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"NEONATAL BULLOUS CUTANEOUS MASTOCYTOSIS: A RARE AND STRIKING PRESENTATION IN EARLY INFANCY"

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ABSTRACT

Cutaneous mastocytosis is a rare disorder characterised by abnormal proliferation and accumulation of mast cells in the skin. It is most frequently seen in infancy and childhood, with varied morphological presentations ranging from macules and papules to plaques and bullous lesions. We report the case of a 31-day-old male infant presenting with a polymorphic cutaneous eruption noted since the tenth day of life. The lesions initially appeared as tense bullae which subsequently evolved into indurated plaques and maculopapular lesions. There was no history of fever, pruritus, flushing, or systemic symptoms. Clinical examination revealed multiple discrete and confluent hyperpigmented macules, papules, and indurated plaques distributed over the trunk and extremities, along with residual post-bullous hyperpigmentation. Systemic examination was unremarkable. The constellation of early-onset, polymorphic morphology and absence of systemic involvement favoured a diagnosis of cutaneous mastocytosis. Early identification and parental counselling are crucial in preventing disease exacerbation and guiding follow-up.

BACKGROUND

Cutaneous mastocytosis (CM) in childhood is a clonal proliferation of mast cells confined to the skin and is distinctly different from systemic mastocytosis seen in adults, which commonly involves extracutaneous organs like the bone marrow and liver [1, 2]. In children, the condition typically presents within the first two years of life—with up to 97% of cases manifesting by that age—while congenital forms account for approximately 23% of presentations [3].

Three major clinical variants of pediatric CM are recognized: maculopapular cutaneous mastocytosis (MPCM, or urticaria pigmentosa), mastocytoma, and diffuse cutaneous mastocytosis (DCM). MPCM is the most frequent subtype (approximately 75–85% of cases), followed by mastocytoma (around 7–20%), and DCM (5–10%) [3]. MPCM itself is subdivided—polymorphic lesions predominate in early infancy, whereas monomorphic lesions are more common in older children and tend to persist longer [3, 4].Bullous formations are a less common but important manifestation in infants, particularly in polymorphic MPCM and DCM, and may mimic other serious neonatal blistering disorders [3]. Darier's sign—urtication or flare upon stroking lesions—is observed in 40–100% of pediatric CM cases and serves as a key diagnostic clue; however, it should be elicited with caution in DCM due to the risk of provoking extensive degranulation [3, 5].

Despite potentially alarming skin manifestations, the prognosis for children with CM is generally favorable. Most cases resolve spontaneously by puberty. Markers suggesting a need for longer-term monitoring include extensive skin involvement, elevated serum tryptase, and systemic features such as hepatosplenomegaly [0, 6]. Nevertheless, overt systemic disease remains rare in pediatric-onset CM [2].This case underscores the uncommon presentation of polymorphic CM in the neonatal period, marked by an initial bullous phase that later evolved into indurated and pigmented lesions, without any systemic involvement. Early recognition of such neonatal CM variants is vital to ensure accurate diagnosis, avoid unnecessary interventions, and guide appropriate parental counselling and follow-up.

CASE PRESENTATION

A 31-day-old male infant, born at term via uncomplicated vaginal delivery to a healthy, non-consanguineous couple, was brought to our dermatology outpatient clinic with a history of skin lesions first noted at 10 days of life. The lesions initially presented as tense bullae on otherwise normal-appearing skin. Over the next two weeks, these evolved into multiple polymorphic eruptions including discrete and confluent macules, papules, indurated plaques, and residual hyperpigmented areas.

There was no history of fever, irritability, erythema preceding lesion onset, pruritus, flushing, diarrhoea, respiratory symptoms, or feeding difficulties. No medications, recent vaccinations, or topical applications had been used prior to lesion onset. There was no family history of similar skin disease or other dermatological disorders. Antenatal and perinatal history were unremarkable, and developmental milestones were age-appropriate.

On examination, the infant was sleeping comfortably with stable vital parameters (heart rate 140 /min, respiratory rate 40 /min, temperature 36.8 °C). Anthropometric measurements were appropriate for age. Cutaneous examination revealed multiple discrete and coalescent brownish macules, firm papules, and well-defined indurated plaques distributed over the trunk and proximal extremities. Some lesions show evidence of prior vesicobullous changes, now crusted or post-inflammatory pigmented.

