considering bullous mastocytosis in the differential diagnosis of infantile vesiculobullous disorders, as timely diagnosis and appropriate management can prevent morbidity.

References

1. Meni C, Bruneau J, Gorgin-Lavialle S, et al. Paediatric mastocytosis: a systematic review of 1747 cases. Br J Dermatol. 2015;172(3):642-51. https://doi.org/10.1111/bjd.13567
2. Nettleship E. Rare orms of urticaria. Br Med J. 1869;2:323-4.
3. Lange M, Niedoszytko M, Renke J, et al. Clinical aspects of paediatric mastocytosis: a review of 101 cases. J Eur Acad Dermatol Venereol. 2013;27(1):97-102. https://doi.org/10.1111/j.1468-3083.2011.04365.x
4. Castells M, Butterfield J. Mast cell activation syndrome and mastocytosis: initial treatment options and long-term management. J Allergy Clin Immunol Pract. 2019;7(4):1097-106. https://doi.org/10.1016/j.jaip.2019.02.002
5. Lange M, Hartmann K, Carter MC, Siebenhaar F, Alvarez-Twose I, Torrado I, et al. Molecular background, clinical features and management of pediatric mastocytosis: status 2021. Int J Mol Sci. 2021;22(6):2586. https://doi.org/10.3390/ijms22052586
6. Husak R, Blume-Peytavi U, Pfrommer C, Geilen CC, Goerdt S, Orfanos CE. Nodular and bullous cutaneous mastocytosis of the xanthelasmoid type: case report. Br J Dermatol. 2001;144(2):355-8. https://doi.org/10.1046/j.1365-2133.2001.04026.x
7. Achehboune K, Elboukhari K, Baybay H, Elloudi S, Mernissi FZ. Bullous disease in infant: think of cutaneous mastocytosis. Pan Afr Med J Clin Med. 2020;2:49. https://doi.org/10.11604/pamj-cm.2020.2.49.21265
8. Valent P, Horny HP, Escribano L, Longley BJ, Li CY, Schwartz LB, et al. Diagnostic criteria and classification of mastocytosis: a consensus proposal. Leuk Res. 2001;25(7):603-25. https://doi.org/10.1016/S0145-2126(01)00038-8
9. Valent P, Aberer E, Beham-Schmid C, Fellinger C, Fuchs W, Gleixner KV, et al. Guidelines and diagnostic algorithm for patients with suspected systemic mastocytosis: a proposal of the Austrian competence network (AUCNM). Am J Blood Res. 2013;3(2):174-90.
10. Sandes AF, Medeiros RS, Rizzatti EG. Diagnosis and treatment of mast cell disorders: practical recommendations. Sao Paulo Med J. 2013;131(4):264-74. https://doi.org/10.1590/1516-3180.2013.1314590
11. Asati DP, Tiwari A. Bullous mastocytosis in a 3-month-old infant. Indian Dermatol Online J. 2014;5(4):497-500. https://doi.org/10.4103/2229-5178.142520
12. Verma KK, Bhat R, Singh MK. Bullous mastocytosis treated with oral betamethasone therapy. Indian J Pediatr. 2004;71(3):261-3. https://doi.org/10.1007/BF02724280
13. Ridlo M, Mahadi I, Siregar R. Diffuse cutaneous mastocytosis with generalized bullae mimicking bullous pemphigoid: single case report. In: Proceedings of the 2nd International Conference on Tropical Medicine and Infectious Disease (ICTROMI). 2019;23-25:436-40. https://doi.org/10.5220/0009991004360440
14. Almheiri SK, Pakran J, AlFalasi AA, El Bahtimi R. Bullous mastocytosis: a rare variant of diffuse cutaneous mastocytosis. Cureus. 2024 Jan 4;16(1):e51660. https://doi.org/10.7759/cureus.51660
15. Dev PP, Lakhani R, Bansal S, Khunger N. Bullous mastocytosis: a rare but challenging diagnosis in infancy. CosmoDerma. 2023;3:53. https://doi.org/10.25259/CSDM_49_2023