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Decoding Cytoplasmic Vacuolization: Unravelling the Diagnostic Challenges of Vexas Syndrome

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A 67-year-old male with a history of Ankylosing Spondylitis, diagnosed in 2019 and treated with methylprednisolone and methotrexate, presented in 2023 with testicular pain, orchitis-epididymitis, and Fournier gangrene. Despite surgical intervention and antibiotic therapy, he developed multiple skin abscesses. Laboratory results showed CRP 80.6 mg/L, Hb 10.2 g/dL, MCV 100 fL, PLT 22 K/uL, and INR 1.15. Pathological examination of skin and wound biopsies revealed necrosis and abscess formation.
A repeat bone marrow biopsy demonstrated marked hyperplasia in the granulocytic and megakaryocytic series, dysplastic changes, and patchy plasma cell infiltration. Bone marrow aspiration revealed vacuoles in progenitor cells. Persistent cytopenias, fever, abscesses, and bone marrow abnormalities raised suspicion for VEXAS syndrome. Genetic testing confirmed a somatic c.122T>C (p.M41T) mutation in the UBA1 gene at 81%, establishing the diagnosis.
This case highlights the importance of considering VEXAS syndrome in patients presenting with systemic inflammation, cytopenias, refractory abscesses, and vacuolated progenitor cells in the bone marrow. The findings emphasize the pivotal role of genetic testing for UBA1 mutations in confirming the diagnosis and suggest that vacuolization in bone marrow precursors should prompt suspicion for VEXAS syndrome.