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Cutaneous Mastocytosis: Case Series and Clinical Images

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1. Introduction

Mastocytosis is a disorder that affects children and adults, characterized by an excess of mast cells in the tissues. These excessive cells are most commonly found in the skin and, less frequently, in the lining of the stomach, intestines, bones, and connective tissue. The condition is triggered by a genetic alteration in the KIT protein present in mast cells, leading to the exaggerated accumulation of these cells, which are rich in inflammation mediators such as histamine, leukotriene C4, prostaglandins, tryptase, TNFα, and IL-8, in various parts of the body. The prevalence of mastocytosis is estimated at one occurrence per 10,000 people, while the incidence is approximately one per 100,000 people/year.

There are several types of cutaneous mastocytosis, while those involving skin plus internal organs are called systemic. Symptoms related to mastocytic degranulation are frequently reported in all categories of mastocytosis. These may be: dyspepsia, diarrhea, abdominal pain, musculoskeletal pain and hypotension. On the other hand, mastocytosis in adults progresses in a less favorable way.

2. Case 1

A healthy 1-year-old boy came to consult with a generalized hyperpigmented macules, since birth. On examination, we saw asymptomatic disseminated dark-brown macules (Figure 1). He had a normal nutritional status, vital signs were also O.K. and no enlarged lymph nodes were detected. Darier sign (upon rubbing his skin with a pen, an elevated papule resulted) was positive. He had no associated conditions. He was being treated with topical corticosteroids without any improvement of the lesions. The whole picture spoke in favor of Urticaria Pigmentosa. Lab tests were requested and all of which were within the normal range.

3. Case 2

A 39-year-old otherwise healthy male was evaluated for a 5-year-long history of rash on the on the back a(Fig. 2And that they were increasing and with a lot of itching. On the initial clinical examination, he looked well, he had normal vital signs and his systemic examinations were within the normal range. Strolling the lesion with a wooden end of a pencil induced a wheal confined to the stroked site (positive Darier sign). (Fig. 2C). The patient complained of systemic symptoms like flushing, exacerbated by physical activities (football) and stress. Due to the extreme facial flushing hat appeared during the games, he ended up being bullied and gave up playing. Routine laboratory findings including blood and urine, hepatorenal function and immunity items were normal ruling out systemic involvement. Subsequently, a 4 mm punch biopsy was done, which revealed increased numbers of mast cell aggregates and filling the papillary dermis (Figure 2C).

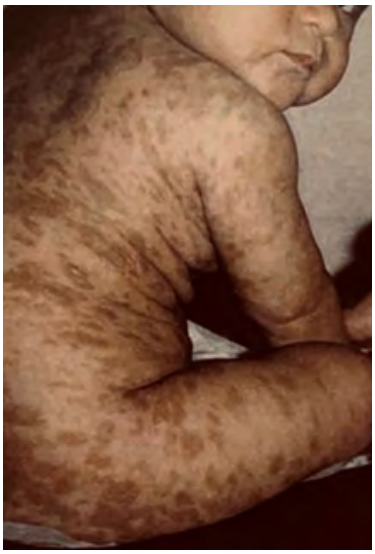

Figure 1: Disseminated dark-brown macules of cutaneous mastocytosis characteristics of urticaria pigmentosa in an infant. These macules were present in small numbers at birth and increased over the months. The tendency of this variant is the disappearance around puberty.

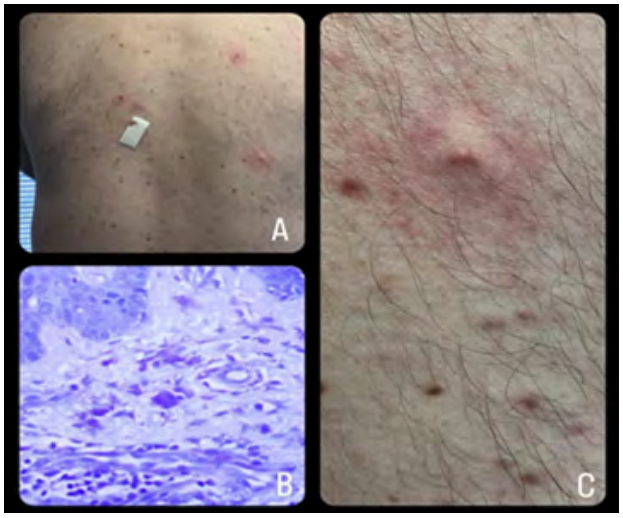

Figure 2: A) Darier sign is characteristic and diagnostic of mastocytosis: upon rubbing an affected area of skin with a pen, an elevated red papule results within a few minutes becoming pruritic. B) Skin biopsy showing increased numbers of mast cell aggregates and filling the papillary dermis.

