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Case report

URTICARIA PIGMENTOSA: A CASE OF PEDIATRIC RARE CUTANEOUS MACULOPAPULAR MASTOCYTOSIS

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Abstract

Mastocytosis in children is a rare disorder characterized by an abnormal accumulation of mast cells in the skin and/or different organs Based on the site of mast cell accumulation, pediatric mastocytosis can be classified into two categories: cutaneous mastocytosis and systemic mastocytosis. The World Health Organization classification system differentiates three clinical variants of cutaneous mastocytosis: mastocytoma, urticaria pigmentosa also known as maculopapular cutaneous mastocytosis, and diffuse cutaneous mastocytosis. We present a clinical case of urticaria pigmentosa in an infant.

In January 2025, a five-month-old infant was referred to the University Clinic for Dermatology, due to the presence of multiple brownish macules disseminated across the trunk and upper extremities, accompanied by a mild degree of pruritus. The infant was generally healthy, born from an uncomplicated pregnancy, with no family history of dermatological diseases. The lesions were not present at birth. The first signs of pigmentation were noticed by the parents approximately three months prior, with a gradual increase in both the number and size. The dermatological status revealed the presence of more than 20 pigmented macules, each approximately 1 cm in diameter, localized on the trunk and upper extremities. A diagnosis of urticaria pigmentosa was established based on the clinical appearance and presence of positive Darier's sign, whereas a skin biopsy was unnecessary. The treatment regimen for the infant included only oral antihistamines. Long-term follow-up of this patient is needed, as the child has not yet reached puberty, in order to assess the severity and prognosis of the disease.

Keywords: pediatric mastocytosis, cutaneous mastocytosis, urticaria pigmentosa, maculopapular cutaneous mastocytosis, mast cells, Darier's sign

Introduction

Mastocytosis in children is a rare disorder characterized by an abnormal accumulation of mast cells in the skin and/or different organs [1,2]. Based on the site of mast cell accumulation, pediatric mastocytosis can be classified into two categories: cutaneous mastocytosis and systemic mastocytosis (with bone marrow involvement, followed by the gastrointestinal tract, liver, spleen, lymph nodes and any other organs) [1,2]. Systemic mastocytosis is rarely diagnosed in children and more often arises in adults [1,2].

The World Health Organization classifies mastocytosis into three clinical variants: mastocytoma, urticaria pigmentosa, and diffuse cutaneous mastocytosis [1,2,3].

In children, urticaria pigmentosa has been considered a relatively benign, self-limiting disease with onset in infancy or early childhood [4,5]. Clinically, it presents with skin lesions and pruritus [4,5]. Skin changes are characterized by a generalized eruption of red-brown to

yellowish macules and papules that vary in size, from several millimeters to centimeters in diameter, predominantly involving the trunk and occasionally the extremities [4-6]. Acral areas are often spared [4-6]. The lesions have a tendency to urticate when stroked and this is known as positive Darier's sign [1,4,6]. Additional symptoms may include: flushing, headache, bone and abdominal pain, and anaphylaxis [2,4,6]. Pathogenesis is still unclear [6,7]. However, studies have shown a close association with mutations in the c-KIT gene, as well as an exposition to certain trigger factors, like: temperature changes, friction, stress, exercise, infections, hot and spicy food, alcohol and some drugs (nonsteroidal anti-inflammatory drugs, narcotics, anticholinergic agents, anesthetic and other) [5,7]. Degranulated mast cells release mediators like histamine, tryptase, prostaglandins, leukotrienes, interleukins and other, responsible for many of the acute signs and symptoms [4,5,8].

This paper focused on a clinical case of a pediatric patient with urticaria pigmentosa, also known as maculopapular cutaneous mastocytosis, with classic skin lesions as described earlier.

Case report

We present a clinical case of urticaria pigmentosa in an infant. In January 2025, a 5-month-old male infant was brought for an examination to the University Clinic for Dermatology due to the presence of diffuse multiple brownish macules on the trunk and upper extremities (Figures 1 and 2). Mild pruritus was the only accompanying symptom. The infant was generally healthy, born from an uncomplicated pregnancy, with no family history of dermatological diseases. At the time of birth, the infant lacked any skin lesions. As reported by the parents, the first signs of pigmentation began approximately three months prior, with a gradual increase in both the number and size of the lesion.

The dermatological status revealed the presence of more than 20 pigmented macules, approximately 1 cm in diameter, localized on the trunk and upper extremities. Acral regions lacked lesions. Positive Darier's sign was present in three regions (upper, lower trunk and left upper extremity) after 30 seconds of rubbing with a blunt object (Figures 1 and 2). Physical examination excluded presence of hepatosplenomegaly or lymphadenopathy. The laboratory test results were within reference values.

