CASE REPORT

Severe maculo-papular mastocytosis in a preterm twin newborn without systemic involvement, evolution at 10 years. Clinical case report

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Key words: Cutaneous mastocytosis, neonate, dizygotic twin.

How to cite this article: Guglielmetti Macchiavello M, Coke González VP, Guglielmetti Valenzuela A. Severe maculo-papular mastocytosis in a preterm twin newborn without systemic involvement, evolution to 10 years. Clinical case report. Rev Dermatol Cent Úraga. 2025; 7(1).

Reception date: 15/09/2024
Acceptance date: 12/12/2024

ABSTRACT

Cutaneous mastocytosis is a rare disease characterized by the clonal proliferation of mast cells, mainly in the skin. In preterm neonates, it can be confused with other skin pathologies, being a great diagnostic challenge. We present the case of a twin preterm newborn with generalized skin lesions from birth. Initially, the diagnosis was cutaneous histiocytosis based on histopathological findings. However, at 11 months, the appearance of Darier's sign and a second biopsy confirmed cutaneous mastocytosis. Management consisted of antihistamines and general measures. During 10 years of follow-up, the patient showed no systemic involvement. The initial differential diagnosis included cutaneous histiocytosis, highlighting the importance of repeating diagnostic studies when the clinical and pathological findings are not concordant. This case highlights that pediatric cutaneous mastocytosis can present from birth and requires a high diagnostic suspicion to avoid misdiagnosis and adequately manage the clinical course. The benign course observed in this case reinforces the self-limiting nature of pediatric cutaneous mastocytosis, although it emphasizes the need for prolonged follow-up to detect possible systemic progressions. This report contributes to the scarce literature on mastocytosis in preterm neonates.

INTRODUCTION

Mastocytosis encompasses a heterogeneous group of diseases characterized by clonal proliferation of mast cells in various organs, with the skin being the most common location, although cutaneous mastocytosis is rare.[1,2]

In children, the most common forms include maculopapular mastocytosis (urticaria pigmentosa), solitary mastocytoma, and diffuse cutaneous mastocytosis. Maculopapular mastocytosis is the most common pediatric variant, with a median onset of 3 months and occurring before 2 years of age in 94% of cases. The most common symptoms are Darier's sign in 86.7% of patients, pruritus in 43% and manifestations such as flushing and bullae. Systemic symptoms such as diarrhea and abdominal pain

are also described, with a direct relationship between the number of lesions and systemic involvement.[3]

Darier's sign, considered pathognomonic, is characterized by a wheal after the friction of the lesion, which can trigger acute systemic symptoms by release of mast cell mediators, especially in large lesions or with blistering, observed in infants and young children.[4]

Serum tryptase levels, a marker of systemic involvement, are suspect if they exceed 20 ug/L. In children under 2 years of age, 66% have levels <6.6 ug/L, while values >30.8 ug/L are associated with greater systemic symptoms.[2]

The diagnosis of cutaneous mastocytosis is based on clinical evaluation of lesions, positive Darier sign, and skin biopsy.[5]

Management focuses on managing symptoms and preventing triggers, such as temperature changes, certain foods, and medications. Antihistamines control pruritus, while in severe cases topical or systemic corticosteroids and targeted therapies are used depending on the extent of the disease.[6]

In newborns and young children, cutaneous mastocytosis is usually benign and resolves spontaneously, although it requires follow-up to monitor its evolution and adjust management.[3,5,6]

