Chilblains as a Clue for the Diagnosis of Essential Thrombocythemia

Perniose como Pista para o Diagnóstico de Trombocitose Essencial

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Dear Editor,

A 75-year-old man, Fitzpatrick's phototype II, with a past medical history relevant for atrial fibrillation, was referred to the Dermatology department due to actinic keratosis on his face. However, on dermatological examination, severe chilblain-like lesions on his fingers were also evident (Fig. 1). After detailed anamnesis, the patient mentioned these painful violaceous and ulcerated patches had lasted for two years, leading to nail dystrophy. In addition, these skin lesions worsened with cold weather. For this reason, the patient was previously examined at a private reumatology clinic with suspected perniosis and was treated with nifedipine, with no clinical improvement. The patient denied any other systemic signs or symptoms.

At this point, laboratory examination revealed a thrombocytosis of 772 000/mm^3, antinuclear antibodies (ANA) test of 1/160, negative extractable nuclear antigen antibodies (ENA) panel, negative cryoglobulins, negative rheumatoid factor, negative antiphospholipid antibodies and the serum protein electrophoresis did not yield any significant changes. Subsequently, the patient was referred to the Hematology department, where the diagnosis of essential thrombocythemia (ET) was established. The patient started treatment with hydroxyurea 500 mg every other day with a drop in platelets to 584 000/mm^3 and a slight improvement in skin lesions and pain.

Essential thrombocythemia is a myeloproliferative neoplasm defined by an increase in blood platelets and a predisposition to vascular events (both thrombotic and, less frequently, hemorrhagic).[1] Cutaneous lesions may be the first manifestation of ET[1,2] and generally include microcirculatory abnormalities such as erythromelalgia, (characterized by recurrent episodes of burning pain and erythema in the distal extremities, which is exacerbated by heat and exercise), acrocyanosis, hematoma, ecchymosis, petechiae, purpura, superficial thrombophlebitis, Raynaud's phenomenon, livedo reticularis, ulceration, and ischemic gangrene.[1] Recognizing dermatologic manifestations of ET may be a clue to its early diagnosis.

Chilblains are a prevalent dermatological condition that typically manifests during the winter months because of an abnormal immune response from exposure to cold temperatures. These lesions typically manifest as painful, edematous, erythematous or violaceous papules and plaques with associated pruritus, burning or pain. In addition, erosions, ulcerations, or blisters may be superimposed. The diagnosis can be established clinically.[3] Primary chilblains are uncommon in the elderly population and a systemic underlying cause should be investigated, particularly in severe cases,

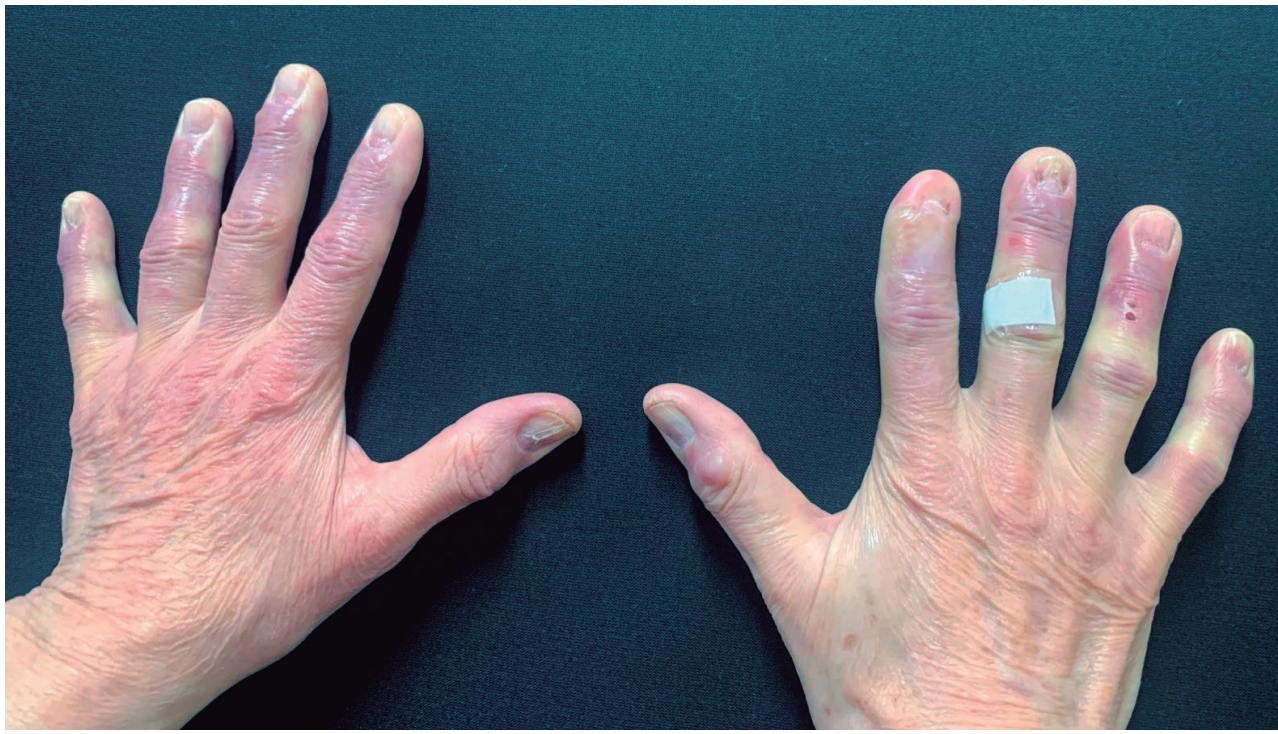

Figure 1 – Erythematous-to-violaceous plaques with ill-defined borders on the distal dorsal aspect of the fingers, accompanied by erosions and nail dystrophy, including dorsal pterygium, anonychia (absence of the nail plate), and onychorrhexis (longitudinal ridging and fragility of the nail plate).

or in those occurring outside of the typical seasonal period, or which are refractory to conventional therapy.[4]

Our clinical case highlights the importance of physical examination in modern medicine, not only in the cutaneous manifestation of systemic diseases but also in the identification of signs and symptoms that were not the initial reason for the appointment.

AUTHOR CONTRIBUTIONS

HL: Data collection, literature search, writing of the manuscript.

JR: Critical review of the manuscript.

AR: Data collection, critical review of the manuscript.

All authors approved the final version to be published.

PROTECTION OF HUMANS AND ANIMALS

The authors declare that the procedures were followed according to the regulations established by the Clinical Research and Ethics Committee and to the Helsinki Declaration of the World Medical Association updated in October 2024.

DATA CONFIDENTIALITY

The authors declare having followed the protocols in use at their working center regarding patients' data publication.

PATIENT CONSENT

Obtained.

COMPETING INTERESTS

The authors have declared that no competing interests exist.

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Hugo J. LEME✉[1], José RAMOS[1], Ângela RODA[1]
1. Department of Dermatology and Venereology. Unidade Local de Saúde de Almada-Seixal. Almada. Portugal.
✉ **Autor correspondente:** Hugo J. Leme. hugojleme@gmail.com
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