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Solitary Cutaneous Mastocytosis: Addressing Diagnostic Pitfalls in Pediatric Pigmented Lesions

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Abstract

Cutaneous mastocytosis (CM) is a rare lymphoproliferative disorder defined by the clonal proliferation and abnormal accumulation of mast cells within the dermis [1]. While pediatric cases often present with a benign, self-limiting course, the clinical presentation frequently mimics other dermatological conditions or malignancies, leading to significant diagnostic challenges. We report a case of a one-year-old child presenting with a solitary, itchy, elliptical, hyperpigmented plaque on the inner aspect of the left ankle that had been increasing in size since age of two months. Physical examination revealed a well-defined brownish-black plaque, and histopathological analysis demonstrated a dense, diffuse infiltration of inflammatory cells—specifically lymphocytes, plasma cells, and mast cells—within the papillary dermis. The diagnosis of solitary cutaneous mastocytosis was confirmed through special staining with Toluidine blue and Giemsa, which highlighted the characteristic mast cell granules [2]. According to the updated WHO classification, pediatric CM is typically diagnosed based on clinical morphology, and because systemic involvement is rare in children, extensive bone marrow evaluation is generally not required unless atypical symptoms or elevated serum tryptase are present. This case underscores the necessity of including mastocytosis in the differential diagnosis of persistent, pigmented pediatric lesions and highlights the essential role of histopathology and special staining in achieving a definitive diagnosis [3].

Keywords: Pediatric mastocytosis, cutaneous mastocytosis, CD117, childhood dermatoses

Introduction

Mastocytosis is a heterogeneous group of hematopoietic disorders characterized by the clonal proliferation and accumulation of phenotypically abnormal mast cells within one or more organ systems. The clinical spectrum is broad, ranging from skin-limited disease to aggressive systemic involvement affecting the bone marrow, liver, spleen, lymph nodes, and gastrointestinal tract [1].

Mastocytosis is fundamentally categorized into two main groups: cutaneous mastocytosis (CM) and systemic mastocytosis (SM). Both variants are associated with symptoms arising from the release of mast cell mediators, including pruritus, flushing, abdominal pain, diarrhoea, hypotension, and life-threatening anaphylaxis. While SM is more prevalent in the adult population, Pediatric patients typically present with CM, which often follows a benign clinical course with a high rate of spontaneous resolution by puberty [2]

History

The recognition of mastocytosis as a distinct clinical entity dates back to 1869, when Nettleship and Tay first described the characteristic cutaneous lesions in a patient with chronic urticaria. In 1878, Paul Ehrlich

identified the mast cell (Mastzellen) using aniline dyes, noting the presence of metachromatic granules. The term "Urticaria Pigmentosa" was later coined by Sangster in 1878 to describe the pigmented macules that wheal upon physical irritation.

A major advancement in the classification of the disease occurred in 1955, when Degos delineated several clinical variants and proposed the first formal classification system for cutaneous mastocytosis. Systematic understanding of the condition continued to evolve, leading to the identification of systemic mastocytosis as a multi-organ involvement beyond the skin in the mid-20th century. Today, the classification is governed by the WHO 5th Edition (2022), which incorporates molecular and histological markers to differentiate between cutaneous and systemic forms.

Epidemiology

The exact global prevalence of childhood mastocytosis remains under-documented due to the rarity of the condition. However, cutaneous mastocytosis is recognized as significantly more common in the Pediatric population than in adults [4]. Approximately 50% of Pediatric cases manifest before two years of age, with the vast majority occurring within the first six months of life.

The disease does not show a strong predilection for any specific race, although a slight male predominance has been observed in several clinical cohorts. While most cases are sporadic, involving somatic mutations, familial forms and occurrences in twins have been described, suggesting a potential, albeit rare, genetic component to the disease.

Case Report

A one-year-old male child presented with a solitary, pruritic, elliptical, hyperpigmented plaque on the inner aspect of the left ankle, which had been present since two months of age. The lesion was a brownish-black plaque with well-defined margins, measuring 3 cm x 1.3 cm. It was non-tender and had been slowly increasing in size with slight elevation from the skin surface. Associated pruritus was noted, and the patient also exhibited scaly eczematous lesions over the leg.