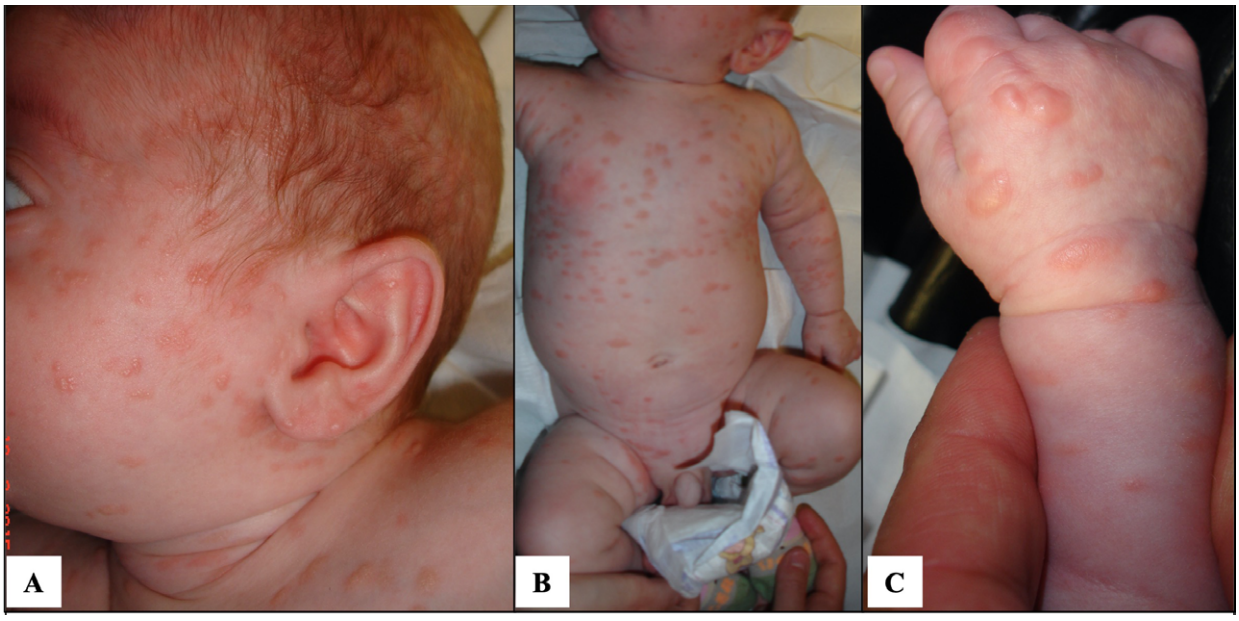
Reports of mastocytosis in preterm newborns are scarce, which led to the presentation of this clinical case as a diagnostic challenge and its evolution after 10 years of follow-up.

CLINICAL CASE

Preterm newborn (biamniotic bichororial twin pregnancy) is evaluated at 5 months of chronological age for generalized lesions present from birth. On physical examination, raised papular lesions, yellow-brown or reddish-brown, some eroded, converging to form plaques measuring from 5 mm to 2 cm, located on the face, trunk, upper and lower extremities, suggestive of a generalized infiltrative rash, without compromise of general condition (Figure 1).

A study was performed with a general laboratory with normal results and a biopsy was performed, whose histopathological report was suggestive of cutaneous

Figure 1. Initial presentation 5 months

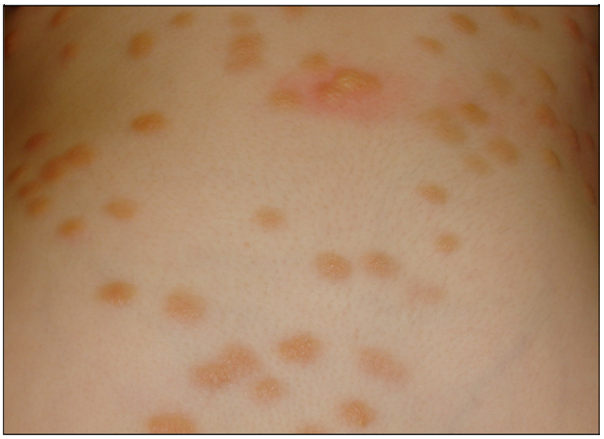


A. Facial involvement B. involvement of the trunk and genitals. C. Involvement of the extremities.

histiocytosis of an indeterminate type with an immunohistochemical study with a negative S-100 and CD1a reaction.

Serum tryptase levels are requested: 19.20 ug/L. Imaging studies were performed: abdominal ultrasound and x-rays of upper and lower extremities without pathological findings.

Figure 2. Darier sign



On clinical evaluation at 11 months, Darier's sign (+) appears in a lesion of the dorsum (Figure 2), which raises questions about the pathological diagnosis and raises the suspicion of mastocytosis.

