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Chained to a Diagnosis?

A 57-year-old Chinese woman presented to the emergency department for diarrhea.

She reported 15 months of gradually worsening diarrhea, with a colon biopsy from 6 months prior reported as ischemic colitis. Other symptoms included sicca, oral ulcers, alopecia, acral paresthesias, night sweats, and a 25-pound weight loss. An initial exam revealed severe orthostatic hypotension and hemorrhagic oral ulcers. Recent serologies were positive for an antinuclear antibody (1:80, speckled and cytoplasmic), and anti-Sjogren-syndrome-related antibodies A and B (anti-SSA and -SSB). The initial suspicion was connective tissue disease (CTD).

Further investigations revealed severe autonomic dysfunction. Nerve conduction studies ordered for the paresthesias were normal but incidentally showed myopathic changes in the quadriceps. A muscle MRI was therefore requested and showed fascial edema and mild myositis. Investigations for possible overlapping myocarditis revealed a positive troponin I, and a transthoracic echocardiogram showed abnormal global longitudinal strain, described as consistent with myocarditis. Surprisingly, a minor salivary gland biopsy was reported as normal and repeat anti-SSA and -SSB were negative.

Additionally, she was found to have elevated lambda free light chains (FLC). In this context, Congo red staining was retrospectively requested on the colon and salivary gland biopsies and demonstrated amyloid. Systemic involvement was subsequently confirmed by cardiac MRI, muscle biopsy, and bone marrow biopsy. She was diagnosed with AL amyloidosis and treated with daratumumab, cyclophosphamide, bortezomib, and dexamethasone. She reached clinical remission and remains on maintenance daratumumab.

This is a case of AL amyloidosis mimicking Sjogren's syndrome. Amyloidosis is a disorder of abnormal protein folding causing organ dysfunction. It is diagnosed histologically by the pathognomonic apple green birefringence with Congo red staining. Multiple organs may be affected, including the heart, peripheral and autonomous nervous systems, and gastrointestinal tract. Fatigue, weight loss, pain, purpura, and bleeding are common symptoms, whereas alopecia, sicca, and myopathy are rare. Nonetheless, approximately 3% of amyloidosis patients are initially labelled with autoimmune disease, despite negative autoantibodies (Prokaeva, 2007).

AL amyloidosis coexisting with Sjogren's syndrome has been reported (Delèveaux, 2001), but this was not the case here given the negative salivary gland biopsy. Rather, our patient's positive then negative serologies may have been affected by the high lambda FLC, a phenomenon called immunoassay interference (Tate, 2004), highlighting pitfalls and limitations of serologic testing.

In conclusion, AL amyloidosis may present similarly to a CTD and should be included in the differential diagnosis in the face of serological and histological discordance.