Anaemia, thrombocytopenia and skin lesions

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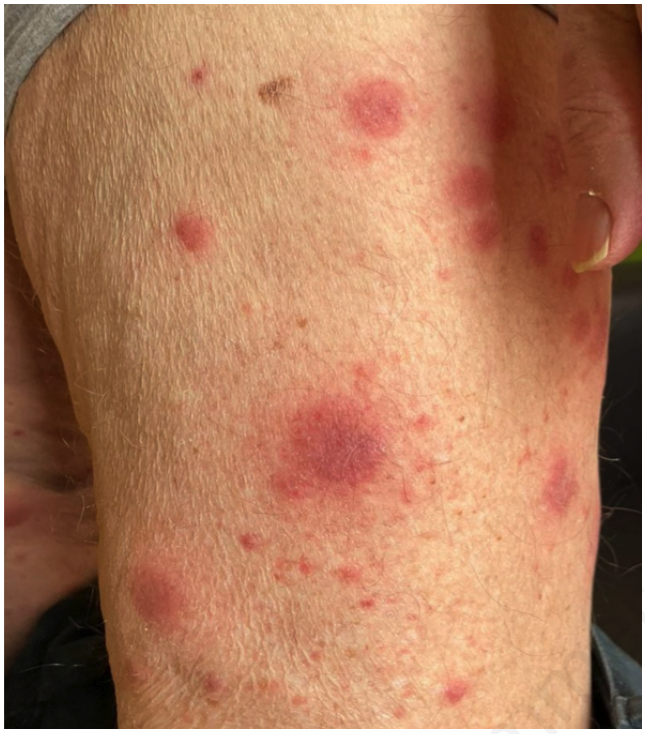

Descriptive legend. A 73-year-old man affected by hyperuricemia, dyslipidaemia and hypothyroidism presented to the emergency room with a 3-month history of fever, exertional dyspnea, progressive asthenia, and painless not itchy skin lesions. Physical exam showed purplish papules and plaques affecting any area of his body, and a slight bilateral oedema of his legs. Laboratory studies revealed a severe macrocytic anaemia (haemoglobin 4.8 g/dL, mean cell volume 119 fL) and thrombocytopenia (34,000/mm³) with hyperferritinemia (1894 ng/mL, normal value <400) and increased serum B12 (1412 pg/mL, normal value 197-771), associated with ESR 71 mm/h (normal value 1-15), CRP 139 mg/L (normal value <6), and procalcitonin 1.05 ng/mL (normal value <0.5).

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Conflicts of interest: The authors declare no conflict of interest. EP is member of the editorial board of Emergency Care Journal.

Availability of data and materials: All data underlying the findings are fully available upon reasonable request to Erika Poggiali, E.Poggiali@ausl.pc.it

Ethics approval and consent to participate: As this was a descriptive case report and data was collected without patient identifiers, ethics approval was not required under our hospital's Institutional Review Board guidelines.

Informed consent: The patient provided consent for the access to medical records at the time of admission.

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Question

Given the patient's history, what is the most likely diagnosis?

1. Erythema nodosum
2. Paraneoplastic pemphigus
3. Fungal skin infection
4. Urticarial vasculitis

Answer

Paraneoplastic Pemphigus (PNP) is the most likely. The patient was investigated with a colonoscopy and an upper endoscopy, that excluded a gastrointestinal bleeding and neoplasms. PET did not detect any occult malignancies. A bone marrow biopsy was performed, and a diagnosis of Refractory Anaemia with Excess Blasts type 2 (RAEB-2) was done. A punch biopsy confirmed the diagnosis of PNP. PNP showed a good response to the targeted therapy for RAEB-2, but not a complete resolution.

PNP is a rare life-threatening mucocutaneous autoimmune disease associated with malignancies,[1] the most frequent of which include lymphoma, leukaemia, and Castleman's disease.[2,3] Since some cases of PNP were diagnosed before an underlying malignancy was detected, PNP can be considered as a marker for occult malignancy. PNP manifests as polymorphic mucocutaneous eruptions, ranging from blisters and erosions to lichenoid eruptions, as reported in our case, to onychodystrophy and alopecia.[4] A single patient can present different types of lesions. Some patients suffer from dyspnea due to bronchiolitis obliterans as extracutaneous manifestation of PNP, more common in Castleman's disease.[5] The pathogenetic mechanism is not completely known. PNP is characterized by the production of autoantibodies against the plakin family proteins, which are target antigens of ordinary pemphigus. The course of PNP is not correlated with that of the associated malignancy, and the prognosis is generally poor, with high mortality rate due to sepsis or multi-organ failure. There are no consistently effective treatments. Rituximab, intravenous immunoglobulins, and plasmapheresis have shown promising effects.[6] Bronchiolitis obliterans is resistant to therapy, and lung transplantation is the last therapeutic option for respiratory failure.[7]

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