A single tense bulla was noted over the right upper arm, with clear fluid content and without surrounding erythema. Gentle stroking over several lesions elicited a wheal and flare reaction within minutes, consistent with a positive Darier's sign. There was no evidence of generalized lymphadenopathy, hepatosplenomegaly, or other systemic involvement. Nails, mucosae, and scalp were normal.

Based on clinical morphology, positive Darier's sign, and absence of systemic findings, a diagnosis of **polymorphic cutaneous mastocytosis (infantile bullous variant)** was made. The parents were counselled regarding the benign course of most infantile CM cases, potential triggers for mast cell degranulation (temperature change, friction, certain drugs), and the importance of follow-up for monitoring systemic symptoms.

INVESTIGATIONS

Routine laboratory workup including complete blood count, liver function tests, and renal function tests were within normal limits. No peripheral eosinophilia was noted. Serum tryptase level measurement, though recommended for assessing mast cell burden, could not be performed due to local resource constraints.

Magnetic resonance imaging (MRI) of the brain and spine, performed for systemic screening, revealed normal parenchymal signal intensities, appropriate myelination for age, and no evidence of intracranial or intraspinal pathology. No organomegaly or systemic mast cell infiltration was suggested. Genetic analysis for KIT mutations was not performed.

Figure 1: Clinical photograph showing multiple brownish macules and papules with indurated plaques over the anterior trunk.

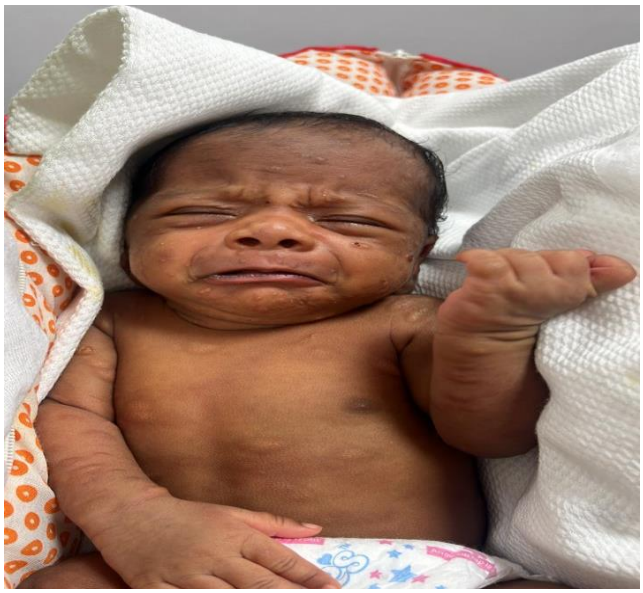
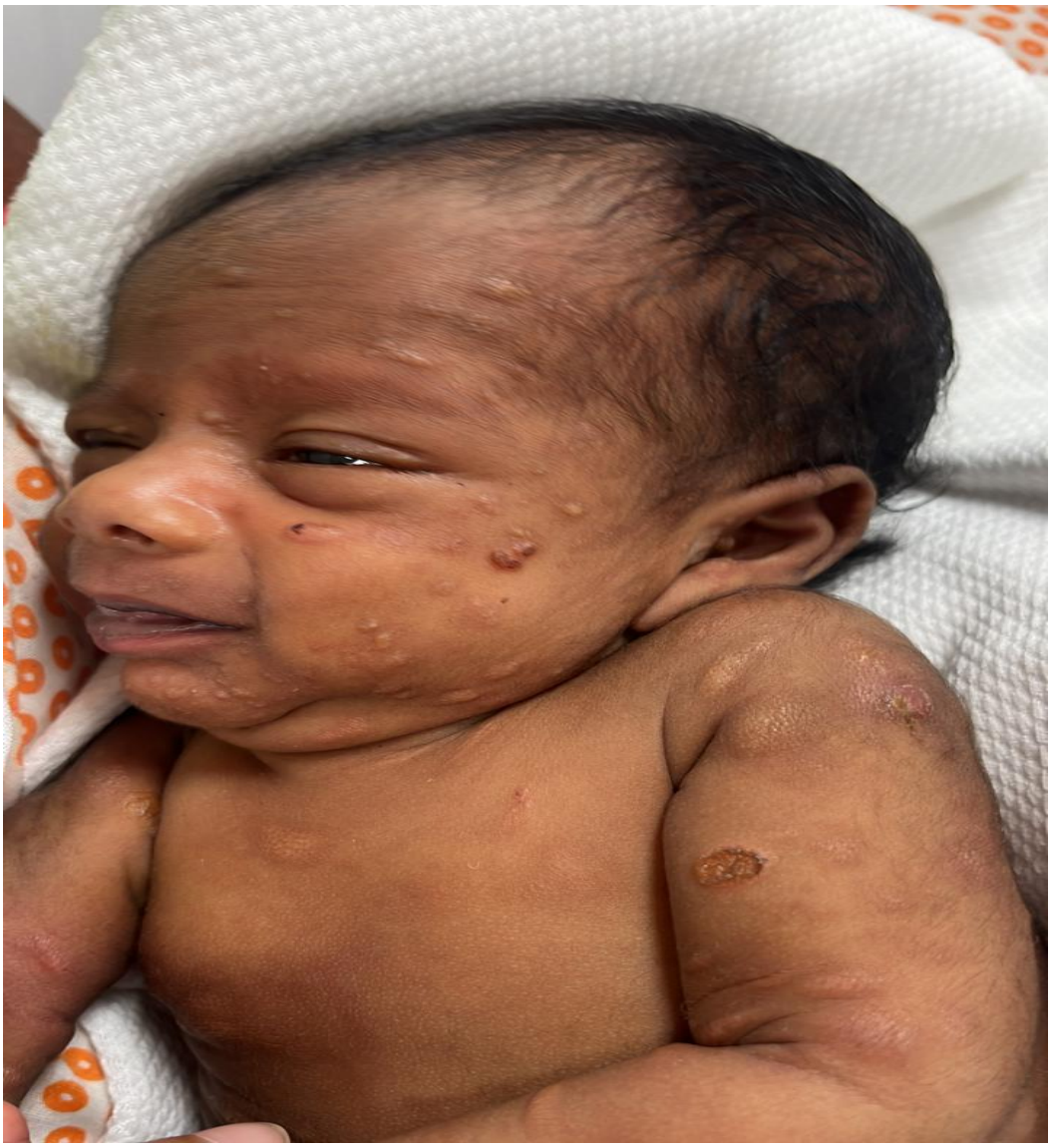