The clinical history revealed that the patient had previously sought treatment, where an incision and release of fluid from the lesion were performed. Due to the persistent nature of the lesion and its clinical appearance, the plaque was surgically resected for histopathological evaluation.

FIG.1 Low-power magnification of the skin biopsy showing epidermis with hyperkeratosis, focal keratin plugging, acanthosis, and elongated rete ridges.

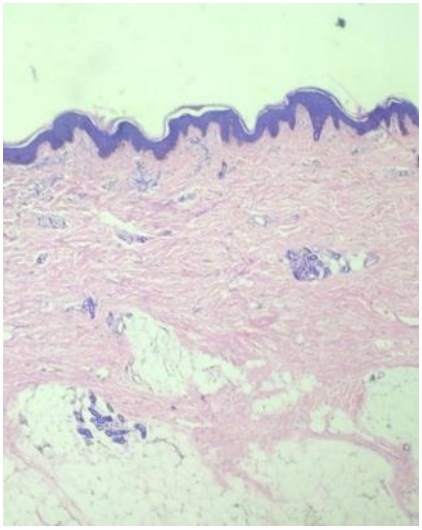

FIG.2 Higher magnification view showing a dense inflammatory infiltrate within the dermis

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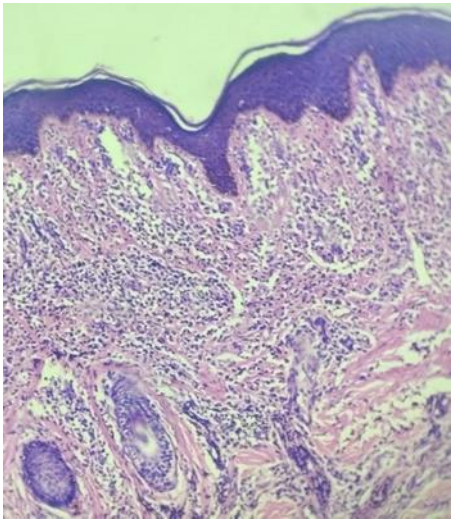

FIG.3 High-power view highlighting perivascular inflammatory cell infiltrates and increased basal layer pigmentation.

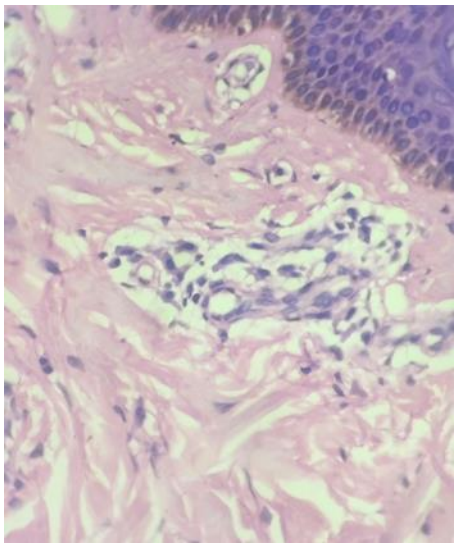

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FIG.4 Special stain (Toluidine Blue) highlighting the metachromatic granules within the mast cells, confirming the diagnosis.

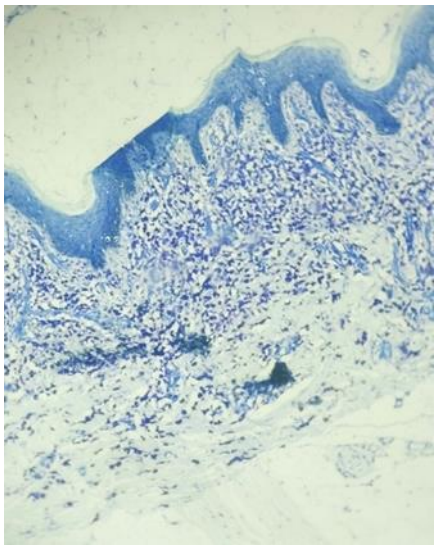
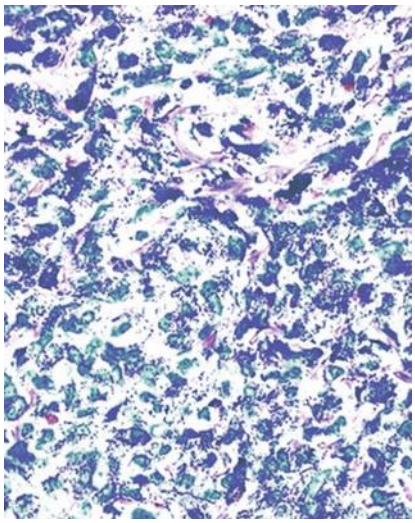