4. Case 3

A 34-year-old healthy male was referred for evaluation for multiple reddish, flat and elevated spots surrounded by wheels on his back. His systemic complaints were abdominal pain and diarrhea. Intense dermographism were noted (Figure 3). The patient was also referred for evaluation to hematology service and no systemic changes were detected.

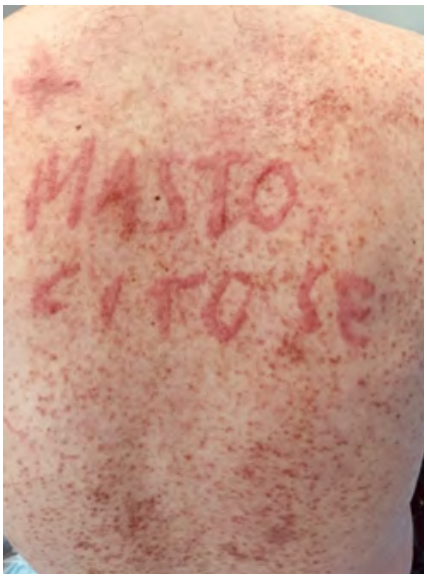

Figure 3: Patient3. Intense dermographism and serum tryptase level >20 ug/- L.

5. Discussion

Maculopapular cutaneous mastocytosis (MPCM), also known as Urticaria Pigmentosa (UP) is the most common pattern of cutaneous mastocytosis and manifest as brown patches on the skin and symptoms of itching, swelling and persistent urticaria-like lesions. In children, the spots can be small or large, while in adults they are more commonly so small that they are sometimes misdiagnosed as freckles and the tendency is the disappearance around puberty. The increased number of mast cells appears as pink or dark marks that may itch.

Darier sign is pathognomonic of the disease. Strolling the lesion with a wooden end of a pencil (or other small object), a wheel confined to the stroked site appears in some minutes representing mast cell degranulation induced by physical stimulation.

There are different patterns of this rare disease and all types can cause anaphylactic reactions. These patients have a good prognosis, with reduction or regression of the disease until before puberty. Rarely, clonal mast cells may also be present in the bone marrow, spleen, lymph nodes and gastrointestinal tract leading to symptoms of abdominal pain with diarrhea, dyspnea, hypotension, black-outs, and bone pain. Flushing is common. Serum tryptase level persistently >20 ug/L indicates systemic involvement and blood and marrow smears and abdominal ultrasound are mandatory. All our patients had normal values. Mast cell leukemia is an exceedingly rare aggressive hematological malignancy where immature mast cells in bone marrow aspirate greater than 20% and high levels of tryptase (>200 ng/ml). Mastocytosis may also be rarely associated with myeloproliferative disorders and leukemias.

As mastocytosis involves an excess of histamine-producing cells, hives-like reactions can occur in certain circumstances. It is important to note that each patient has specific sensitivities, with no fixed patterns of reactions, and triggering factors can vary over time.

Among the relevant situations to avoid triggers that lead to histamine release are imaging exams requiring radiocontrast, the use of morphine, codeine, and their derivatives, exposure to sudden changes in temperature, contact with inhalants such as perfumes, smoke, chemicals, and natural aromas, emotional factors, respiratory and gastrointestinal viral infections, use of nonsteroidal anti-inflammatory drugs (NSAIDs), administration of anesthetics, beta-lactams, vaccines, and antibiotics, consumption of alcoholic beverages and certain foods (with individual variations), and exposure to insects. A leaflet must be given to patients.

To ensure a prompt response in case of emergency, it is advisable for susceptible patients to always have antihistamines or EPIPEN self-application devices available. These measures aim to reduce the risks of severe reactions and provide greater safety and quality of life for individuals with mastocytosis.