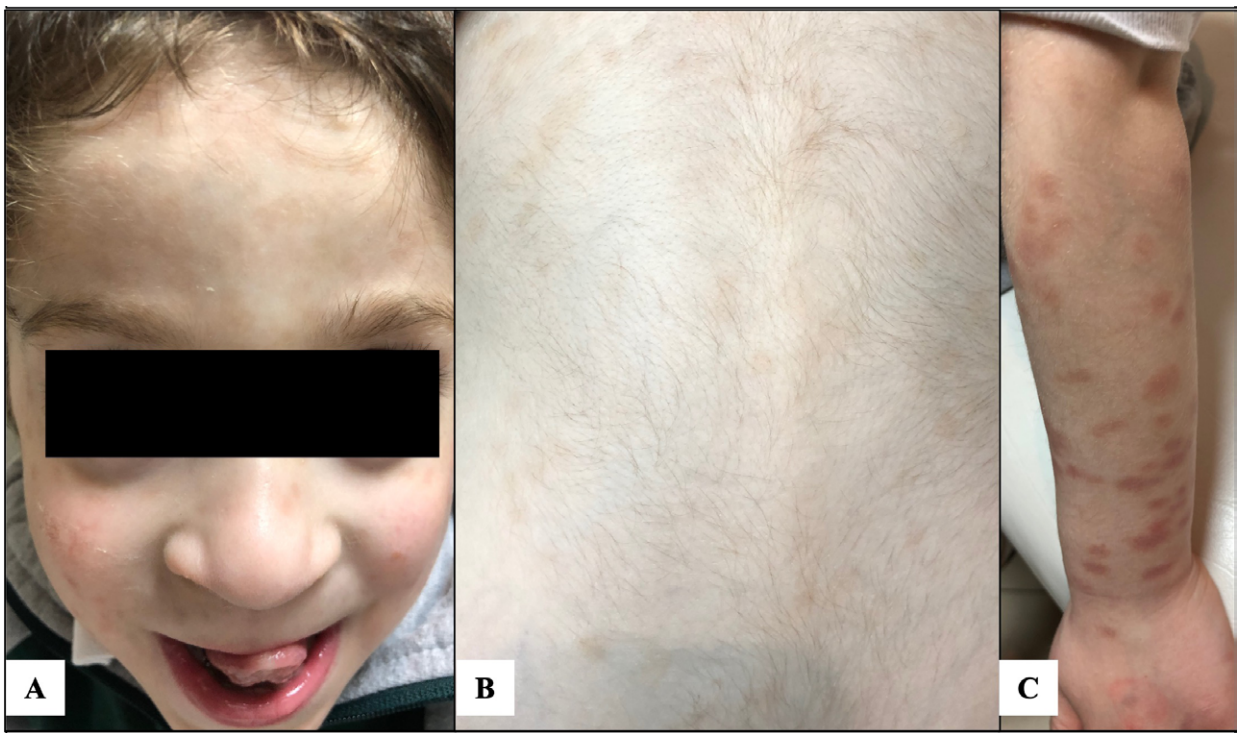
A new biopsy was performed with Giemsa stain and transmission electron microscopy that confirmed cutaneous mastocytosis.

The patient has evolved with a decrease in skin lesions at 5 years (Figure 3), followed up for 10 years in controls every 6 months, with general management based on diet and antihistamines, maintaining normal serum tryptase levels, with no evidence of systemic compromise.

DISCUSSION

Despite the characteristic appearance of the disease, the interesting thing about this case is that it appears in a twin preterm newborn that is initially clinically and histopathologically confused with histiocytosis. The benign course and Darier's sign were key in the clinical suspicion.

Figure 3. Clinical evolution, 5 years



A. Facial involvement B. involvement of the trunk and genitals. C. Involvement of the extremities.

Cutaneous mastocytosis, especially in its maculopapular form, is the most common presentation in pediatrics and usually appears in the first months of life. In this case, the lesions were present from birth, which is less common according to the literature, where the onset is usually at 3 months in most cases. The presence from birth raises the possibility of a congenital form, which reinforces the importance of including this pathology in the differential diagnosis of skin lesions in neonates, especially when the characteristics are persistent and generalized.

The lesions in this patient are described as raised papular, yellow-brown or reddish-brown, some with erosions and forming plaques. These characteristics coincide with the classic description of lesions in cutaneous mastocytosis. The subsequent onset of Darier's sign at 11 months confirmed the clinical diagnosis. This sign, considered pathognomonic, appeared relatively late, which underlines that the initial absence does not exclude the disease and that the clinical course and the appropriate pathological correlation are essential to establish the diagnosis.

The case illustrates the challenges in the early diagnosis of cutaneous mastocytosis, especially in newborns. Initially, the differential diagnosis included cutaneous histiocytosis, which is reasonable given the pattern of infiltration observed in the initial biopsy. Benign cephalic histiocytosis (BCH) is a rare non-Langerhans cell histiocytosis that develops during infancy or early childhood, which may confound or raise a possible differential diagnosis.[7,8,9]

Additionally, serum tryptase levels, normal in this patient, were useful to rule out early systemic involvement.

The patient's evolution over 10 years without evidence of systemic involvement and with conservative management is consistent with the benign course of cutaneous mastocytosis in pediatrics. Spontaneous resolution was not fully observed, as the lesions persisted during childhood, although they were kept under control with antihistamines and general measures.

CONCLUSION

This case emphasizes the importance of clinical follow-up and accurate diagnosis in neonates with persistent skin lesions. Cutaneous mastocytosis, although rare, can present significant diagnostic challenges, and confirmation requires clinical, histological, and laboratory correlation. The benign course observed in this patient reinforces the self-limiting nature of the disease in most pediatric cases, but underscores the need for comprehensive management and long-term monitoring.

INFORMED CONSENT

The patient included in this study and their representative have signed the informed consent, approving the use of their images and clinical data exclusively for research and scientific publication purposes. It is guaranteed that no personal data has been provided and no photographs have been used to allow identification.

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