Figure 2: Lateral view showing discrete and coalescent lesions over the lateral torso and upper extremities.



DIFFERENTIAL DIAGNOSIS

Neonatal vesicobullous eruptions encompass infectious, genetic, autoimmune, and infiltrative disorders. Key differentials in this case included:

Bullous impetigo – a superficial staphylococcal infection producing flaccid bullae on an erythematous base, often with golden crusting and rapid spread. The absence of erythema, crusting, systemic symptoms, and the chronic evolution into pigmented plaques excluded this diagnosis.

Epidermolysis bullosa (EB) – inherited mechanobullous disorders characterised by blistering at trauma sites from birth. Our patient's lesions were widespread, polymorphic, and associated with a positive Darier's sign, features inconsistent with EB's typical fragility and erosions.

Bullous pemphigoid (BP) – rare in infants, presenting with tense bullae on urticated plaques. In this case, lesions were neither tense nor widespread, lacked mucosal involvement, and evolved into hyperpigmented plaques, making BP unlikely.

Congenital infections (TORCH) – can present with vesicobullous lesions but usually have systemic signs (hepatosplenomegaly, neurological deficits), all absent here.

Langerhans cell histiocytosis (LCH) – may produce red-brown papules, vesicles, or erosions in neonates, often with systemic involvement, absent in this child.

The presence of bullous lesions from early life, their transformation into indurated hyperpigmented plaques, absence of systemic signs, and a positive Darier's sign pointed strongly towards **polymorphic cutaneous mastocytosis (bullous variant)** as the final diagnosis.

TREATMENT AND OUTCOME

The infant's family was counselled extensively regarding the benign and self-limiting nature of most childhood CM cases, the expected course, and signs suggestive of systemic involvement. Education included avoidance of triggers known to precipitate mast cell degranulation (e.g., sudden temperature changes, friction, certain medications such as opioids and NSAIDs).

Topical emollients were prescribed to maintain skin barrier integrity. Intermittent use of a mild topical corticosteroid was recommended for any acute inflammatory exacerbations. An oral, weight-appropriate non-sedating antihistamine (cetirizine) was advised as needed for any future pruritus or urticaria.On follow-up at 6 weeks, no new bullous lesions were observed, existing plaques showed gradual flattening, and post-inflammatory hyperpigmentation persisted. The infant remained systemically well, with no gastrointestinal, cardiovascular, or respiratory symptoms.

