DOI: 10.7860/JCDR/2025/77007.21439

Rare Clinical Image of Cutaneous Mastocytosis

SHUBHAM KHANKE[1], MUJAHID KHAN[2], KEYUR DUDHAT[3], PRATIKSHA MAHURE[4]

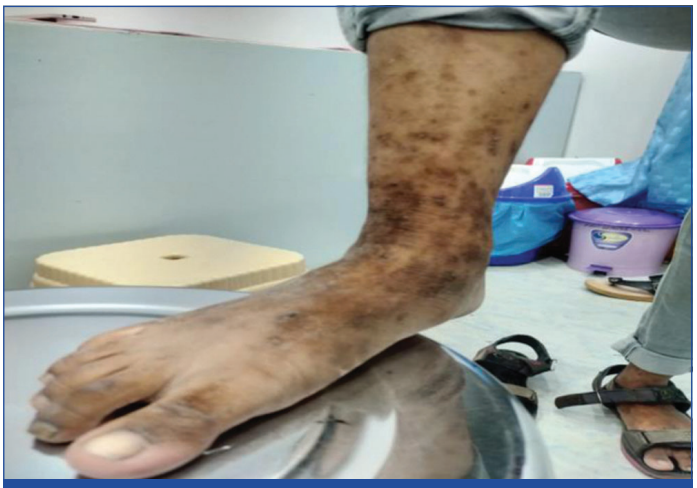
Keywords: Anaphylaxis, Maculopapular, Mast cells, Omalizumab

Mastocytosis, a rare condition also referred to as a clonal bone marrow disorder, is characterised by an excessive presence of CD34+ mast cell progenitors and functionally impaired mast cells. This disorder can affect both adults and children, leading to symptoms such as hives, itching, and potentially life-threatening anaphylactic shock. Often misdiagnosed, mastocytosis is frequently associated with other underlying conditions, indicating that its prevalence may be underestimated. The disease can present in two distinct forms: Cutaneous Mastocytosis (CM) and systemic mastocytosis. These conditions impact multiple bodily systems, leading to a diverse array of symptoms and clinical manifestations [1].

An 18-year-old male presented with a one-year history of pruritus and discolouration affecting his legs. Upon examination, pigmented lesions were observed, some of which were elevated and exhibited red or brown hues, as seen in [Table/Fig-1]. These lesions were prone to blistering when subjected to friction, and no systemic abnormalities were noted. Accompanying symptoms, including pruritus, erythema, musculoskeletal pain, and fatigue, were discovered. The diagnosis was made based on a Complete Blood Count (CBC), serum tryptase, and skin biopsy confirming mastocytosis, as shown in [Table/Fig-2]. The serum tryptase level was 16 ng/mL. The histopathological findings revealed a mild to moderate perivascular and interstitial infiltrate of mast cells in the dermis. The mast cells were round to oval in shape with granular cytoplasm. No significant atypia was observed in the mast cell morphology. There was an absence of epidermal involvement, and the overlying epidermis appeared unremarkable. Dermal collagen appeared intact with no evidence of fibrosis. Blood vessels within the dermis were unremarkable, without evidence of vasculitis. Toluidine Blue Stain highlighted positive for metachromatic granules, confirming the presence of mast cells. The differential diagnosis for mastocytosis includes urticaria, pruritic skin disorders, melanocytic nevi, autoimmune bullous skin disorders, and Langerhans cell histiocytosis [2]. The treatment was administered as Gandhak Rasayan 250 mg, two tablets BD after food, Panchatikta Ghrita one teaspoon in the morning, Arogyavardhini Vati 250 mg BD, and Cutis ointment for local application for fifteen days.

The skin is the organ most commonly affected by mastocytosis. CM is categorised based on its clinical manifestations and is also characterised by the timing of disease onset. Typically, CM manifests during early childhood; however, cases of adult-onset CM are also observed [3]. In pediatric patients, the likelihood of systemic involvement is relatively low. Childhood-onset mastocytosis shows spontaneous, partial, or complete remission in 67% of cases, whereas in adults, systemic mastocytosis is present in more than 25% of CM cases [4]. Mastocytosis is defined by the clonal expansion of mast cells across multiple organs. In the case of childhood-onset mastocytosis, approximately 90% of instances manifest before the age of 2, with a minority presenting at birth [5].

Adult-onset mastocytosis typically manifests between the ages of 20 and 35 years and is frequently linked to mutations in the c-KIT gene, particularly in exon 17. In 2-4% of instances, mastocytosis may present as a familial condition, with at least one first-degree relative also exhibiting the disorder. There are three forms of CM, with Maculopapular Cutaneous Mastocytosis (MPCM) and Urticaria Pigmentosa (UP) being the most prevalent, accounting for 70-90% of cases [6]. This is followed by mastocytoma of the skin, which occurs primarily in children and represents 10-30% of cases. Diffuse CM is exceedingly uncommon, typically presenting at birth or during early infancy, with an incidence of 1-3%. The main objective of the treatment for mastocytosis is to manage symptoms associated with mast cell mediators. The therapeutic foundation lies in antagonists of the histamine receptor and short-term topical corticosteroids.



[Table/Fig-1]: Maculopapular lesions on right ankle.

