Dermatology Practical & Conceptual

Cutaneous and Orbital Myeloid Sarcoma as the Initial Clinical Presentation of Acute Myeloid Leukemia in an Infant

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Introduction

Cutaneous myeloid sarcoma is a rare disease that predominantly occurs in association with myeloid malignancies, with a particularly high incidence in acute myeloid leukemia (AML). This paper presents a rare case of cutaneous and orbital myeloid sarcoma in an infant without a prior diagnosis of AML, contributing to the limited literature on the subject.

Case Presentation

A five-month-old male infant presented with a three-month history of asymptomatic cutaneous lesions. The lesions initially appeared on the abdomen and subsequently spread over the trunk and limbs, accompanied by a one-week period of swelling in the right lower eyelid. No fever, hemorrhage, or arthralgia was reported.

On examination, erythematous-violaceous macules and patches were observed on the torso and extremities, along with painless nodules. An erythematous mass was noted in the lower right eyelid region (Figures 1A-B). Laboratory tests revealed anemia (hemoglobin 7.9 g/dL) and elevated lactic dehydrogenase (606 U/L), with negative HIV and hepatitis B/C serologies.

A skin biopsy showed blue round cells infiltrating vessels, adnexals, and the interstitium, extending into the hypodermis, with hyperchromatic nuclei and frequent mitosis, suggesting hematopoietic lineage (Figures 2A-C). Immunohistochemical analysis was positive for CD45, CD43, CD4, and CD15, but negative for myeloperoxidase (MPO), CD34,

