Case of new-onset scleromyxoedema–scleroedema spectrum disorder in a patient with CLL

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Scleromyxoedema, a mimic of systemic sclerosis (SSc), is a rare progressive fibromucinous disease clinically characterised by symmetrical progressive skin thickening on the extremities and face.1 While scleromyxoedema has been described in patients with neoplasms, none has been described in patients with chronic lymphocytic leukaemia (CLL).2–5 Here, we present a patient whose new-onset scleromyxoedema likely coincided with his CLL relapse.

A 72-year-old man with a history of CLL in remission for 2 years was admitted for a 3-month history of skin thickening, concerning for SSc. Patient’s skin thickening started on his extremities and rapidly progressed to his face and trunk within 2 months, limiting his chest expansion and causing dyspnoea. He also had subjective fever, dysphagia and unintentional weight loss. He denies having Raynaud’s phenomenon, puffy hands, arthralgia, sicca, cough or bowel movement changes.

Patient’s CLL was diagnosed 6 years prior to the admission via flow cytometry (aberrant CD5, CD19, partially decreased CD20, decreased CD22, negative CD23, positive CD38, CD43, CD79b, CD200, kappa and negative lambda). Serum electrophoresis with immunofixation (SPEP with IFE) was not done at the time. When patient developed lymphadenopathy 2 years later, he was treated with ibrutinib and venetoclax, which drove his cancer into remission 2 years afterwards.

Patient’s physical exam showed diffuse skin thickening around his glabella, cheeks, neck, upper back, trunk, bilateral proximal arms and thighs, and dorsal hands. He had no skin thickening in his fingers or ears, no digital pits or ulcers, and no lung cracks. Nailfold capillaroscopy showed no abnormal capillary loops.

His complete metabolic profile, thyroid studies and urine samples were normal. However, his complete blood count (CBC) showed an elevated white blood count (WBC) of 19.6 × 10⁹/L with 83% lymphocytes in contrast to his normal CBC only 4 months prior. Autoimmune labs showed a positive 1:640 Anti-nuclear antibody (ANA) with speckled pattern and an anti-RNP autoantibody at 2 units (reference range: < 1 unit). Anti-dsDNA, anti-Smith, anti-SCL70, anti-RNA polymerase III, anti-centromere, rheumatoid factor and anticitrullinated peptide autoantibody were negative.

Patient’s endoscopy for his dysphagia was normal. Chest CT did not reveal interstitial lung disease (ILD). Transthoracic echocardiogram showed a normal right ventricular systolic pressure. His bone marrow biopsy demonstrated recurrent CLL/small lymphocytic lymphoma involving 70%–80% of the marrow cellularity.

Patient’s skin biopsy (figure 1) illustrated dermis with thickened collagen bundles and dermal mucin with minimal lymphocytic infiltration and fibroblast infiltration. Although the low number of dermal fibroblasts favoured a diagnosis of scleromyxoedema, scleromyxoedema was the eventual diagnosis given the numerous amount of mucin, IgM kappa monoclonal gammopathy on SPEP with IFE, and his lymphoproliferative cancer. Mixed connective tissue disease was ruled out as he lacked other rheumatological features such as arthralgia, rash, neuropathy, myositis, ILD, nephritis, pulmonary hypertension and Raynaud’s phenomenon.

Patient’s CLL was treated with obinutuzumab and acalabrutinib. Monthly intravenous immunoglobulin (IVIG) at 2 g/kg and UVA1 therapy were added to treat his scleromyxoedema in the hopes of quickly...
relieving his dyspnoea due to skin thickening. At 4-month follow-up, patient’s CLL had good clinical response with WBC at 5600/μL, and his scleromyxoedema also had dramatic improvement.

Our patient’s scleromyxoedema was likely related to his CLL relapse, which has never been described. Our case highlights the importance of considering SSc mimics when evaluating a patient for SSc. While both disease may be characterised by skin thickening, SSc more commonly presents with Raynaud’s phenomenon, abnormal nail capillaroscopy findings, arthritis, pulmonary hypertension and ILD. Finally, although his scleromyxoedema could have been resolved with cancer treatments alone, the IVIG therapy may have contributed to his quick recovery and, more importantly, did not interfere with his CLL treatments.

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