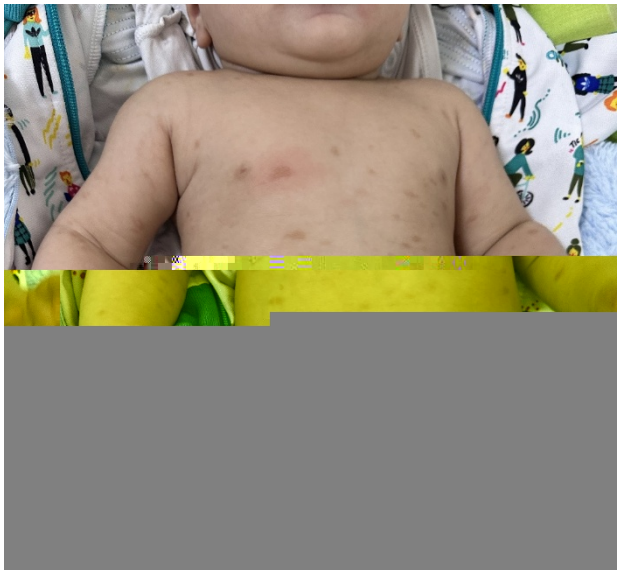

Fig. 1. Urticaria pigmentosa: multiple brownish papules with positive Darier's sign in two lesions of the trunk

Diagnosis of urticaria pigmentosa was established based on the clinical appearance and presence of positive Darier's sign; skin biopsy was not required. The treatment regimen for the infant included only oral antihistamines - cetirizine. The parents were consulted about the course and nature of the disease and advised for regular ambulatory follow-up. Long-term follow-up of this patient is needed, as the child has not yet reached puberty, in order to assess the severity and prognosis of the disease.

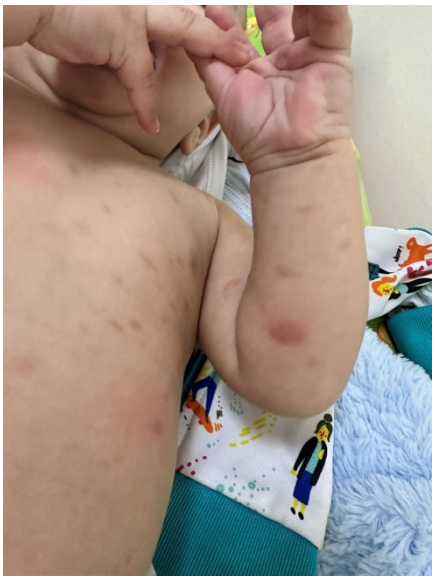

Fig. 2. Urticaria pigmentosa: positive Darier's sign on the left upper extremity

Discussion

Diagnosis of cutaneous mastocytosis may be challenging [2,5]. The criteria for diagnosing cutaneous mastocytosis must include one major and one minor criterion [5]. Major criteria include presence of the typical skin lesions [5]. Minor criteria include presence of mast cells accumulation in the dermis with aggregates of >15 mast cells per cluster or scattered mast cells with >20 per high microscopic power field in skin biopsy and/or detection of a c-KIT mutation in the affected skin as a molecular criterion [5]. It is well known that over 95% of patients harbor the driver KIT D816V mutation, the most common c-KIT mutation associated with systemic mastocytosis [9].

Beside the major and minor criteria, it is necessary to perform a complete blood count, along with serum tryptase [1,2]. Serum tryptase levels greater than 20 ng/mL are considered high and correlate with the increased mast cell proliferation and arouse suspicion of systemic involvement [2,3]. In this case bone marrow biopsy should always be performed [2,3].

The majority of skin lesions and the severity of their symptoms will resolve over time; therefore, it is important to educate and reassure families [8]. The first line of therapy is to avoid the potential trigger factors that cause mast cell degranulation [4,8]. Treatment usually includes oral H1 and/or H2 antihistamines, topical steroid creams and/or topical calcineurin inhibitors [4,5,8].

Conclusion

In summary, urticaria pigmentosa in children is benign, self-limiting disease with generally good prognosis and complete spontaneous regression before adolescence. However, if persists beyond adolescence, there is a risk of systemic involvement with a poor prognosis. Patients should be carefully investigated for underlying systemic involvement and malignancies. Numerous knowledge gaps exist, which make obstacle in understanding the

exact pathophysiologic mechanism. Further investigations are needed in this field in order to define standardized diagnostic criteria and management guidelines. A long-term follow-up is necessary for all pediatric patients with mastocytosis.

Conflict of interest statement. None declared.

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