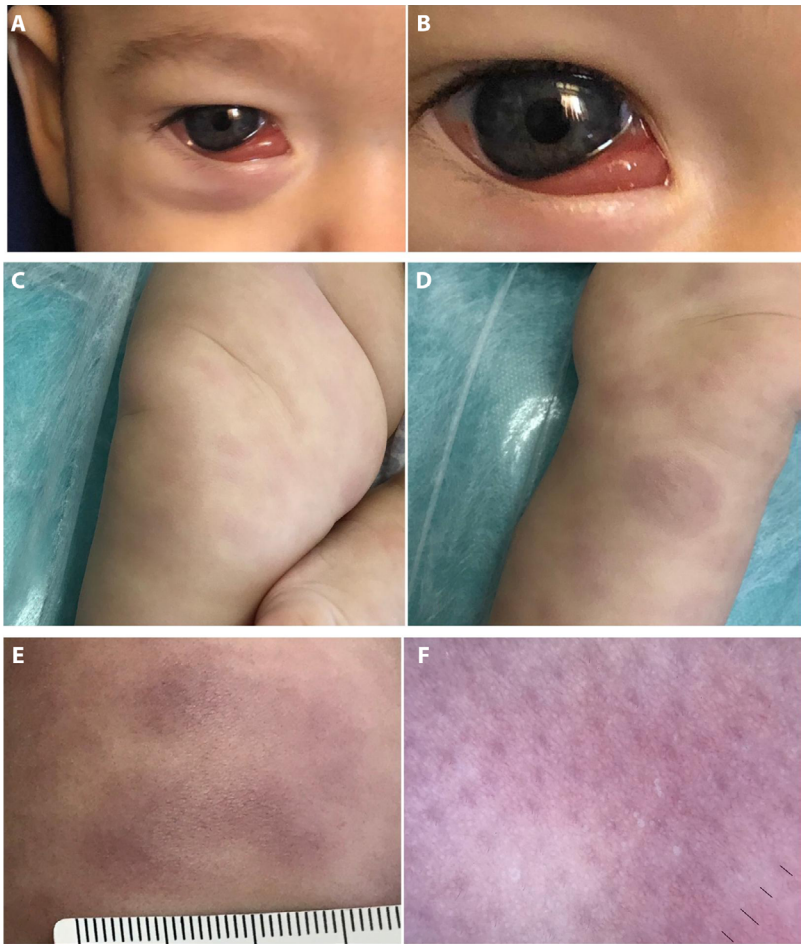

Figure 1. (A) Right eye displaying an erythematous mass in the lower eyelid region, extending to the medial portion of the sclera. (B) Close-up view of the affected area. (C-D) Clinical image showing erythematous-violaceous macular patches and the presence of painless nodules that were detected upon palpation of the lower limbs. (E) Close-up view of erythematous-violaceous infiltrated lesions. (F) A dermatoscopic evaluation of the disseminated erythematous-violaceous lesions discloses the presence of dotted vessels on a conspicuous violaceous background. However, no discernible abnormality in the skin texture is observed.

CD117, and TdT, indicating a myeloid lineage consistent with cutaneous myeloid sarcoma. Bone marrow biopsy confirmed acute myeloid leukemia.

The patient received chemotherapy (cytarabine and daunorubicin), achieving disease-free status. Two years later, the patient's leukemia returned. A bone marrow transplant was successful, resulting in long-term remission.

Discussion

Myeloid sarcoma may occur with hematological conditions like AML, CML, myelodysplastic syndrome, or without prior diagnoses [1]. In the pediatric age group, infiltration of the skin is the most common extramedullary manifestation of myeloid sarcoma, affecting 12.3% of pediatric AML patients, with higher prevalence in children under age one year [2]. The occurrence of orbital involvement in patients with AML is estimated to be between 3% and 9% and is most prevalent in young children [3].

Cutaneous myeloid sarcoma presents as papules, plaques, or nodules, typically on the trunk, head, neck, or extremities [4]. At histopathological examination, the leukemic infiltrate can be subtle and easily overlooked, and the rate of histopathological diagnostic error may be high, especially in patients lacking a history of AML [5]. Thus, clinical suspicion together with immunohistochemical analysis is important for the correct diagnosis. Dermatoscopy of cutaneous myeloid sarcoma shows nonspecific features such as polymorphic vessels (dotted, linear, or serpentine) on a pink-to-violaceous stroma (Figure 2F). These patterns overlap with conditions like leukemia cutis or inflammatory dermatoses, making dermatoscopy a useful but nonspecific tool [6]. While it helps guide biopsy, definitive diagnosis depends on histopathology and immunohistochemistry.

