

Cutaneous mastocytosis presenting as urticaria pigmentosa with prominent bone involvement: A case report

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Abstract

Mastocytosis comprises a rare group of disorders characterized by the accumulation of mast cells, primarily in the skin and/or internal organs. Cutaneous mastocytosis includes maculopapular cutaneous mastocytosis (urticaria pigmentosa), diffuse cutaneous mastocytosis, and cutaneous mastocytoma, with urticaria pigmentosa representing the most frequent clinical variant. It typically presents as erythematous to brownish macules or papules, often associated with pruritus.

We report the case of a 48-year-old woman with a history of rheumatologic and cardiac disease, who presented with multiple disseminated, well-demarcated, livid-brown, mildly pruritic macules involving the trunk and extremities. Skin biopsy revealed a perivascular mastocytic inflammatory infiltrate, with immunohistochemistry strongly positive for CD117, confirming the diagnosis of cutaneous mastocytosis. Serum tryptase levels were within normal limits. Systemic evaluation revealed skeletal involvement, including femoral osteopenia and vertebral osteoporosis.

The patient was managed with lifestyle and dietary modifications aimed at avoiding known triggers, in addition to H1-antihistamine therapy, resulting in good functional improvement.

This case is of interest due to its presentation in adulthood with systemic skeletal involvement despite normal serum tryptase levels, highlighting the importance of comprehensive evaluation even in apparently indolent cutaneous forms. It also underscores potential limitations, including the absence of KIT mutation analysis and bone marrow biopsy, which could further refine disease classification and prognostic assessment.

Keywords: Mastocytosis; Urticaria Pigmentosa; Mast Cells; Skin Disorder; Systemic Skeletal Involvement

1. Introduction

Mastocytosis is defined as a heterogeneous group of disorders characterized by abnormal activation and/or proliferation of neoplastic mast cells. It is a rare condition that may involve the skin alone or other organs, including the bone marrow, gastrointestinal tract, bones, liver, spleen, and lymph nodes in systemic forms. Several clinical subtypes have been described [1]. We report a case of cutaneous mastocytosis presenting as urticaria pigmentosa.

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2. Case report

A 65-year-old woman with a history of rheumatologic and cardiac disease, receiving acenocoumarol (Sintrom), presented with pruritic erythematous lesions on the trunk that had been evolving for 5 years, associated with episodes of flushing and mixed-pattern arthralgia.

Physical examination (Figure 1) revealed multiple erythematous-to-brownish pigmented plaques with ill-defined margins, distributed over the trunk (Figure a), flanks (Figure b), back (Figure c), and limbs (Figure d). Darier's sign was negative.

Dermoscopic examination revealed a diffuse erythematous-to-light-brown background with a poorly organized pigmentary pattern and multiple fine dotted vascular structures irregularly distributed within the lesion (Figure 2).

Skin biopsy showed cutaneous tissue lined by a regular epidermis with overlying orthokeratotic hyperkeratosis. The dermis demonstrated a perivascular inflammatory infiltrate rich in mononuclear cells composed of lymphocytes and regularly shaped cells with a mast cell-like appearance. Basal layer hyperpigmentation was also observed. Immunohistochemical analysis revealed positive staining for anti-CD117 antibody in mast cells with both perivascular and interstitial distribution.

Laboratory investigations revealed a normal complete blood count, preserved renal function, mild hepatic cytolysis with ALT (GPT) at 41 IU/L (1.2 times the upper normal limit), normal electrolyte levels, LDH at 211 IU/L, and slightly elevated β 2-microglobulin at 2.7 mg/L. Phosphocalcic evaluation showed serum calcium at 100 mg/L, phosphorus at 28 mg/L, and severe vitamin D deficiency. Serum tryptase levels were negative.

Radiological assessment demonstrated diffuse bone demineralization. Radiographs of the long bones showed cortical thickening, while arm radiographs revealed duplication of the diaphyseal cortex. Hand radiographs demonstrated band-like demineralization of the metacarpophalangeal joints. Spine radiographs showed L2-L3 disc space narrowing with osteophyte formation. Bone densitometry revealed femoral osteopenia and spinal osteoporosis.

A diagnosis of cutaneous mastocytosis presenting as urticaria pigmentosa was established.

The patient received therapeutic education including avoidance of scratching, hot baths, and spicy foods in order to prevent lesion exacerbation. Photoprotection was recommended, and treatment with a daily H1-antihistamine associated with an evening emollient cream was initiated because of intense pruritus.





Multiple erythematous to hyperpigmented macular plaques with poorly defined, irregular borders, located on the trunk (a), flanks (b), lower back (c), and limbs (d).

Figure 1 Clinical appearance



Dermoscopy reveals a diffuse erythematous-to-light brown background (star) with a subtle, poorly organized pigmented pattern (arrow). Multiple fine dotted vascular structures (triangle) are observed, distributed irregularly within the lesion.

Figure 2 Dermoscopic appearance of a cutaneous lesion in a patient with mastocytosis.

3. Discussion

Cutaneous mastocytosis is a rare disorder resulting from abnormal mast cell accumulation or excessive mast cell activation [1]. Urticaria pigmentosa, also known as maculopapular cutaneous mastocytosis, is the most common clinical form. It typically presents as pruritic pigmented macules or papules that may coalesce into symmetrically distributed plaques. Lesion color may range from reddish-purple to brownish-beige. The lesions predominantly involve the trunk but may also affect the limbs and, less frequently, the face, scalp, palms, soles, and even mucous membranes. The appearance of lesions after rubbing is referred to as Darier's sign [2].

