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Steroid Cover-up: VEXAS Syndrome in a patient with Subacute Cutaneous Lupus (SCLE)

SUMMARY

A man in his 60s suffered from refractory, biopsy-proven subacute cutaneous lupus erythematosus that required chronic, moderate dose steroids to manage. His rash was accompanied by arthralgias and negative autoantibody testing. His SCLE was responsive to tofacitinib but thrombotic complications limited the use of this medication. He continued on prednisone 20 mg daily to manage his symptoms until the use of anifrolumab completely cleared his skin. During a subsequent prednisone taper, he developed a macrocytic anemia and elevated liver function tests (LFTs) that continued to progress. Ultimately a bone marrow biopsy and myeloid next generation sequencing revealed cellular vacuoles and UBA1 gene mutation respectively, consistent with a diagnosis of VEXAS syndrome. We believe the chronic steroid use to control his SCLE masked the underlying diagnosis for many years.

BACKGROUND

Subacute Cutaneous Lupus Erythematosus (SCLE) is an autoimmune disease that results in erythematous papules and plaques typically in sun-exposed areas of the skin. SCLE can occur in isolation, be medication induced, or be associated with Systemic lupus erythematosus (SLE), a heterogeneous autoimmune disease that can affect most organs in the body including the joints and bone marrow [1]. The risk factors for progression to SLE include female sex, ANA antibodies, disseminated lesions, and abnormal laboratory findings [2].

VEXAS (vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic) syndrome was first described in a subset of men in 2020 who had features of adult-onset inflammatory disease and myeloid abnormalities [3], [4]. The disease can present with wide ranging symptomatology and can cause skin rash, ocular inflammation, cardiovascular events, cytopenias, and arthritis. Diagnosis is made by appropriate clinical picture, vacuoles in myeloid populations on bone marrow aspirate, and mutations in the *UBA1* gene.

Here, we present a case of VEXAS in a patient with widespread and refractory SCLE. This case demonstrates the challenges of diagnosing VEXAS in a patient with a pre-existing autoimmune diagnosis and emphasizes the importance of re-evaluating a diagnosis when a patient is not improving on current standard of care therapies. We present the first reported case of a patient with underlying SCLE and new onset VEXAS.