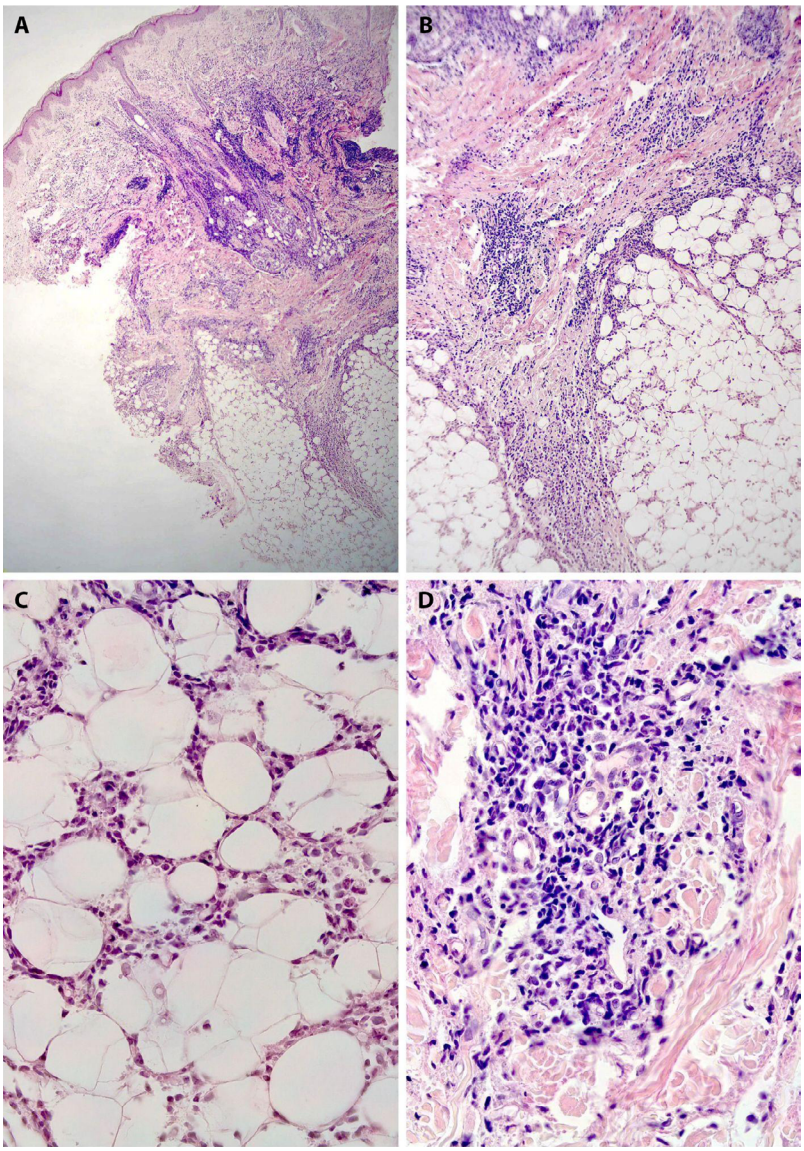

Figure 2. (A) Histopathology reveals dermal infiltration of isolated round cells surrounding vessels, adnexal structures, and the interstitium (H&E staining). (B) Histopathology shows infiltration of isolated round cells extending into the hypodermis. (C) Hypodermal infiltration by blue round cells. (D) Dermal infiltration by small-to-medium-sized cells with hyperchromatic nuclei and a high nucleus-to-cytoplasm ratio.

Conclusion

Cutaneous myeloid sarcoma may precede bone marrow involvement and be the first sign of AML or its recurrence. Prognosis is poor, particularly in pediatric cases [1]. Early detection is essential, as it can indicate imminent leukemia. Dermatologists play a key role in early diagnosis, which is critical to initiating treatment and improving outcomes.

References

1. Vasconcelos ERA, Bauk AR, Rochael MC. Cutaneous myeloid sarcoma associated with chronic myeloid leukemia. *An Bras Dermatol.* 2017;92(5 Suppl 1):50-52. DOI: 10.1590/abd1806-4841.20176042. PMID: 29267445.
2. Xu LH, Wang Y, Chen ZY, Fang JP. Myeloid sarcoma is associated with poor clinical outcome in pediatric patients with acute myeloid leukemia. *J Cancer Res Clin Oncol.* 2020;146(4):1011-1020. DOI:10.1007/s00432-020-03128-7. PMID: 31919567.
3. Lo WP, Kuo CH, Kuo MT, Fang PC. Isolated conjunctival myeloid sarcoma as a presenting sign of acute leukemia. *Chang Gung Med J.* 2010;33(4):334-337. PMID: 20584512.
4. Raja V, Bryant B, Bessman DJ, Alperin JB. Granulocytic sarcoma: presentation with nodal and skin involvement. *J Clin Oncol.* 2004;22(10):2026-2027. DOI:10.1200/JCO.2004.06.103. PMID: 15143096.
5. de Arruda Câmara VM, Morais JC, Portugal R, da Silva Carneiro SC, Ramos-e-Silva M. Cutaneous granulocytic sarcoma in myelodysplastic syndrome. *J Cutan Pathol.* 2008;35(9):876-879. DOI:10.1111/j.1600-0560.2007.00916.x. PMID: 18494822.
6. Sławińska M, et al. Dermoscopic features of leukemia cutis: case series. *Indian J Dermatol.* 2021;66(2):187-190. DOI:10.4103/ijd.IJD_534_19. PMID: 34188276.