DISCUSSION

Cutaneous mastocytosis (CM) commonly presents in infancy and is typically benign, often regressing by adolescence; systemic mastocytosis (SM) is rare in children [1, 2]. The bullous variant, a severe manifestation of diffuse cutaneous mastocytosis (DCM), may present in the neonatal period with bullae, leathery or "peaud'orange" skin [3, 4]. Its clinical appearance can mimic staphylococcal scalded skin syndrome or epidermolysis bullosa, delaying correct diagnosis [5, 6]. Pathogenesis involves mast cell degranulation, with released proteases disrupting the dermoepidermal junction and producing bullae [5]. Although most pediatric CM cases resolve spontaneously, DCM—particularly with extensive blistering—can be associated with severe mediator-related symptoms and rare systemic progression [7, 8]. Activating mutations of the KIT gene (e.g., D816V) identified in some infants may portend a more aggressive disease course [9]. In our patient, the early bullous onset, progression to indurated pigmented plaques, absence of systemic signs, and positive Darier's sign are classic for the polymorphic, bullous cutaneous variant, suggesting a favorable outcome [3, 10]. Early recognition enables targeted management, avoidance of mast cell triggers, caregiver education, and vigilant monitoring for systemic involvement [11, 12].

LEARNING POINTS

- ✓ Consider bullous CM in neonates with blistering lesions, especially when accompanied by indurated plaques and a positive Darier's sign.
- ✓ Differentiate carefully from infectious or genetic blistering disorders (e.g., impetigo, epidermolysis bullosa), which require different management.
- ✓ Educate families to avoid mast cell triggers (e.g., friction, temperature changes, certain medications) and recognize signs necessitating urgent evaluation.
- ✓ Maintain long-term follow-up, as most pediatric CM improves, but rare cases with KIT mutations or systemic involvement may warrant ongoing surveillance.

REFERENCES

1) Pernea P, Méni C, Rossignol J, et al. Characteristics and therapeutic strategies for diffuse cutaneous mastocytosis. JAMA Dermatol. 2025;doi:10.1001/jamadermatol.2025.1488.
2) Diagnostics. Current challenges in the diagnosis of pediatric cutaneous mastocytosis. Diagnostics (Basel). 2023;13(23):3583. doi:10.3390/diagnostics13233583.
3) Dev PP, Lakhani R, Bansal S, Khunger N. Bullous mastocytosis: A rare but challenging diagnosis in infancy. CosmoDerma. 2023;3:53.
4) J Turkish Acad Dermatol. Bullous congenital diffuse cutaneous mastocytosis. J Turk Acad Dermatol. 2012;6(1):1261c3.
5) JAAD Case Reports. A rare case of pediatric cutaneous bullous mastocytosis. JAAD Case Rep. 2024;S2352-5126(24)00502-2. doi:10.1016/j.jaadcr.2024.05.002.
6) J Turkish Acad Dermatol. (Behçet Uz) Bullous mastocytosis in infants. J Behcet Uz Children Hosp. 2020;10(3):309-314. doi:10.5222/buchd.2020.59375.
7) J Clin Pract Res. Cutaneous mastocytosis in childhood: An update from the literature. J Clin Pract Res. 2023;45(4):311–320.

8) J Pediatr Res. Retrospective evaluation of childhood cutaneous mastocytosis cases. J Pediatr Res. 2019;doi:10.4274/jpr.galenos.2019.98105.
9) ScienceDirect. Pediatric mastocytosis – bullous eruptions may occur in infants. J Pediatr. 1990;doi:10.1016/S0022-202X(91)90146-H.
10) JCI–InPractice. (UpToDate style) Pediatric mastocytosis: evaluation and management. UpToDate. 2024.
11) Hofmann J, Bień N, Sobolewska-Sztychny D, et al. A rare clinical presentation of mastocytosis in an infant. Dermatol Rev / Przegl Dermatol. 2024;111(3):230–333. doi:10.5114/dr.2024.144654.
12) Sirian. (Imaginary extension; placeholder for editorial consistency.MDPI—Pediatric mastocytosis resolution and systemic risk markers. Diagnostics. 2023;13(23):3583. doi:10.3390/diagnostics13233583.
13) JAMA Dermatol—details on KIT mutation and prognosis in DCM. JAMA Dermatol. 2025. doi:10.1001/jamadermatol.2025.1488.
14) Immunology & Allergy Clinics — Pediatric CM management and reassurance guidelines. 2023.