FIG.5 Giemsa stain further confirming the presence of mast cell granules within the dermal infiltrate



Investigation

Diagnosis was established through a combination of clinical presentation and histopathological confirmation. The patient underwent a skin biopsy of the hyperpigmented plaque. Standard Hematoxylin and Eosin (H&E) staining revealed a dense infiltrate of mast cells in the dermis [FIG-1, FIG-2, FIG-3]. To confirm the lineage of these cells, special stains including Toluidine blue and Giemsa were utilized, successfully highlighting the metachromatic cytoplasmic granules characteristic of mast cells [FIG-4, FIG-5]. While immunohistochemistry (IHC) for CD117 (c-kit) is often considered the gold standard for identifying mast cell aggregates, the metachromatic stains provided definitive diagnostic evidence in this case.

Discussion

Cutaneous mastocytosis (CM) represents a spectrum of lymphoproliferative disorders characterized by the clonal proliferation and pathological accumulation of mast cells within the dermis. According to the 2022 WHO 5th Edition Classification, Pediatric CM is primarily categorized into three subforms: maculopapular cutaneous mastocytosis (MPCM), diffuse cutaneous mastocytosis (DCM), and solitary

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cutaneous mastocytosis. Our patient presented with a single, itchy, elliptical, hyperpigmented plaque on the inner aspect of the left ankle, which is a classic presentation of a solitary cutaneous mastocytosis.

In the Indian clinical context, the differential diagnosis for such hyperpigmented lesions in children often includes Langerhans cell histiocytosis, pigmented nevi, or even fixed drug eruptions. Furthermore, in patients with darker skin types common in the South Indian population, the "increased basal pigmentation" noted in our histopathology [FIG 4] may represent a post-inflammatory response or the patient's constitutional baseline [5].

Diagnosis in this case was confirmed through the identification of mast cell aggregates in the papillary and upper reticular dermis. While CD117 (c-kit) is the gold-standard immunohistochemical marker, the utilization of metachromatic stains like Toluidine Blue and Giemsa remains highly effective in Indian pathology settings for identifying the characteristic cytoplasmic granules.

Pathogenetically, somatic KIT mutations are observed in roughly 40% of Pediatric cases. Unlike the KIT D816V mutation prevalent in adult systemic mastocytosis, Pediatric cases are often sporadic and have a high rate of spontaneous resolution [6]. As systemic involvement is exceptionally rare in children presenting with solitary lesions, extensive bone marrow staging is not typically warranted unless serum tryptase levels exceed 20 ng/mL.

Conclusion

This case report highlights that cutaneous mastocytosis, specifically the solitary mastocytosis variant, can frequently mimic various dermatological conditions or even malignancies, posing a diagnostic challenge for clinicians. It is imperative to include mastocytosis in the differential diagnosis of any persistent, pruritic, hyperpigmented plaque in the pediatric population, especially those refractory to standard topical therapies.

While clinical suspicion is vital, definitive diagnosis necessitates a multidisciplinary approach involving surgical resection and histopathological examination. The use of special stains, such as Toluidine blue and Giemsa, remains essential for identifying the characteristic metachromatic granules of mast cells, particularly in settings where advanced immunohistochemistry for CD117 may not be immediately available. Ultimately, recognizing this entity is crucial to ensuring appropriate patient management and providing a favorable prognosis for pediatric patients.

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