

## Langerhans Cell Histiocytosis and Mastocytosis: A Rare Association

Romain Boisseau<sup>1</sup>, Lea Scaramuzzino<sup>1</sup>, Gregory Bosphore<sup>1</sup>, Kim Heang Ly<sup>1</sup>

<sup>1</sup> CHU Dupuytren, Dermatology Department, Limoges, France

**Key words:** Langerhans cell histiocytosis, Mastocytosis, Rare association, Case report, Clinical features

**Citation:** Boisseau R, Scaramuzzino L, Bosphore G, Ly KH. Langerhans Cell Histiocytosis and Mastocytosis: A Rare Association. *Dermatol Pract Concept*. 2025;15(4):5660. DOI: <https://doi.org/10.5826/dpc.1504a5660>

**Accepted:** April 6, 2025; **Published:** October 2025

**Copyright:** ©2025 Boisseau et al. This is an open-access article distributed under the terms of the Creative Commons Attribution-NonCommercial License (BY-NC-4.0), <https://creativecommons.org/licenses/by-nc/4.0/>, which permits unrestricted noncommercial use, distribution, and reproduction in any medium, provided the original authors and source are credited.

**Funding:** None.

**Competing Interests:** None.

**Authorship:** All authors have contributed significantly to this publication.

**Corresponding Author:** Romain Boisseau, MD, CHU Dupuytren, Dermatology Department, Limoges, France.  
Email: [romain.boisseau@chu-limoges.fr](mailto:romain.boisseau@chu-limoges.fr)

### Introduction

Histiocytoses are rare disorders characterized by dysfunction or proliferation of mononuclear phagocytic system cells. These disorders can affect any organ, with the skeleton, skin, and pituitary gland most commonly involved [1]. Mastocytoses are marked by the accumulation of clonal mast cells in both the skin and internal organs, including the bone marrow, spleen, lymph nodes, liver, and gastrointestinal tract [2]. Therefore, histiocytoses and mastocytoses are distinct clonal diseases, with no documented overlap.

### Case Presentation

A 25-year-old male patient presented to the emergency department (ED) in October 2023 with progressive right-sided facial swelling over two months, which had recently prevented him from opening his right eye (Figure 1). A computed tomography (CT) scan identified a 2-cm lytic lesion in the frontal bone (Figure 2). The patient remained in good health. Two days following his ED visit, the patient attended the Dermatology Department. Examination revealed pigmented

macules on his limbs, abdomen, and lower back, which he reported having since childhood. Darier's sign was positive. A skin biopsy indicated cutaneous mastocytosis, and serum tryptase levels were 1.5 times above normal. Molecular analysis detected a KIT D816V mutation in both blood and skin samples. In December 2023, curettage of the frontal bone lesion revealed Langerhans cell histiocytosis (LCH), confirmed by immunohistochemistry and molecular testing for a *BRAF V600E* mutation. Pituitary function tests were normal. A chest CT scan showed multiple bilateral punched-out nodules in the upper lung lobes and scattered centrilobular micronodules, suggesting early pulmonary histiocytosis. By June 2024, post-surgical assessments showed resolved facial swelling. Magnetic resonance imaging (MRI) confirmed the stability of the right frontal bone lesion. A follow-up chest CT indicated resolution of the pulmonary nodules. All laboratory results were normal, except for elevated tryptase levels. The patient quit smoking and remained in excellent health. In November 2024, repeat MRI of the facial mass confirmed the stability of the frontal lesion. Based on these findings, treatment for both conditions was deemed unnecessary. A biannual follow-up plan was established.



Figure 1. Progressive right-sided facial swelling over two months.

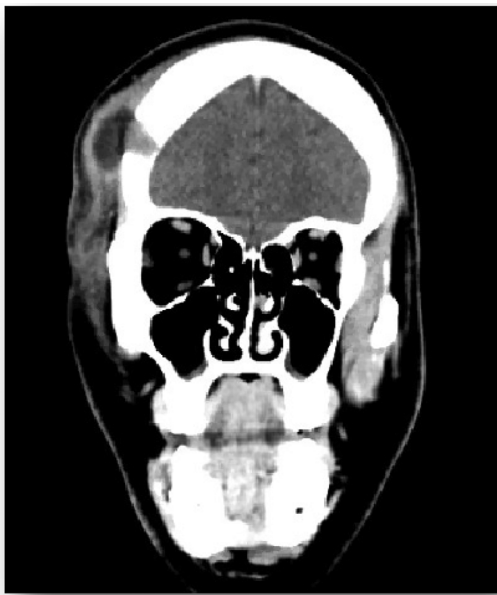


Figure 2. CT scan identifying a 2-cm lytic lesion in the frontal bone.

## Conclusion

LCH exhibits a broad spectrum of severity. Treatment strategies are determined by the extent of disease, with localized single-system disease often managed through local therapy or observation, while multisystemic disease necessitates systemic treatment. This approach has led to improved 5-year survival rates of 90% in children and 70% in adults [3]. Typically beginning in early childhood, pediatric mastocytosis primarily affects the skin and regresses in ~75% of cases within six years [4]. However, the lesions can persist. Aggressive forms are symptomatic from the onset [4].

Histiocytoses and mastocytoses have occasionally been reported to co-occur [5]. However, the specific coexistence of mastocytosis and LCH is exceptional. The literature records only a single case involving a young child, who presented with LCH affecting the eye and skin at 15 days old, resolving within two months, followed by the emergence of two mastocytomas [5]. The coexistence of LCH and mastocytosis in adults remains virtually undocumented. Both diseases in our patient currently do not require treatment. However, as lesions persist, regular follow-up remains essential.

## References

1. Haupt R, Minkov M, Astigarraga I, et al. Langerhans cell histiocytosis (LCH): guidelines for diagnosis, clinical work-up, and treatment for patients till the age of 18 years. *Pediatr Blood Cancer*. 2013;60(2):175-184. DOI:10.1002/pbc.24367. PMID: 23109216
2. Valent P, Akin C, Hartmann K, et al. Updated Diagnostic Criteria and Classification of Mast Cell Disorders: A Consensus Proposal. *Hemasphere*. 2021;5(11):e646. Published 2021 Oct 13. DOI:10.1097/HS9.0000000000000646. PMID: 34901755
3. Carlos Rodriguez-Galindo and Carl E. Allen. Langerhans cell histiocytosis. *Blood*. 2020 Apr 16;135(16):1319-1331. DOI: 10.1182/blood.2019000934. PMID: 32106306
4. Laura Polivka, Julien Rossignol, Antoine Neuraz et al. Criteria for the regression of pediatric mastocytosis: a long-term follow-up. *The Journal of Allergy and Clinical Immunology: In Practice*. Volume 9, Issue 4, April 2021 Pages 1695-1704.e5. DOI: 10.1016/j.jaip.2020.12.019. PMID: 33338682
5. Lozano Masdemont B, Campos Dominguez M, Gomez-Recuero Munoz L, Moreno Garcia B, Parra Blanco V, Suarez Fernandez R. Congenital cutaneous Langerhans cell histiocytosis and cutaneous mastocytoma in a child. *Dermatol Online J*. 2016;22(4). DOI: 13030/qt4kv9x9rh. PMID: 27617456