Investigation	Observed value	Unit	Method
Hb%	14.8	gm%,	Photometric measurement
MCV	84.4	Fl	RBC histogram
MCH	28.1	pico-gm	(Hb/RBC) X 10
MCHC	33.1	%	(Hb/RBC) X 100
Total RBC count	5.2	millions/cu.mm	Coulter principle
RDW	13.3	%	RBC histogram
HCT	43.5	%	RBC X MCV/10
Total WBC count	6800	/cu.mm	Coulter principle
Monocytes	03	%	Coulter VCS technology
Granulocytes	64	%	Coulter VCS technology
Lymphocytes	25	%	Coulter VCS technology
Eosinophils	8	%	Coulter VCS technology
Basophils	00	%	Coulter VCS technology
Total platelets count	2.77	Lacs/cu.mm	Coulter principle

[Table/Fig-2]: CBC investigation on cell counter with Peripheral Smear (PS). Peripheral Smear RBCs – Normocytic Normochromic Platelets – Adequate on smear. No Haemoparasite seen; Hb: Haemoglobin; MCV: Mean corpuscular volume; MCH: Mean corpuscular haemoglobin; MCHC: Mean corpuscular haemoglobin concentration; RBC: Red blood cell; RDW: Red cell distribution width; HCT: Haematocrit; WBC: White blood cell; VCS: Volume conductivity scatter

First-generation and second-generation H1 antihistamines can effectively manage skin manifestations, including pruritus, flushing, and a burning sensation on the skin. The first-line drugs are nonsedating H1 antihistamines [7]. Phototherapy serves as a secondary treatment option that may be utilised when antihistamines fail to yield the anticipated benefits. Emerging therapeutic alternatives encompass omalizumab and agents that target KIT [8]. In recent years, omalizumab, a monoclonal antibody that specifically targets human IgE, has emerged as a promising treatment alternative for mastocytosis linked to recurrent anaphylaxis [9].

REFERENCES

[1] Li JY, Ryder CB, Zhang H, Cockey SG, Hyjek E, Moscinski LC, et al. Review and updates on systemic mastocytosis and related entities. Cancers (Basel). 2023;15(23):5626. Doi: 10.3390/cancers15235626. PMID: 38067330; PMCID: PMC10705510.

[2] Matos AL, Figueiredo C, Gonçalo M. Differential diagnosis of urticarial lesions. Front Allergy. 2022;3:808543. Doi: 10.3389/falgy.2022.808543. PMID: 36238930; PMCID: PMC9552949.

[3] Sandru F, Petca RC, Costescu M, Dumitraşcu MC, Popa A, Petca A, et al. Cutaneous mastocytosis in childhood-update from the literature. J Clin Med. 2021;10(7):1474. Doi: 10.3390/jcm10071474. PMID: 33918305; PMCID: PMC8038134.

[4] Giona F. Pediatric mastocytosis: An update. Mediterr J Hematol Infect Dis. 2021;13(1):e2021069. Doi: 10.4084/MJHID.2021.069. PMID: 34804443; PMCID: PMC8577558.

[5] Sathishkumar D, Balasundaram A, Mathew SM, Mathew L, Thomas M, Balasubramanian P, et al. Clinicopathological profile of childhood onset cutaneous mastocytosis from a tertiary care center in South India. Indian Dermatol Online J. 2021;12(5):706-13. Doi: 10.4103/idoj.IDOJ_924_20. PMID: 34667757; PMCID: PMC8456265.

[6] Nemat K, Abraham S. Cutaneous mastocytosis in childhood. Allergol Select. 2022;6:01-10. Doi: 10.5414/ALX02304E. PMID: 35028497; PMCID: PMC8750954.

[7] Gangireddy M, Ciofoaia GA. Systemic Mastocytosis. [Updated 2023 Jul 4]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025. Available from: https://www.ncbi.nlm.nih.gov/books/NBK544345/.

[8] Giustozzi MI, Torre AC, Ritchie C, Parisi CAS. Phototherapy as an alternative in the treatment of chronic spontaneous urticaria. Front Allergy. 2024;5:1468983. Doi: 10.3389/falgy.2024.1468983. PMID: 39640431; PMCID: PMC11617558.

[9] Pelaia C, Calabrese C, Terracciano R, de Blasio F, Vatrella A, Pelaia G. Omalizumab, the first available antibody for biological treatment of severe asthma: More than a decade of real-life effectiveness. Ther Adv Respir Dis. 2018;12:1753466618810192. Doi: 10.1177/1753466618810192. PMID: 30400762; PMCID: PMC6236630.

PARTICULARS OF CONTRIBUTORS:

1. Postgraduate Scholar, Department of Ayurveda, Mahatma Gandhi Ayurved College, Hospital and Research Centre, Wardha, Maharashtra, India.
2. Associate Professor, Department of Ayurveda, Mahatma Gandhi Ayurved College, Hospital and Research Centre, Wardha, Maharashtra, India.
3. Postgraduate Scholar, Department of Ayurveda, Mahatma Gandhi Ayurved College, Hospital and Research Centre, Wardha, Maharashtra, India.
4. Postgraduate Scholar, Department of Ayurveda, Mahatma Gandhi Ayurved College, Hospital and Research Centre, Wardha, Maharashtra, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Shubham Khanke,
Salod, Hirapur, Wardha, Maharashtra, India.
E-mail: shubhamkhanke@gmail.com

PLAGIARISM CHECKING METHODS: [Jain H et al.]

- Plagiarism X-checker: Nov 28, 2024
- Manual Googling: Apr 10, 2025
- iThenticate Software: Apr 12, 2025 (5%)

ETYMOLOGY: Author Origin

EMENDATIONS: 7

AUTHOR DECLARATION:

- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Nov 27, 2024**
Date of Peer Review: **Feb 19, 2025**
Date of Acceptance: **Apr 14, 2025**
Date of Publishing: **Sep 01, 2025**