The differential diagnosis of urticaria pigmentosa may be challenging, particularly in atypical presentations. Bullous impetigo should be considered in the presence of erosive or crusted lesions, especially in children, whereas urticaria is usually characterized by transient wheals without persistent pigmented macules. Lichen planus pigmentosus may mimic the pigmented appearance of the lesions but typically lacks pruritic whealing and Darier's sign. Juvenile xanthogranuloma can also present as yellow-brown papules or nodules; however, its histopathological features are distinct. Insect bites may simulate urticarial papules but are generally associated with an acute onset and a history of exposure. Autoimmune bullous dermatoses, particularly in bullous forms of mastocytosis, should also be considered and can be excluded by histopathology and direct immunofluorescence studies [3,4].

Histopathological examination, supported by immunohistochemistry, remains essential for confirming the diagnosis of cutaneous mastocytosis in adults and should be interpreted in correlation with clinical findings [5].

Functional investigations aimed at detecting systemic involvement are mandatory in the presence of suggestive clinical features, including bone involvement, gastrointestinal manifestations particularly hepatic involvement, hematological abnormalities, bone marrow involvement, as well as other manifestations such as respiratory or cardiac involvement [3,6].

No systematic workup is recommended unless diffuse disease or alarming clinical features are present [6]. In our patient, the reported joint pain prompted radiological investigations and bone densitometry in anticipation of preventive management for osteoporosis.

There is currently no established consensus regarding the treatment of cutaneous mastocytosis; however, several measures may significantly improve patient's quality of life. The main recommendations include avoidance of triggering foods and medications, prevention of abrupt temperature changes, and avoidance of iodinated contrast agents. In abdominal assessment, magnetic resonance imaging is preferred over computed tomography whenever possible [7].

For symptomatic management, H1-antihistamines remain the first-line therapy. Bisphosphonate supplementation appears necessary in patients with a bone densitometry T-score below -2.5, whereas vitamin D and calcium supplementation may be sufficient in cases of osteopenia [6].

Recent therapeutic advances in mastocytosis have led to the development of targeted KIT inhibitors such as midostaurin and avapritinib, which have demonstrated significant clinical efficacy in reducing mast cell burden, improving symptom control, and enhancing quality of life. Ongoing studies are currently evaluating next-generation tyrosine kinase inhibitors (e.g., bezuclastinib and elenestinib), as well as emerging biologic and immunomodulatory approaches, highlighting a rapidly evolving therapeutic landscape [8].

4. Conclusion

Mastocytosis is a rare and heterogeneous disease with mainly cutaneous forms that are generally indolent but can significantly impact quality of life. Management is primarily based on patient education and avoidance of triggering factors which may provoke mast cell degranulation and symptom flares. H1-antihistamines remain the main symptomatic treatment. In this case, strict trigger avoidance combined with regular follow-up is essential to ensure adequate disease control and to monitor for possible systemic involvement.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

References

- [1] Pileri A, et al. Cutaneous mastocytosis: clinical, dermoscopic and pathological features. *Ann Dermatol Venereol*. 2016. from: <https://www.sciencedirect.com/science/article/abs/pii/S1773035X16300120>
- [2] Macri A, Tefferi A. Urticaria Pigmentosa. In: *StatPearls*. Treasure Island (FL): StatPearls Publishing; 2026. <https://www.ncbi.nlm.nih.gov/books/NBK482503/>

- [3] Zhang Y, et al. Urticaria pigmentosa: a case report with review of the literature. *Int J Clin Dermatol*. 2024;7(1):16. from: <https://sciencepublishinggroup.com/article/10.11648/j.ijcd.20240701.16>
- [4] Hartmann K, et al. Challenges in the diagnosis of cutaneous mastocytosis. *Diagnostics (Basel)*. 2024;14(2):161. <https://www.mdpi.com/2075-4418/14/2/161>
- [5] Méni C, et al. Mastocytosis in the skin: approach to diagnosis, evaluation, and management in adult and pediatric patients. *J Eur Acad Dermatol Venereol*. 2022. PMID: 34901755. <https://pubmed.ncbi.nlm.nih.gov/articles/34901755/>
- [6] Valent P, Akin C, Hartmann K, Alvarez-Twose I, Brockow K, Hermine O, et al. Updated diagnostic criteria and classification of mast cell disorders: a consensus proposal. *Hemasphere*. 2021;5(11):e646. doi:10.1097/HS9.0000000000000646. PMID: 34901755.
- [7] Tamhankar PM, Suvarna J, Deshmukh CT. Cutaneous mastocytosis: getting beneath the skin of the issue: a case report. *Cases Journal*. 2009 Jan 20;2:69. doi:10.1186/1757-1626-2-69. <https://link.springer.com/article/10.1186/1757-1626-2-69>
- [8] Chifotides HT, Bose P. SOHO state of the art update and next questions: current and emerging therapies for systemic mastocytosis. *Clin Lymphoma Myeloma Leuk*. 2025;25(1):1-12. doi: 10.1016/j.clml.2024.06.005. available from <http://pubmed.ncbi.nlm.nih.gov/39168723>